Consensus Statement: Chromosomal Microarray Is a Fi
Individuals with Developmental Disabilities or Congeni

American Journal of Human Genetics

86, 749-764

DOI: 10.1016/j.ajhg.2010.04.006

Citation Report

#	Article	IF	Citations
1	Molecular genetics and diagnostic techniques. , 2000, , 481-497.		0
2	Introduction to Human Genetics. , 2009, , 265-287.		2
3	Prenatal genetic screening and diagnosis for pediatricians. Current Opinion in Pediatrics, 2010, 22, 809-813.	1.0	3
4	Deletion 17q12 Is a Recurrent Copy Number Variant that Confers High Risk of Autism and Schizophrenia. American Journal of Human Genetics, 2010, 87, 618-630.	2.6	282
5	Clinically detectable copy number variations in a Canadian catchment population of schizophrenia. Journal of Psychiatric Research, 2010, 44, 1005-1009.	1.5	62
6	An introduction to standardized clinical nomenclature for dysmorphic features: the Elements of Morphology project. BMC Medicine, 2010, 8, 56.	2.3	1
7	Pathogenesis, neuroimaging and management in children with cerebral palsy born preterm. Developmental Disabilities Research Reviews, 2010, 16, 302-312.	2.9	17
8	Mosaic down syndrome in a patient with lowâ€level mosaicism detected by microarray. American Journal of Medical Genetics, Part A, 2010, 152A, 3154-3156.	0.7	16
9	Comparative analysis of copy number detection by whole-genome BAC and oligonucleotide array CGH. Molecular Cytogenetics, 2010, 3, 11.	0.4	56
10	MLPA for confirmation of array CGH results and determination of inheritance. Molecular Cytogenetics, 2010, 3, 19.	0.4	26
11	The use of array-CGH in a cohort of Greek children with developmental delay. Molecular Cytogenetics, 2010, 3, 22.	0.4	20
12	Fragile X and autism: Intertwined at the molecular level leading to targeted treatments. Molecular Autism, 2010, 1, 12.	2.6	204
13	Diagnostic utility of arrayâ€based comparative genomic hybridization (aCGH) in a prenatal setting. Prenatal Diagnosis, 2010, 30, 1131-1137.	1.1	70
14	Targeted array comparative genomic hybridisation (array CGH) identifies genomic imbalances associated with isolated congenital diaphragmatic hernia (CDH). Prenatal Diagnosis, 2010, 30, 1198-1206.	1.1	49
15	Public data archives for genomic structural variation. Nature Genetics, 2010, 42, 813-814.	9.4	71
16	Gene-environment interaction influences the reactivity of autoantibodies to citrullinated antigens in rheumatoid arthritis. Nature Genetics, 2010, 42, 814-816.	9.4	65
17	Lack of microarray uptake compromises diagnosis of young. Nature Medicine, 2010, 16, 725-725.	15.2	0
19	Genomic Copy Number Variation in Disorders of Cognitive Development. Journal of the American Academy of Child and Adolescent Psychiatry, 2010, 49, 1091-1104.	0.3	15

#	Article	IF	Citations
21	Classification of pathogenic or benign status of CNVs detected by microarray analysis. Expert Review of Molecular Diagnostics, 2010, 10, 717-721.	1.5	12
22	CHROMOSOMAL MICROARRAYS: THE BENEFITS AND CHALLENGES OF INTRODUCTION INTO PRENATAL DIAGNOSIS. Fetal and Maternal Medicine Review, 2010, 21, 307-322.	0.3	0
23	Approaches to the Patient with Neurogenetic Disease. Clinics in Laboratory Medicine, 2010, 30, 785-793.	0.7	2
24	Molecular diagnosis of acute myeloid leukemia. Expert Review of Molecular Diagnostics, 2010, 10, 993-1012.	1.5	15
25	Polygenic Heterogeneity: A Complex Model of Genetic Inheritance in Psychiatric Disorders. Biological Psychiatry, 2010, 68, 312-313.	0.7	8
26	Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. Genetics in Medicine, 2010, 12, 742-745.	1.1	517
27	Realising the benefits of genetics for health. Lancet, The, 2010, 376, 1370-1371.	6.3	3
28	Genomic Copy Number Variation in Disorders of Cognitive Development. Journal of the American Academy of Child and Adolescent Psychiatry, 2010, 49, 1091-1104.	0.3	106
29	Prenatal diagnosis of fetal aneuploidies: post-genomic developments. Genome Medicine, 2010, 2, 50.	3.6	8
30	Ethical issues raised by common copy number variants and single nucleotide polymorphisms of certain and uncertain significance in general medical practice. Genome Medicine, 2010, 2, 42.	3.6	20
31	Genetically complex epilepsies, copy number variants and syndrome constellations. Genome Medicine, 2010, 2, 71.	3.6	32
32	Genetic testing for autism: recent advances and clinical implications. Expert Review of Molecular Diagnostics, 2010, 10, 837-840.	1.5	17
33	Living Without a Diagnosis: The Parental Experience. Genetic Testing and Molecular Biomarkers, 2010, 14, 807-815.	0.3	55
34	Phenotypic variability and genetic susceptibility to genomic disorders. Human Molecular Genetics, 2010, 19, R176-R187.	1.4	234
35	Association between deletion size and important phenotypes expands the genomic region of interest in Phelan-McDermid syndrome (22q13 deletion syndrome). Journal of Medical Genetics, 2011, 48, 761-766.	1.5	112
36	Diagnostic Implications of Excessive Homozygosity Detected by SNP-Based Microarrays: Consanguinity, Uniparental Disomy, and Recessive Single-Gene Mutations. Clinics in Laboratory Medicine, 2011, 31, 595-613.	0.7	139
37	Interpretation of Array Comparative Genome Hybridization Data: A Major Challenge. Cytogenetic and Genome Research, 2011, 135, 222-227.	0.6	31
38	Rapid High-Resolution Mapping of Balanced Chromosomal Rearrangements on Tiling CGH Arrays. Journal of Molecular Diagnostics, 2011, 13, 621-633.	1.2	21

#	Article	IF	Citations
42	Clinical application of array-based comparative genomic hybridization by two-stage screening for 536 patients with mental retardation and multiple congenital anomalies. Journal of Human Genetics, 2011, 56, 110-124.	1.1	22
43	Autism spectrum disordersâ€"A genetics review. Genetics in Medicine, 2011, 13, 278-294.	1.1	466
44	Copy Number and SNP Arrays in Clinical Diagnostics. Annual Review of Genomics and Human Genetics, 2011, 12, 25-51.	2.5	143
45	Development of new postnatal diagnostic methods for chromosome disorders. Seminars in Fetal and Neonatal Medicine, 2011, 16, 114-118.	1.1	4
46	Array technology in prenatal diagnosis. Seminars in Fetal and Neonatal Medicine, 2011, 16, 94-98.	1.1	20
47	Fetal Surgery for Congenital Diaphragmatic Hernia Is Back from Never Gone. Fetal Diagnosis and Therapy, 2011, 29, 6-17.	0.6	86
48	Clinical Laboratory Implementation of Cytogenomic Microarrays. Cytogenetic and Genome Research, 2011, 135, 203-211.	0.6	8
49	Early-onset seizures due to mosaic exonic deletions of CDKL5 in a male and two females. Genetics in Medicine, 2011, 13, 447-452.	1.1	45
50	A copy number variation morbidity map of developmental delay. Nature Genetics, 2011, 43, 838-846.	9.4	1,141
51	Copy number variation in the dosage-sensitive 16p11.2 interval accounts for only a small proportion of autism incidence: A systematic review and meta-analysis. Genetics in Medicine, 2011, 13, 377-384.	1.1	50
52	Laboratory Aspects of Prenatal Microarray Analysis. Clinics in Laboratory Medicine, 2011, 31, 615-630.	0.7	5
53	In search of biomarkers for autism: scientific, social and ethical challenges. Nature Reviews Neuroscience, 2011, 12, 603-612.	4.9	209
54	Patterns of cardiac and extracardiac anomalies in adults with tetralogy of Fallot. American Heart Journal, 2011, 161, 131-137.	1.2	30
56	Genomic alterations in myeloid neoplasms withÂnovel, apparently balanced translocations. Cancer Genetics, 2011, 204, 68-76.	0.2	5
57	Implementation of high resolution single nucleotide polymorphism array analysis asÂaÂclinical test for patients with hematologic malignancies. Cancer Genetics, 2011, 204, 26-38.	0.2	29
58	A family with Xq22.3q25 interstitial deletion and normal ovarian function. Fertility and Sterility, 2011, 96, e29-e34.	0.5	0
59	Clinical report: An interstitial deletion of 16p13.11 detected by array CGH in a patient with infantile spasms. European Journal of Medical Genetics, 2011, 54, 314-318.	0.7	15
60	Using a combination of MLPA kits to detect chromosomal imbalances in patients with multiple congenital anomalies and mental retardation is a valuable choice for developing countries. European Journal of Medical Genetics, 2011, 54, e425-e432.	0.7	44

#	Article	IF	Citations
61	Oligonucleotide microarrays in constitutional genetic diagnosis. Expert Review of Molecular Diagnostics, 2011, 11, 521-532.	1.5	15
62	Copy-number changes in prenatal diagnosis. Expert Review of Molecular Diagnostics, 2011, 11, 579-592.	1.5	23
63	Genome Arrays for the Detection of Copy Number Variations in Idiopathic Mental Retardation, Idiopathic Generalized Epilepsy and Neuropsychiatric Disorders: Lessons for Diagnostic Workflow and Research. Cytogenetic and Genome Research, 2011, 135, 174-202.	0.6	103
64	American College of Medical Genetics standards and guidelines for interpretation and reporting of postnatal constitutional copy number variants. Genetics in Medicine, 2011, 13, 680-685.	1.1	794
65	American College of Medical Genetics recommendations for the design and performance expectations for clinical genomic copy number microarrays intended for use in the postnatal setting for detection of constitutional abnormalities. Genetics in Medicine, 2011, 13, 676-679.	1.1	91
66	Genetic and Epigenetic Networks in Intellectual Disabilities. Annual Review of Genetics, 2011, 45, 81-104.	3.2	329
67	Developments in Molecular Genetic Diagnostics: An Update for the Pediatric Epilepsy Specialist. Pediatric Neurology, 2011, 44, 317-327.	1.0	22
68	Microdeletion of Chromosome 15q26.1 in a Child With Intractable Generalized Epilepsy. Pediatric Neurology, 2011, 45, 60-62.	1.0	16
69	Human Copy Number Variation and Complex Genetic Disease. Annual Review of Genetics, 2011, 45, 203-226.	3.2	344
70	An evidence-based approach to establish the functional and clinical significance of copy number variants in intellectual and developmental disabilities. Genetics in Medicine, 2011, 13, 777-784.	1.1	371
71	From Karyotyping to Array-CGH in Prenatal Diagnosis. Cytogenetic and Genome Research, 2011, 135, 241-250.	0.6	61
72	SNP Array Analysis in Constitutional and Cancer Genome Diagnostics â€“ Copy Number Variants, Genotyping and Quality Control. Cytogenetic and Genome Research, 2011, 135, 212-221.	0.6	34
73	Genetic Testing in Epilepsy: What Should you be Doing?. Epilepsy Currents, 2011, 11, 107-111.	0.4	33
74	Array Comparative Genomic Hybridization and Cytogenetic Analysis in Pediatric Acute Leukemias. Current Oncology, 2011, 18, 210-217.	0.9	11
75	Miller-Dieker Syndrome with der(17)t(12;17)(q24.33;p13.3)pat Presenting with a Potential Risk of Mis-identification as a de novo Submicroscopic Deletion of 17p13.3. Annals of Laboratory Medicine, 2011, 31, 49-53.	1.2	4
76	Innovations in the Early Diagnosis of Chromosomal Disorders Associated with Intellectual Disability. International Review of Research in Developmental Disabilities, 2011, , 211-228.	0.6	0
77	Just in Time: An Update on Continuum Neurogenetics. CONTINUUM Lifelong Learning in Neurology, 2011, 17, 239-248.	0.4	0
78	Craniofacial Dysmorphism and Developmental Disorders Among Children with Chromosomal Microdeletions and Duplications of Unknown Significance. Journal of Developmental and Behavioral Pediatrics, 2011, 32, 600-604.	0.6	9

#	Article	IF	Citations
79	Genetic Testing in Clinical Practice. CONTINUUM Lifelong Learning in Neurology, 2011, 17, 316-325.	0.4	0
80	Evolving applications of microarray analysis in prenatal diagnosis. Current Opinion in Obstetrics and Gynecology, 2011, 23, 103-108.	0.9	37
82	Genetic basis of autism: is there a way forward?. Current Opinion in Psychiatry, 2011, 24, 226-236.	3.1	74
83	Genomic Medicine and Neurology. CONTINUUM Lifelong Learning in Neurology, 2011, 17, 249-267.	0.4	4
84	16q24.1 microdeletion in a premature newborn: Usefulness of array-based comparative genomic hybridization in persistent pulmonary hypertension of the newborn. Pediatric Critical Care Medicine, 2011, 12, e427-e432.	0.2	21
85	Chromosome microarray analysis in a clinical environment: new perspective and new challenge. British Journal of Biomedical Science, 2011, 68, 100-108.	1.2	5
86	College of American Pathologists/American College of Medical Genetics proficiency testing for constitutional cytogenomic microarray analysis. Genetics in Medicine, 2011, 13, 765-769.	1.1	13
87	Sequencing revolution. Developmental Medicine and Child Neurology, 2011, 53, 673-674.	1.1	0
88	Microarray as a first genetic test in global developmental delay: a cost-effectiveness analysis. Developmental Medicine and Child Neurology, 2011, 53, 994-999.	1.1	38
89	What price a diagnosis?. Developmental Medicine and Child Neurology, 2011, 53, 971-971.	1.1	5
90	Epilepsy and the new cytogenetics. Epilepsia, 2011, 52, 423-432.	2.6	56
91	Genome structural variation discovery and genotyping. Nature Reviews Genetics, 2011, 12, 363-376.	7.7	1,240
92	Needles in stacks of needles: finding disease-causal variants in a wealth of genomic data. Nature Reviews Genetics, 2011, 12, 628-640.	7.7	531
93	Rapid testing versus karyotyping in Down's syndrome screening: cost-effectiveness and detection of clinically significant chromosome abnormalities. European Journal of Human Genetics, 2011, 19, 3-9.	1.4	44
94	The causality of de novo copy number variants is overestimated. European Journal of Human Genetics, 2011, 19, 1112-1113.	1.4	43
95	Charting a course for genomic medicine from base pairs to bedside. Nature, 2011, 470, 204-213.	13.7	823
96	Comprehensive assessment of array-based platforms and calling algorithms for detection of copy number variants. Nature Biotechnology, 2011, 29, 512-520.	9.4	384
97	Education of a Child Neurologist: Developmental Neuroscience Relevant to Child Neurology. Seminars in Pediatric Neurology, 2011, 18, 133-138.	1.0	5

#	Article	IF	Citations
98	Practical Guidelines for Managing Patients with 22q11.2 Deletion Syndrome. Journal of Pediatrics, 2011, 159, 332-339.e1.	0.9	481
99	Analysis of a purported SHANK3 mutation in a boy with autism: Clinical impact of rare variant research in neurodevelopmental disabilities. Brain Research, 2011, 1380, 98-105.	1.1	28
100	Prenatal testing for intellectual disability: Misperceptions and reality with lessons from down syndrome. Developmental Disabilities Research Reviews, 2011, 17, 27-31.	2.9	13
101	Bridging autism, science and society: moving toward an ethically informed approach to autism research. Autism Research, 2011, 4, 271-282.	2.1	143
102	A new case of rare proximal 3q13 interstitial deletion. Open Medicine (Poland), 2011, 6, 625-630.	0.6	2
103	Genetics of Schizophrenia: New Findings and Challenges. Annual Review of Genomics and Human Genetics, 2011, 12, 121-144.	2.5	160
104	Next-Generation Sequencing Strategies Enable Routine Detection of Balanced Chromosome Rearrangements for Clinical Diagnostics and Genetic Research. American Journal of Human Genetics, 2011, 88, 469-481.	2.6	154
105	Identification of submicroscopic chromosomal aberrations in fetuses with increased nuchal translucency and apparently normal karyotype. Ultrasound in Obstetrics and Gynecology, 2011, 38, 314-319.	0.9	102
106	Risk factors for autism: translating genomic discoveries into diagnostics. Human Genetics, 2011, 130, 123-148.	1.8	102
107	The Contribution of Chromosomal Abnormalities to Congenital Heart Defects: A Population-Based Study. Pediatric Cardiology, 2011, 32, 1147-1157.	0.6	161
108	Genetic testing: predictive value of genotyping for diagnosis and management of disease. EPMA Journal, 2011, 2, 173-179.	3.3	6
109	Copy Number Variants: A New Molecular Frontier in Clinical Psychiatry. Current Psychiatry Reports, 2011, 13, 129-137.	2.1	11
110	Following the genes: a framework for animal modeling of psychiatric disorders. BMC Biology, 2011, 9, 76.	1.7	27
111	Clinical implementation of whole-genome array CGH as a first-tier test in 5080 pre and postnatal cases. Molecular Cytogenetics, 2011, 4, 12.	0.4	67
112	Evaluation of chronic lymphocytic leukemia by BAC-based microarray analysis. Molecular Cytogenetics, 2011, 4, 4.	0.4	8
113	Prenatal BACsâ€onâ€Beads^{â„¢}: a new technology for rapid detection of aneuploidies and microdeletions in prenatal diagnosis. Prenatal Diagnosis, 2011, 31, 500-508.	1.1	47
114	The development of a rapid assay for prenatal testing of common aneuploidies and microdeletion syndromes. Prenatal Diagnosis, 2011, 31, 778-787.	1.1	41
115	Introducing array comparative genomic hybridization into routine prenatal diagnosis practice: a prospective study on over 1000 consecutive clinical cases. Prenatal Diagnosis, 2011, 31, 1270-1282.	1.1	95

#	Article	IF	Citations
116	Suspected trisomy 22: Modification, clarification, or confirmation of the diagnosis by aCGH. American Journal of Medical Genetics, Part A, 2011, 155, 434-438.	0.7	7
117	2q23 de novo microdeletion involving the MBD5 gene in a patient with developmental delay, postnatal microcephaly and distinct facial features. American Journal of Medical Genetics, Part A, 2011, 155, 424-429.	0.7	27
118	11p14.1 microdeletions associated with ADHD, autism, developmental delay, and obesity. American Journal of Medical Genetics, Part A, 2011, 155, 1272-1280.	0.7	84
119	Pharmaco-genetically guided treatment of recurrent rage outbursts in an adult male with 15q13.3 deletion syndrome. , 2011, 155, 805-810.		46
120	Commentary: Recognizing syndromes with overlapping features: How difficult is it? Considerations generated by the article on differential diagnosis of Smithâ€“Magenis syndrome by Vieira and colleagues. American Journal of Medical Genetics, Part A, 2011, 155, 986-987.	0.7	2
121	The atypical 16p11.2 deletion: A not so atypical microdeletion syndrome?. American Journal of Medical Genetics, Part A, 2011, 155, 1066-1072.	0.7	46
122	Submicroscopic deletion in 7q31 encompassing CADPS2 and TSPAN12 in a child with autism spectrum disorder and PHPV. American Journal of Medical Genetics, Part A, 2011, 155, 1568-1573.	0.7	24
123	Familial intellectual disability and autistic behavior caused by a small FMR2 gene deletion. American Journal of Medical Genetics, Part A, 2011, 155, 2003-2007.	0.7	25
124	Marfanoid hypermobility caused by an 862â€‰kb deletion of Xq22.3 in a patient with Sotos syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 2293-2297.	0.7	6
125	Clinically relevant single gene or intragenic deletions encompassing critical neurodevelopmental genes in patients with developmental delay, mental retardation, and/or autism spectrum disorders. American Journal of Medical Genetics, Part A, 2011, 155, 2386-2396.	0.7	159
126	A de novo 2.1â€Mb deletion of 13q12.11 in a child with developmental delay and minor dysmorphic features. American Journal of Medical Genetics, Part A, 2011, 155, 2538-2542.	0.7	9
127	No association between a common single nucleotide polymorphism, rs4141463, in the MACROD2 gene and autism spectrum disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 633-639.	1.1	30
128	Rare copy number variants are an important cause of epileptic encephalopathies. Annals of Neurology, 2011, 70, 974-985.	2.8	222
129	Links between genetics and pathophysiology in the autism spectrum disorders. EMBO Molecular Medicine, 2011, 3, 438-450.	3.3	39
130	High-Resolution genomic arrays identify CNVs that phenocopy the chromosome 22q11.2 deletion syndrome. Human Mutation, 2011, 32, 91-97.	1.1	17
131	Characterization of copy numberâ€stable regions in the human genome. Human Mutation, 2011, 32, 947-955.	1.1	19
132	Extending the scope of diagnostic chromosome analysis: Detection of single gene defects using high-resolution SNP microarrays. Human Mutation, 2011, 32, 1500-1506.	1.1	41
133	So what? Does the test lead to improved health outcomes?. Neurology, 2011, 77, 1586-1587.	1.5	17

#	Article	IF	Citations
134	Diagnosis and management of autism in childhood. BMJ: British Medical Journal, 2011, 343, d6238-d6238.	2.4	35
135	Assessment and diagnosis of autism spectrum disorders. BMJ: British Medical Journal, 2011, 343, d6628-d6628.	2.4	2
136	Parental Origin, DNA Structure, and the Schizophrenia Spectrum. American Journal of Psychiatry, 2011, 168, 350-353.	4.0	12
137	Whole exome and whole genome sequencing. Current Opinion in Pediatrics, 2011, 23, 594-600.	1.0	124
138	Recurrence Risk for Autism Spectrum Disorders: A Baby Siblings Research Consortium Study. Pediatrics, 2011, 128, e488-e495.	1.0	1,088
139	Validation of Multiplex Ligation-Dependent Probe Amplification for Confirmation of Array Comparative Genomic Hybridization. Diagnostic Molecular Pathology, 2011, 20, 166-174.	2.1	7
140	Chromosomal abnormalities resembling Joubert syndrome. Clinical Dysmorphology, 2011, 20, 136-142.	0.1	4
141	Comparison of Constitutional and Replication Stress-Induced Genome Structural Variation by SNP Array and Mate-Pair Sequencing. Genetics, 2011, 187, 675-683.	1.2	20
142	Statin use and outcome after intracerebral hemorrhage: Case-control study and meta-analysis. Neurology, 2011, 77, 2073-2074.	1.5	6
143	Minor physical anomalies in children with autism spectrum disorders. Autism, 2011, 15, 746-760.	2.4	20
144	Neurogenetics. Neurology: Clinical Practice, 2011, 1, 41-48.	0.8	3
145	Motor Chip: A Comparative Genomic Hybridization Microarray for Copy-Number Mutations in 245 Neuromuscular Disorders. Clinical Chemistry, 2011, 57, 1584-1596.	1.5	48
146	Mosaic trisomy 13: understanding origin using SNP array. Journal of Medical Genetics, 2011, 48, 323-326.	1.5	19
147	Adaptor protein complex-4 (AP-4) deficiency causes a novel autosomal recessive cerebral palsy syndrome with microcephaly and intellectual disability. Journal of Medical Genetics, 2011, 48, 141-144.	1.5	169
148	Chorea, ballism, and athetosis. , 2011, , 335-349.		2
149	Standardization and Diversification of Copy Number Microarray Testing for Clinical Diagnosticsâ€"Implications of the Cross-Platform/Algorithm Study on Clinical Diagnostic Chromosomal Microarray Analysis. Clinical Chemistry, 2011, 57, 1354-1356.	1.5	1
150	Commentary on â€˜Identification of a microdeletion at Xp22.13 in a Taiwanese family presenting with Nanceâ€"Horan syndromeâ€™. Journal of Human Genetics, 2011, 56, 4-4.	1.1	0
151	Going BAC or oligo microarray to the well: A commentary on Clinical application of array-based comparative genomic hybridization by two-stage screening for 536 patients with mental retardation and multiple congenital anomalies. Journal of Human Genetics, 2011, 56, 104-105.	1.1	1

#	Article	IF	Citations
152	Autism, authenticated. Nature Medicine, 2011, 17, 1336-1338.	15.2	2
153	The UCSC Genome Browser database: update 2011. Nucleic Acids Research, 2011, 39, D876-D882.	6.5	958
154	Genetic risk in autism: new associations and clinical testing. Expert Opinion on Medical Diagnostics, 2011, 5, 347-356.	1.6	6
155	Neurodevelopmental Disabilities. , 2011, , .		18
156	Molecular Technologies Open New Clinical Genetic Vistas. Science Translational Medicine, 2011, 3, 65ps2.	5.8	24
157	Balanced into array: genome-wide array analysis in 54 patients with an apparently balanced de novo chromosome rearrangement and a meta-analysis. European Journal of Human Genetics, 2011, 19, 1152-1160.	1.4	41
158	Downsizing genomic medicine: Approaching the ethical complexity of whole-genome sequencing by starting small. Genetics in Medicine, 2011, 13, 191-194.	1.1	62
159	Clinical validity of karyotyping for the diagnosis of chromosomal imbalance following array comparative genomic hybridisation. Journal of Medical Genetics, 2011, 48, 851-855.	1.5	4
160	Genome and exome sequencing in the clinic: unbiased genomic approaches with a high diagnostic yield. Pharmacogenomics, 2012, 13, 511-514.	0.6	19
161	A Discovery Resource of Rare Copy Number Variations in Individuals with Autism Spectrum Disorder. G3: Genes, Genomes, Genetics, 2012, 2, 1665-1685.	0.8	175
162	Prenatal Diagnosis of Cystic Hygroma related to a Deletion of 16q24.1 with Haploinsufficiency of FOXF1 and FOXC2 Genes. Case Reports in Genetics, 2012, 2012, 1-4.	0.1	17
163	A 600â€…kb deletion syndrome at 16p11.2 leads to energy imbalance and neuropsychiatric disorders. Journal of Medical Genetics, 2012, 49, 660-668.	1.5	251
164	Rare Copy Number Variations in Adults with Tetralogy of Fallot Implicate Novel Risk Gene Pathways. PLoS Genetics, 2012, 8, e1002843.	1.5	149
165	A Rare Chromosome 3 Imbalance and Its Clinical Implications. Case Reports in Pediatrics, 2012, 2012, 1-5.	0.2	3
166	From Genetics to Genomics of Epilepsy. Neurology Research International, 2012, 2012, 1-18.	0.5	13
167	Further clinical and molecular delineation of the 15q24 microdeletion syndrome. Journal of Medical Genetics, 2012, 49, 110-118.	1.5	40
168	Ethics and neuropsychiatric genetics: a review of major issues. International Journal of Neuropsychopharmacology, 2012, 15, 1547-1557.	1.0	34
169	Discovery of variants unmasked by hemizygous deletions. European Journal of Human Genetics, 2012, 20, 748-753.	1.4	19

#	Article	IF	Citations
170	Core Concepts: Chromosome Aneuploidies. NeoReviews, 2012, 13, e30-e39.	0.4	3
171	The Autism Treatment Network and Autism Intervention Research Network on Physical Health: Future Directions. Pediatrics, 2012, 130, S198-S201.	1.0	5
173	Genetic investigation for adults with intellectual disability. Current Opinion in Neurology, 2012, 25, 150-158.	1.8	17
174	Current World Literature. Current Opinion in Neurology, 2012, 25, 208-213.	1.8	0
175	14q32 deletion syndrome. Clinical Dysmorphology, 2012, 21, 42-44.	0.1	10
176	Genomic medicine: what are the challenges for the National Health Service?. Personalized Medicine, 2012, 9, 539-545.	0.8	2
177	Detection of Microdeletion 22q11.2 in a Fetus by Next-Generation Sequencing of Maternal Plasma. Clinical Chemistry, 2012, 58, 1148-1151.	1.5	115
178	Microarrays in Diagnostics and Biomarker Development. , 2012, , .		3
179	â€œWhat does it mean?â€• Uncertainties in understanding results of chromosomal microarray testing. Genetics in Medicine, 2012, 14, 250-258.	1.1	101
180	Exon-level array CGH in a large clinical cohort demonstrates increased sensitivity of diagnostic testing for Mendelian disorders. Genetics in Medicine, 2012, 14, 594-603.	1.1	48
181	Variability in laboratory reporting practices for regions of homozygosity indicating parental relatedness as identified by SNP microarray testing. Genetics in Medicine, 2012, 14, 971-976.	1.1	21
182	Chromosomal microarray analysis in ocular developmental anomalies. Expert Review of Molecular Diagnostics, 2012, 12, 425-427.	1.5	2
183	Diagnosing fetal alcohol syndrome: new insights from newer genetic technologies. Archives of Disease in Childhood, 2012, 97, 812-817.	1.0	36
184	Application of Genomic Technology in Prenatal Diagnosis. New England Journal of Medicine, 2012, 367, 2249-2251.	13.9	7
185	Congenital diaphragmatic hernia. European Respiratory Journal, 2012, 39, 820-829.	3.1	127
187	Characterizing Small Supernumerary Marker Chromosomes with Combination of Multiple Techniques. Cytogenetic and Genome Research, 2012, 136, 6-14.	0.6	19
188	Clinical Significance of Rare Copy Number Variations in Epilepsy. Archives of Neurology, 2012, 69, 322.	4.9	61
189	Molekulare Karyotypisierung in der Routinediagnostik â€“ RÃ¼ckblick und Ausblick/Molecular karyotyping in routine diagnostics â€“ a view back and forth. Laboratoriums Medizin, 2012, 36, .	0.1	0

#	Article	IF	Citations
190	Clinical Utility of Chromosomal Microarray Analysis. Pediatrics, 2012, 130, e1085-e1095.	1.0	45
191	Clinical implementation of chromosomal microarray technology in prenatal diagnosis (Review). Molecular Medicine Reports, 2012, 6, 1219-1222.	1.1	7
192	5q31 Microdeletions: Definition of a Critical Region and Analysis of <i>LRRTM2,</i> a Candidate Gene for Intellectual Disability. Molecular Syndromology, 2012, 3, 68-75.	0.3	11
193	VACTERL Association Etiology: The Impact of de novo and Rare Copy Number Variations. Molecular Syndromology, 2013, 4, 20-26.	0.3	30
194	So What? Does the Test Lead to Improved Health Outcomes?. Neurology, 2012, 78, 440-442.	1.5	2
195	Exome and genome sequencing of neonates with neurodevelopmental disorders. Future Neurology, 2012, 7, 655-658.	0.9	4
197	Autism screening and diagnostic evaluation. , 0, , 179-200.		1
198	Chromosomal microarrays: influential players in the diagnosis of developmental disorders. Personalized Medicine, 2012, 9, 167-169.	0.8	3
199	Chromosomal microarray testing. JAAPA: Official Journal of the American Academy of Physician Assistants, 2012, 25, 65-68.	0.1	4
200	Interstitial 4q Deletion and Isodicentric Y-Chromosome in a Patient with Dysmorphic Features. Molecular Syndromology, 2012, 3, 39-43.	0.3	3
201	Copy neutral <scp>LOH</scp> of <scp>FANC F</scp> gene: implication on differential diagnosis and treatment of aplastic anemia: letter. European Journal of Haematology, 2012, 89, 365-366.	1.1	0
202	Diagnostic Exome Sequencing in Persons with Severe Intellectual Disability. New England Journal of Medicine, 2012, 367, 1921-1929.	13.9	1,367
203	A Clinical Approach to a Family History of Sudden Death. Circulation: Cardiovascular Genetics, 2012, 5, 697-705.	5.1	6
204	Incidental findings in genetic research and clinical diagnostic tests: A systematic review. American Journal of Medical Genetics, Part A, 2012, 158A, 3159-3167.	0.7	41
205	A genetic model for neurodevelopmental disease. Current Opinion in Neurobiology, 2012, 22, 829-836.	2.0	47
206	Diagnostic Exome Sequencing â€" Are We There Yet?. New England Journal of Medicine, 2012, 367, 1951-1953.	13.9	25
207	Chromosomal Microarray versus Karyotyping for Prenatal Diagnosis. New England Journal of Medicine, 2012, 367, 2175-2184.	13.9	1,082
208	Karyotype versus Microarray Testing for Genetic Abnormalities after Stillbirth. New England Journal of Medicine, 2012, 367, 2185-2193.	13.9	215

#	Article	IF	Citations
210	What is complex about complex disorders?. Genome Biology, 2012, 13, 237.	13.9	85
211	Mosaic 18q21.2 deletions including the TCF4 gene: A clinical report. American Journal of Medical Genetics, Part A, 2012, 158A, 3174-3181.	0.7	10
212	Clinical geneticists' views of VACTERL/VATER association. American Journal of Medical Genetics, Part A, 2012, 158A, 3087-3100.	0.7	78
213	Undetected sex chromosome aneuploidy by chromosomal microarray. Prenatal Diagnosis, 2012, 32, 1117-1118.	1.1	5
214	Impact of genetics on the diagnosis and clinical management of syndromic craniosynostoses. Child's Nervous System, 2012, 28, 1447-1463.	0.6	70
215	Development of an Evidenceâ€Based Information Booklet to Support Parents of Children Without a Diagnosis. Journal of Genetic Counseling, 2012, 21, 854-861.	0.9	3
216	The Laboratoryâ€Clinician Team: A Professional Call to Action to Improve Communication and Collaboration for Optimal Patient Care in Chromosomal Microarray Testing. Journal of Genetic Counseling, 2012, 21, 631-637.	0.9	12
217	Genetic Counseling, Activism and â€˜Genotype-Firstâ€™ Diagnosis of Developmental Disorders. Journal of Genetic Counseling, 2012, 21, 770-776.	0.9	7
218	Molecular Karyotyping as a Relevant Diagnostic Tool in Children with Growth Retardation with Silver-Russell Features. Journal of Pediatrics, 2012, 161, 933-942.e1.	0.9	39
219	A GC-Wave Correction Algorithm that Improves the Analytical Performance of aCGH. Journal of Molecular Diagnostics, 2012, 14, 550-559.	1.2	9
220	Approach to the Genetic Evaluation of the Child with Autism. Pediatric Clinics of North America, 2012, 59, 113-128.	0.9	16
221	Clinical impact of copy number variation analysis using high-resolution microarray technologies: advantages, limitations and concerns. Genome Medicine, 2012, 4, 80.	3.6	63
222	Characterising chromosome rearrangements: recent technical advances in molecular cytogenetics. Heredity, 2012, 108, 75-85.	1.2	79
223	Microdeletions detected using chromosome microarray in children with suspected genetic movement disorders: a singleâ€centre study. Developmental Medicine and Child Neurology, 2012, 54, 618-623.	1.1	68
224	The clinical utility of molecular karyotyping using high-resolution array-comparative genomic hybridization. Expert Review of Molecular Diagnostics, 2012, 12, 449-457.	1.5	16
227	Clinical utilization of high-resolution single nucleotide polymorphism based oligonucleotide arrays in diagnostic studies of pediatric patients with solid tumors. Cancer Genetics, 2012, 205, 42-54.	0.2	18
228	CNVs: Harbingers of a Rare Variant Revolution in Psychiatric Genetics. Cell, 2012, 148, 1223-1241.	13.5	759
229	2q23.1 microdeletion of the MBD5 gene in a female with seizures, developmental delay and distinct dysmorphic features. European Journal of Medical Genetics, 2012, 55, 59-62.	0.7	16

#	Article	IF	Citations
230	Clinical review of genetic epileptic encephalopathies. European Journal of Medical Genetics, 2012, 55, 281-298.	0.7	42
231	Array comparative genomic hybridization: Results from an adult population with drug-resistant epilepsy and co-morbidities. European Journal of Medical Genetics, 2012, 55, 342-348.	0.7	28
232	Ring chromosome 20. European Journal of Medical Genetics, 2012, 55, 381-387.	0.7	40
233	2q23.1 microdeletion of the MBD5 gene in a female with seizures, developmental delay and distinct dysmorphic features. European Journal of Medical Genetics, 2012, 55, 354-357.	0.7	9
234	Genetic architecture in autism spectrum disorder. Current Opinion in Genetics and Development, 2012, 22, 229-237.	1.5	445
235	Microdeletion and microduplication 17q21.31 plus an additional CNV, in patients with intellectual disability, identified by array-CGH. Gene, 2012, 492, 319-324.	1.0	28
236	High resolution array in the clinical approach to chromosomal phenotypes. Gene, 2012, 495, 163-169.	1.0	7
237	Cytogenomic characterization of an unexpected 17.6Mb 9p deletion associated to a 14.8Mb 20p duplication in a dysmorphic patient with multiple congenital anomalies presenting a normal G-banding karyotype. Gene, 2012, 496, 59-62.	1.0	5
238	Chromosomal microarray (CMA) analysis in infants with congenital anomalies: when is it really helpful?. Journal of Maternal-Fetal and Neonatal Medicine, 2012, 25, 116-118.	0.7	11
239	Report of two patients and further characterization of interstitial 9p13 deletionâ€"A rare but recurrent microdeletion syndrome?. American Journal of Medical Genetics, Part A, 2012, 158A, 2328-2335.	0.7	9
240	Lifeâ€history chronicle for a patient with the recently described chromosome 4q21 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2606-2609.	0.7	5
241	Microarray comparative genomic hybridization in prenatal diagnosis: a review. Ultrasound in Obstetrics and Gynecology, 2012, 40, 385-391.	0.9	30
242	Deletion of a single-copy DAAM1 gene in congenital heart defect: a case report. BMC Medical Genetics, 2012, 13, 63.	2.1	15
243	High-resolution SNP array analysis of patients with developmental disorder and normal array CGH results. BMC Medical Genetics, 2012, 13, 84.	2.1	9
244	Severe intellectual disability, omphalocele, hypospadia and high blood pressure associated to a deletion at 2q22.1q22.3: case report. Molecular Cytogenetics, 2012, 5, 30.	0.4	32
245	Array-based comparative genomic hybridization is more informative than conventional karyotyping and fluorescence in situ hybridization in the analysis of first-trimester spontaneous abortion. Molecular Cytogenetics, 2012, 5, 33.	0.4	39
246	Array comparative genomic hybridization in prenatal diagnosis of first trimester pregnancies at high risk for chromosomal anomalies. Molecular Cytogenetics, 2012, 5, 38.	0.4	7
247	Evolving applications of microarray technology in postnatal diagnosis. International Journal of Molecular Medicine, 2012, 30, 223-228.	1.8	7

#	Article	IF	Citations
248	How novel molecular diagnostic technologies and biomarkers are revolutionizing genetic testing and patient care. Expert Review of Molecular Diagnostics, 2012, 12, 25-37.	1.5	15
249	Evidence-based psychiatric genetics, AKA the false dichotomy between common and rare variant hypotheses. Molecular Psychiatry, 2012, 17, 474-485.	4.1	124
250	Genomics, Intellectual Disability, and Autism. New England Journal of Medicine, 2012, 366, 733-743.	13.9	276
251	Online Resources for Genomic Structural Variation. Methods in Molecular Biology, 2012, 838, 273-289.	0.4	6
252	Genomic Literacy and Competent Practice. Nursing Clinics of North America, 2012, 47, 529-545.	0.7	23
253	Solving the Puzzle: Case Examples of Array Comparative Genomic Hybridization as a Tool to End the Diagnostic Odyssey. Current Problems in Pediatric and Adolescent Health Care, 2012, 42, 74-78.	0.8	10
254	Advances in Whole-Genome Genetic Testing: From Chromosomes to Microarrays. Current Problems in Pediatric and Adolescent Health Care, 2012, 42, 47-73.	0.8	18
257	Family History: A Guide for Neurologists in the Age of Genomic Medicine. Seminars in Pediatric Neurology, 2012, 19, 160-166.	1.0	3
258	Updates in the Genetic Evaluation of the Child with Global Developmental Delay or Intellectual Disability. Seminars in Pediatric Neurology, 2012, 19, 173-180.	1.0	60
259	The role of the geneticist and genetic counselor in an ACHD clinic. Progress in Pediatric Cardiology, 2012, 34, 15-20.	0.2	16
260	Uniparental disomy: can SNP array data be used for diagnosis?. Genetics in Medicine, 2012, 14, 753-756.	1.1	27
261	Oligonucleotide Microarrays for Clinical Diagnosis of Copy Number Variation and Zygosity Status. Current Protocols in Human Genetics, 2012, 74, Unit8.12.	3.5	11
262	Infantile hypertrophic pyloric stenosisâ€"genetics and syndromes. Nature Reviews Gastroenterology and Hepatology, 2012, 9, 646-660.	8.2	58
264	Omics. , 2012, , 117-152.		5
265	Human Genome Project, Genomics, andÂClinical Research. , 2012, , 707-725.		0
267	Understanding the Value of Information from Pediatric Clinical Research. Paediatric Drugs, 2012, 14, 295-297.	1.3	0
268	Deletion at chromosome 10p11.23-p12.1 defines characteristic phenotypes with marked midface retrusion. Journal of Human Genetics, 2012, 57, 191-196.	1.1	19
269	Genetics in Pediatric Medicine. , 2012, , 3-12.		0

#	Article	IF	Citations
270	Next Generation Sequencing in the Clinical Domain: Clinical Advantages, Practical, and Ethical Challenges. Advances in Protein Chemistry and Structural Biology, 2012, 89, 27-63.	1.0	21
271	Enhancer Chip: Detecting Human Copy Number Variations in Regulatory Elements. PLoS ONE, 2012, 7, e52264.	1.1	8
272	Copy Number Variants in Obesity-Related Syndromes: Review and Perspectives on Novel Molecular Approaches. Journal of Obesity, 2012, 2012, 1-15.	1.1	30
273	Array CGH in Fetal Medicine Diagnosis. , 0, , .		0
274	The Genetics of Fetal and Neonatal Cardiovascular Disease. , 2012, , 343-376.		0
275	Microdeletions in 16p11.2 and 13q31.3 associated with developmental delay and generalized overgrowth. Genetics and Molecular Research, 2012, 11, 3133-3137.	0.3	1
276	Clinical applications of schizophrenia genetics: genetic diagnosis, risk, and counseling in the molecular era. The Application of Clinical Genetics, 2012, 5, 1.	1.4	35
277	DiagnÃ³stico citogenÃ©tico de pacientes com retardo mental idiopÃ¡tico. Jornal Brasileiro De Patologia E Medicina Laboratorial, 2012, 48, 33-39.	0.3	1
278	"Familial" versus "Sporadic" intellectual disability: contribution of common microdeletion and microduplication syndromes. Molecular Cytogenetics, 2012, 5, 9.	0.4	8
279	Microarray application in prenatal diagnosis: a position statement from the cytogenetics working group of the Italian Society of Human Genetics (SIGU), November 2011. Ultrasound in Obstetrics and Gynecology, 2012, 39, 384-388.	0.9	50
280	Developmental and genetic aspects of congenital diaphragmatic hernia. Pediatric Pulmonology, 2012, 47, 534-545.	1.0	70
281	Genomic microarrays: a technology overview. Prenatal Diagnosis, 2012, 32, 336-343.	1.1	50
282	Prenatal BACsâ€onâ€Beads^{TM}: the prospective experience of five prenatal diagnosis laboratories. Prenatal Diagnosis, 2012, 32, 329-335.	1.1	43
283	Genetic counselling and ethical issues with chromosome microarray analysis in prenatal testing. Prenatal Diagnosis, 2012, 32, 389-395.	1.1	82
284	Referral patterns for microarray testing in prenatal diagnosis. Prenatal Diagnosis, 2012, 32, 344-350.	1.1	32
285	Integration of microarray technology into prenatal diagnosis: counselling issues generated during the NICHD clinical trial. Prenatal Diagnosis, 2012, 32, 396-400.	1.1	61
286	Genome-wide arrays: Quality criteria and platforms to be used in routine diagnostics. Human Mutation, 2012, 33, 906-915.	1.1	69
287	Phenotypic spectrum and genotypeâ€“phenotype correlations of NRXN1 exon deletions. European Journal of Human Genetics, 2012, 20, 1240-1247.	1.4	99

#	Article	IF	Citations
288	Chromosome microarrays in human reproduction. Human Reproduction Update, 2012, 18, 555-567.	5.2	34
289	De novo duplication and deletions at 7q in a threeâ€generation family. American Journal of Medical Genetics, Part A, 2012, 158A, 1493-1497.	0.7	1
290	Ambiguous genitalia: What prenatal genetic testing is practical?. American Journal of Medical Genetics, Part A, 2012, 158A, 1337-1343.	0.7	24
291	Familial Xp22.33â€Xp22.12 deletion delineated by chromosomal microarray analysis causes proportionate short stature. American Journal of Medical Genetics, Part A, 2012, 158A, 1462-1466.	0.7	14
292	Clinical phenotype and candidate genes for the 5q31.3 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 1891-1896.	0.7	25
293	Immunodeficiency, centromeric instability, facial anomalies (ICF) syndrome, due to ZBTB24 mutations, presenting with large cerebral cyst. American Journal of Medical Genetics, Part A, 2012, 158A, 2043-2046.	0.7	25
294	Application of array comparative genomic hybridization in 102 patients with epilepsy and additional neurodevelopmental disorders. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2012, 159B, 760-771.	1.1	48
295	From prenatal genomic diagnosis to fetal personalized medicine: progress and challenges. Nature Medicine, 2012, 18, 1041-1051.	15.2	151
296	Detection and Characterization of Copy Number Variation in Autism Spectrum Disorder. Methods in Molecular Biology, 2012, 838, 115-135.	0.4	72
297	Diagnostic Yield of Chromosomal Microarray Analysis in an Autism Primary Care Practice: Which Guidelines to Implement?. Journal of Autism and Developmental Disorders, 2012, 42, 1582-1591.	1.7	56
299	Array-CGH. Medizinische Genetik, 2012, 24, 99-107.	0.1	3
302	Submicroscopic Chromosomal Copy Number Variations Identified in Children With Hypoplastic Left Heart Syndrome. Pediatric Cardiology, 2012, 33, 757-763.	0.6	35
304	Clinical utility of array comparative genomic hybridisation for prenatal diagnosis: a cohort study of 3171 pregnancies. BJOG: an International Journal of Obstetrics and Gynaecology, 2012, 119, 614-625.	1.1	132
305	Wholeâ€genome array CGH identifies pathogenic copy number variations in fetuses with major malformations and a normal karyotype. Clinical Genetics, 2012, 81, 128-141.	1.0	72
306	Caregiver and adult patient perspectives on the importance of a diagnosis of 22q11.2 deletion syndrome. Journal of Intellectual Disability Research, 2012, 56, 641-651.	1.2	43
307	Aetiology of severe mental retardation and further genetic analysis by highâ€resolution microarray in a populationâ€based series of 6â€to 17â€yearâ€old children. Acta Paediatrica, International Journal of Paediatrics, 2012, 101, 85-91.	0.7	12
308	Dup(3) (p23p25) syndrome and array-based comparative genomic hybridization. Tzu Chi Medical Journal, 2012, 24, 90-91.	0.4	0
309	What can we learn from old microdeletion syndromes using arrayâ€CGH screening?. Clinical Genetics, 2012, 82, 41-47.	1.0	4

#	Article	IF	Citations
310	Towards an evidenceâ€based process for the clinical interpretation of copy number variation. Clinical Genetics, 2012, 81, 403-412.	1.0	93
311	Visualising difference, similarity and belonging in paediatric genetics. Sociology of Health and Illness, 2012, 34, 459-474.	1.1	12
312	Chromosome microarray in Australia: A guide for paediatricians. Journal of Paediatrics and Child Health, 2012, 48, E59-67.	0.4	16
313	Array CGH increasingly used in prenatal and postnatal testing. American Journal of Medical Genetics, Part A, 2012, 158A, viii-ix.	0.7	1
314	Balanced reciprocal translocation t(5;13)(q33;q12) and 9q31.1 microduplication in a man suffering from infertility and pollinosis. Journal of Applied Genetics, 2012, 53, 93-97.	1.0	9
315	Clinical utility of chromosomal microarray analysis in invasive prenatal diagnosis. Human Genetics, 2012, 131, 513-523.	1.8	82
316	The clinical application of array CGH for the detection of chromosomal defects in 20,126 unselected newborns. Molecular Cytogenetics, 2013, 6, 21.	0.4	17
317	Array CGH as a first line diagnostic test in place of karyotyping for postnatal referrals - results from four yearsâ€™ clinical application for over 8,700 patients. Molecular Cytogenetics, 2013, 6, 16.	0.4	54
318	A rare de novo duplication of chromosome 21q22.12â€‰â†’â€‰q22.3 with other concomitant deletion and duplication of small fragments in 21q associated with Down syndrome: Prenatal diagnosis, molecular cytogenetic characterization. Molecular Cytogenetics, 2013, 6, 11.	0.4	6
319	Multiplex ligation-dependent probe amplification workflow for the detection of submicroscopic chromosomal abnormalities in patients with developmental delay/intellectual disability. Molecular Cytogenetics, 2013, 6, 7.	0.4	12
320	Multiple samples aCGH analysis for rare CNVs detection. Journal of Clinical Bioinformatics, 2013, 3, 12.	1.2	2
321	Microarray-Based Cytogenetics. , 2013, , 441-450.		0
322	Deletion 12p12 Involving SOX5 in Two Children With Developmental Delay and Dysmorphic Features. Pediatric Neurology, 2013, 48, 317-320.	1.0	23
323	Detection of Clinically Relevant Copy Number Variants with Whole-Exome Sequencing. Human Mutation, 2013, 34, 1439-1448.	1.1	105
324	Experimental Designs for Array Comparative Genomic Hybridization Technology. Cytogenetic and Genome Research, 2013, 139, 250-257.	0.6	10
325	Prenatal diagnosis: array comparative genomic hybridization in fetuses with abnormal sonographic findings. Acta Obstetricia Et Gynecologica Scandinavica, 2013, 92, 762-768.	1.3	29
326	Technology-Driven and Evidence-Based Genomic Analysis for Integrated Pediatric and Prenatal Genetics Evaluation. Journal of Genetics and Genomics, 2013, 40, 1-14.	1.7	28
327	0.5ÂMb Array as a First-Line Prenatal Cytogenetic Test in Cases without Ultrasound Abnormalities and Its Implementation in Clinical Practice. Human Mutation, 2013, 34, 1298-1303.	1.1	24

#	Article	IF	Citations
328	Meeting the challenge of interpreting highâ€•resolution single nucleotide polymorphism array data in prenatal diagnosis: does increased diagnostic power outweigh the dilemma of rare variants?. BJOG: an International Journal of Obstetrics and Gynaecology, 2013, 120, 594-606.	1.1	34
329	Prenatal diagnosis using combined quantitative fluorescent polymerase chain reaction and array comparative genomic hybridization analysis as a firstâ€line test: results from over 1000 consecutive cases. Ultrasound in Obstetrics and Gynecology, 2013, 41, 500-507.	0.9	54
330	Neurodevelopmental disorders and genetic testing: Current approaches and future advances. Annals of Neurology, 2013, 74, 164-170.	2.8	26
331	Regions of homozygosity identified by SNP microarray analysis aid in the diagnosis of autosomal recessive disease and incidentally detect parental blood relationships. Genetics in Medicine, 2013, 15, 70-78.	1.1	69
332	Microarray-based prenatal diagnosis for the identification of fetal chromosome abnormalities. Expert Review of Molecular Diagnostics, 2013, 13, 601-611.	1.5	15
333	Parentsâ€™ Perspectives on Participating in Genetic Research in Autism. Journal of Autism and Developmental Disorders, 2013, 43, 556-568.	1.7	32
334	The disclosure of incidental genomic findings: an â€œethically important momentâ€•in pediatric research and practice. Journal of Community Genetics, 2013, 4, 435-444.	0.5	25
335	Translational Genetics for Diagnosis of Human Disorders of Sex Development. Annual Review of Genomics and Human Genetics, 2013, 14, 371-392.	2.5	55
336	The â€˜thousand-dollar genomeâ€™: an ethical exploration. European Journal of Human Genetics, 2013, 21, S6-S26.	1.4	56
337	Pathogenic or not? Assessing the clinical relevance of copy number variants. Clinical Genetics, 2013, 84, 415-421.	1.0	44
338	Chromosomal Basis of Inheritance. , 2013, , 1-26.		1
339	Genetic Aspects of Human Epilepsy. , 2013, , 1-40.		0
340	Cytogenetic Analysis. , 2013, , 1-18.		0
341	Neurodevelopmental Disabilities. , 2013, , 1-15.		1
342	Deletions and Other Structural Abnormalities of the Autosomes. , 2013, , 1-37.		4
343	Nature and Frequency of Genetic Disease. , 2013, , 1-4.		3
344	Update on the investigation of children with delayed development. Journal of Paediatrics and Child Health, 2013, 49, 519-525.	0.4	39
345	Velopharyngoplasty in patients with 22q11.2 microdeletion syndrome: outcomes following the Newcastle protocol. European Journal of Plastic Surgery, 2013, 36, 607-618.	0.3	2

#	Article	IF	Citations
346	New Approaches to Molecular Diagnosis. JAMA - Journal of the American Medical Association, 2013, 309, 1511.	3.8	116
347	A 1.1Mb deletion in distal 13q deletion syndrome region with congenital heart defect and postaxial polydactyly: Additional support for a CHD locus at distal 13q34 region. Gene, 2013, 528, 51-54.	1.0	20
348	Confirmation of chromosomal microarray as a first-tier clinical diagnostic test for individuals with developmental delay, intellectual disability, autism spectrum disorders and dysmorphic features. European Journal of Paediatric Neurology, 2013, 17, 589-599.	0.7	170
350	Neurodevelopmental Genomics of Autism, Schizophrenia, and Related Disorders. , 2013, , 695-708.		0
351	Medical conditions affect the outcome of early intervention in preschool children with autism spectrum disorders. European Child and Adolescent Psychiatry, 2013, 22, 23-33.	2.8	44
352	The clinical utility of microarray technologies applied to prenatal cytogenetics in the presence of a normal conventional karyotype: a review of the literature. Prenatal Diagnosis, 2013, 33, 1119-1123.	1.1	140
353	Fragile X Syndrome and X-linked Intellectual Disability. , 2013, , 1-27.		1
354	Mental Retardation (Former Term). , 2013, , 1841-1841.		0
355	Report of interstitial 22q13.1q13.2 microduplication in two siblings with distinctive dysmorphic features, heart defect and mental retardation. European Journal of Medical Genetics, 2013, 56, 389-396.	0.7	7
356	The advantage of using SNP array in clinical testing for hematological malignanciesâ€"a comparative study of three genetic testing methods. Cancer Genetics, 2013, 206, 317-326.	0.2	33
357	Hearing impairment in Estonia: An algorithm to investigate genetic causes in pediatric patients. Advances in Medical Sciences, 2013, 58, 419-428.	0.9	9
358	Cytogenetic Evaluation: A Primer for Pediatric Nurse Practitioners. Journal of Pediatric Health Care, 2013, 27, 426-433.	0.6	2
359	Tetraploidy in the era of molecular karyotyping â€“ What we need to remember. Pediatria Polska, 2013, 88, 467-471.	0.1	3
360	Genetic medicine and incidental findings: it is more complicated than deciding whether to disclose or not. Genetics in Medicine, 2013, 15, 896-899.	1.1	31
361	Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. New England Journal of Medicine, 2013, 369, 1502-1511.	13.9	1,717
362	Disruption of the CREBBP gene and decreased expression of CREB, NFÎºB p65, c-JUN, c-FOS, BCL2 and c-MYC suggest immune dysregulation. Human Immunology, 2013, 74, 911-915.	1.2	6
363	Chromosomal microarray analysis as a first-line test in pregnancies with a priori low risk for the detection of submicroscopic chromosomal abnormalities. European Journal of Human Genetics, 2013, 21, 725-730.	1.4	79
364	Pathogenic rare copy number variants in community-based schizophrenia suggest a potential role for clinical microarrays. Human Molecular Genetics, 2013, 22, 4485-4501.	1.4	120

#	Article	IF	Citations
365	Prenatal diagnosis of chromosomal abnormalities in fetuses with abnormal cardiac ultrasound findings: evaluation of chromosomal microarrayâ€based analysis. Ultrasound in Obstetrics and Gynecology, 2013, 41, 375-382.	0.9	59
366	Frequent detection of parental consanguinity in children with developmental disorders by a combined CGH and SNP microarray. Molecular Cytogenetics, 2013, 6, 38.	0.4	25
367	LINS, a modulator of the WNT signaling pathway, is involved in human cognition. Orphanet Journal of Rare Diseases, 2013, 8, 87.	1.2	21
368	Detection limit of intragenic deletions with targeted array comparative genomic hybridization. BMC Genetics, 2013, 14, 116.	2.7	15
369	Clinical Significance of De Novo and Inherited Copy-Number Variation. Human Mutation, 2013, 34, 1679-1687.	1.1	100
371	ACMG Standards and Guidelines for constitutional cytogenomic microarray analysis, including postnatal and prenatal applications: revision 2013. Genetics in Medicine, 2013, 15, 901-909.	1.1	288
372	Detection of Clinically Relevant Genetic Variants in Autism Spectrum Disorder by Whole-Genome Sequencing. American Journal of Human Genetics, 2013, 93, 249-263.	2.6	429
373	Genomic approaches for studying craniofacial disorders. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 218-231.	0.7	29
374	The use of chromosomal microarray in prenatal diagnosis. The Obstetrician and Gynaecologist, 2013, 15, 80-84.	0.2	7
375	Medizin, 2013, 36, ---.	0.1	0
376	Noninvasive Detection of Fetal Subchromosome Abnormalities via Deep Sequencing of Maternal Plasma. American Journal of Human Genetics, 2013, 92, 167-176.	2.6	228
377	Recent advances in the prenatal interrogation of the human fetal genome. Trends in Genetics, 2013, 29, 84-91.	2.9	46
378	Genetic Basis of Intellectual Disability. Annual Review of Medicine, 2013, 64, 441-450.	5.0	85
379	Autism genetic testing: a qualitative study of awareness, attitudes, and experiences among parents of children with autism spectrum disorders. Genetics in Medicine, 2013, 15, 274-281.	1.1	67
380	Clinical utility of the Xâ€chromosome array. American Journal of Medical Genetics, Part A, 2013, 161, 120-130.	0.7	0
381	Different characteristics identified by single nucleotide polymorphism array analysis in leukemia suggest the need for different application strategies depending on disease category. Genes Chromosomes and Cancer, 2013, 52, 44-55.	1.5	12
382	3q26.33â€“3q27.2 microdeletion: A new microdeletion syndrome?. European Journal of Medical Genetics, 2013, 56, 216-221.	0.7	20
383	Clinical Neurogenetics. Neurologic Clinics, 2013, 31, 951-968.	0.8	4

#	Article	IF	Citations
385	De novo trisomy 20p characterized by array comparative genomic hybridization: Report of a novel case and review of the literature. Gene, 2013, 524, 368-372.	1.0	11
386	The psychological impact of cryptic chromosomal abnormalities diagnosis announcement. European Journal of Medical Genetics, 2013, 56, 585-590.	0.7	13
387	Introduction to the Special Section on Genomics. Child Development, 2013, 84, 6-16.	1.7	8
388	The Utility of Chromosomal Microarray Analysis in Developmental and Behavioral Pediatrics. Child Development, 2013, 84, 121-132.	1.7	67
389	Physicians' perspectives on the uncertainties and implications of chromosomal microarray testing of children and families. Clinical Genetics, 2013, 83, 23-30.	1.0	53
390	Prenatal chromosomal microarray analysis: a survey of prenatal genetic counselors' experiences and attitudes. Prenatal Diagnosis, 2013, 33, 371-377.	1.1	22
391	Cryptic 7q36.2q36.3 deletion causes multiple congenital eye anomalies and craniofacial dysmorphism. American Journal of Medical Genetics, Part A, 2013, 161, 589-593.	0.7	9
392	De novo unbalanced translocation leading to monosomy 9p24.3p24.1 and trisomy 19q13.42q13.43 characterized by microarrayâ€based comparative genomic hybridization in a child with partial cortical dysplasia and craniofacial dysmorphisms without trigonocephaly. American Journal of Medical Genetics, Part A, 2013, 161, 632-636.	0.7	6
393	Utility of chromosomal microarray in five cases with cytogenetic abnormalities detected by traditional karyotype. Clinical Genetics, 2013, 84, 600-602.	1.0	2
394	Autism spectrum disorder in the genetics clinic: a review. Clinical Genetics, 2013, 83, 399-407.	1.0	153
395	8p23.1 duplication syndrome; common, confirmed, and novel features in six further patients. American Journal of Medical Genetics, Part A, 2013, 161, 487-500.	0.7	47
396	Array CGH in patients with developmental delay or intellectual disability: are there phenotypic clues to pathogenic copy number variants?. Clinical Genetics, 2013, 83, 53-65.	1.0	60
397	Clinical application of 2.7M Cytogenetics array for CNV detection in subjects with idiopathic autism and/or intellectual disability. Clinical Genetics, 2013, 83, 145-154.	1.0	43
398	A parallel study of different array-CGH platforms in a set of Spanish patients with developmental delay and intellectual disability. Gene, 2013, 521, 82-86.	1.0	15
399	Clinical genetics evaluation in identifying the etiology of autism spectrum disorders: 2013 guideline revisions. Genetics in Medicine, 2013, 15, 399-407.	1.1	402
400	Density matters: comparison of array platforms for detection of copy-number variation and copy-neutral abnormalities. Genetics in Medicine, 2013, 15, 706-712.	1.1	24
401	The future of prenatal diagnosis: karyotype, microarray or both? Technical and ethical considerations. Expert Review of Proteomics, 2013, 10, 131-134.	1.3	6
402	Prenatal diagnosis using array-CGH: A French experience. European Journal of Medical Genetics, 2013, 56, 341-345.	0.7	31

#	Article	IF	Citations
403	Haploinsufficiency of ZNF238 is associated with corpus callosum abnormalities in 1q44 deletions. American Journal of Medical Genetics, Part A, 2013, 161, 711-716.	0.7	28
404	Counting Chromosomes to Exons: Advances in Copy Number Detection. Current Genetic Medicine Reports, 2013, 1, 71-80.	1.9	1
405	The Future of Prenatal Cytogenetics: From Copy Number Variations to Non-invasive Prenatal Testing. Current Genetic Medicine Reports, 2013, 1, 91-98.	1.9	1
406	High resolution chromosomal microarray in undiagnosed neurological disorders. Journal of Paediatrics and Child Health, 2013, 49, 716-724.	0.4	18
407	Here/In This Issue and There/Abstract Thinking: Genetic Testing in Neurodevelopmental Disorders. Journal of the American Academy of Child and Adolescent Psychiatry, 2013, 52, 449-450.	0.3	0
408	Customized high resolution CGHâ€array for clinical diagnosis reveals additional genomic imbalances in previous wellâ€defined pathological samples. American Journal of Medical Genetics, Part A, 2013, 161, 1950-1960.	0.7	32
409	The Genetics of Autism Spectrum Disorders â€“ A Guide for Clinicians. Current Psychiatry Reports, 2013, 15, 334.	2.1	49
410	Clinical Applications of Whole-Genome Chromosomal Microarray Analysis. , 2013, , 133-144.		3
411	Molecular karyotype single nucleotide polymorphism analysis of early fetal demise. Systems Biology in Reproductive Medicine, 2013, 59, 227-231.	1.0	14
412	Comparison of chromosome analysis and chromosomal microarray analysis: what is the value of chromosome analysis in todayâ€™s genomic array era?. Genetics in Medicine, 2013, 15, 450-457.	1.1	63
413	Assessing the Minimally Verbal Schoolâ€Aged Child With Autism Spectrum Disorder. Autism Research, 2013, 6, 479-493.	2.1	219
414	Targeted array comparative genomic hybridization â€“ A new diagnostic tool for the detection of large copy number variations in nemaline myopathy-causing genes. Neuromuscular Disorders, 2013, 23, 56-65.	0.3	31
415	Re: Prenatal diagnosis using combined quantitative fluorescent polymerase chain reaction and array comparative genomic hybridization analysis as a firstâ€line test: results from over 1000 consecutive cases. F. Scott, K. Murphy, L. Carey, W. Greville, N. Mansfield, P. Barahona, R. Robertson and A. McLennan. Ultrasound Obstet Gynecol 2013; 41: 500â€“507. Ultrasound in Obstetrics and Gynecology, 2013, 41, 489-489.	0.9	1
416	Can microarray analysis reveal additional abnormalities in prenatal diagnosis?. Expert Review of Obstetrics and Gynecology, 2013, 8, 193-195.	0.4	3
417	Both Rare and De Novo Copy Number Variants Are Prevalent in Agenesis of the Corpus Callosum but Not in Cerebellar Hypoplasia or Polymicrogyria. PLoS Genetics, 2013, 9, e1003823.	1.5	69
418	Rare Copy Number Variants Are a Common Cause of Short Stature. PLoS Genetics, 2013, 9, e1003365.	1.5	60
420	Structural Chromosome Rearrangements. , 2013, , 139-174.		3
421	CNVinspector:a web-based tool for the interactive evaluation of copy number variations in single patients and in cohorts. Journal of Medical Genetics, 2013, 50, 529-533.	1.5	3

#	Article	IF	Citations
422	Current Progress and Challenges in the Search for Autism Biomarkers. Disease Markers, 2013, 35, 55-65.	0.6	36
423	A Dynamic Database of Microarray-Characterized Cell Lines with Various Cytogenetic and Genomic Backgrounds. G3: Genes, Genomes, Genetics, 2013, 3, 1143-1149.	0.8	12
424	Ethical controversies in prenatal microarray. Current Opinion in Obstetrics and Gynecology, 2013, 25, 133-137.	0.9	23
425	Autisms. , 2013, , 651-694.		9
426	Expanding probe repertoire and improving reproducibility in human genomic hybridization. Nucleic Acids Research, 2013, 41, e81-e81.	6.5	12
427	High-throughput sequencing of autism spectrum disorders comes of age. Genetical Research, 2013, 95, 121-129.	0.3	4
428	The NICE guideline on recognition, referral, diagnosis and management of adults on the autism spectrum. Advances in Mental Health and Intellectual Disabilities, 2013, 8, 3-14.	0.7	21
429	Chromosomal Microarrays in Prenatal Diagnosis: Time for a Change of Policy?. Microarrays (Basel,) Tj ETQq1 1 0.784314 rgBT /Overlo	1.4	9
430	Genetic Literacy and Competency. Pediatrics, 2013, 132, S224-S230.	1.0	22
431	Key Informantsâ€™ Perspectives of Implementing Chromosomal Microarrays Into Clinical Practice in Australia. Twin Research and Human Genetics, 2013, 16, 833-839.	0.3	10
432	BBGRE: brain and body genetic resource exchange. Database: the Journal of Biological Databases and Curation, 2013, 2013, bat067.	1.4	4
433	Providing family guidance in rapidly shifting sand: informed consent for genetic testing. Developmental Medicine and Child Neurology, 2013, 55, 766-768.	1.1	12
434	Impact of new genomic tools on the practice of clinical genetics in consanguineous populations: the Saudi experience. Clinical Genetics, 2013, 84, 203-208.	1.0	44
435	Evaluation of Real-Time Quantitative PCR as a Standard Cytogenetic Diagnostic Tool for Confirmation of Microarray (aCGH) Results and Determination of Inheritance. Genetic Testing and Molecular Biomarkers, 2013, 17, 821-825.	0.3	5
436	An Update of Childhood Genetic Disorders. Journal of Nursing Scholarship, 2013, 45, 34-42.	1.1	4
437	Expanding diagnostic testing beyond cytogenetics: Implications for birth defects research and surveillance. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 726-729.	1.6	2
438	Prenatal Diagnosis of Chromosome Abnormalities: Past, Present, and Future. Clinical Chemistry, 2013, 59, 1432-1434.	1.5	2
439	Estimates of penetrance for recurrent pathogenic copy-number variations. Genetics in Medicine, 2013, 15, 478-481.	1.1	277

#	Article	IF	Citations
440	Hypothesis-generating research and predictive medicine. Genome Research, 2013, 23, 1051-1053.	2.4	66
441	Effect of Copy Number Variants on Outcomes for Infants With Single Ventricle Heart Defects. Circulation: Cardiovascular Genetics, 2013, 6, 444-451.	5.1	89
442	Genetic pathways to autism spectrum disorders. Neuropsychiatry, 2013, 3, 193-207.	0.4	4
443	Prenatal microarray analysis as secondâ€tier diagnostic test: singleâ€center prospective study. Ultrasound in Obstetrics and Gynecology, 2013, 41, 267-273.	0.9	14
444	Array CGH Analysis and Developmental Delay: A Diagnostic Tool for Neurologists. Canadian Journal of Neurological Sciences, 2013, 40, 777-782.	0.3	1
445	Genetic Testing of Epileptic Encephalopathies of Infancy: An Approach. Canadian Journal of Neurological Sciences, 2013, 40, 10-16.	0.3	21
446	Early diagnosis of autism and impact on prognosis: a narrative review. Clinical Epidemiology, 2013, 5, 33.	1.5	150
447	Mouse Behavioral Models for Autism Spectrum Disorders. , 2013, , 363-378.		0
448	Autism Spectrum Disorders, Risk Communication, and the Problem of Inadvertent Harm. Kennedy Institute of Ethics Journal, 2013, 23, 105-138.	0.3	27
449	Children with Autism Spectrum Disorder and Epilepsy. Pediatric Annals, 2013, 42, 255-60.	0.3	14
450	Prioritization of Copy Number Variation Loci Associated with Autism from AutDBâ€“An Integrative Multi-Study Genetic Database. PLoS ONE, 2013, 8, e66707.	1.1	13
451	Array-Based Comparative Genomic Hybridization in 190 Korean Patients with Developmental Delay and/or Intellectual Disability: A Single Tertiary Care University Center Study. Yonsei Medical Journal, 2013, 54, 1463.	0.9	10
452	Sex bias in copy number variation of olfactory receptor gene family depends on ethnicity. Frontiers in Genetics, 2013, 4, 32.	1.1	7
453	Genetic Etiology of Autism. , 0, , .		1
454	Genetic Evaluation of Individuals with Autism Spectrum Disorders. , 0, , .		0
455	Cytogenomic Abnormalities and Dosage-Sensitive Mechanisms for Intellectual and Developmental Disabilities. , 2013, , .		1
456	Mutual Gaze. , 2013, , 1966-1967.		0
457	In Situ Hybridization. , 2014, , 699-704.		0

#	Article	IF	Citations
458	Gene/Genome Mutation Detection and Testing. , 2014, , 3408-3417.		1
459	The Prevalence of Chromosomal Deletions Relating to Developmental Delay and/or Intellectual Disability in Human Euploid Blastocysts. PLoS ONE, 2014, 9, e85207.	1.1	10
460	Uptake and Diagnostic Yield of Chromosomal Microarray in an Australian Child Development Clinic. Children, 2014, 1, 21-30.	0.6	6
461	Microarray Technology for the Diagnosis of Fetal Chromosomal Aberrations: Which Platform Should We Use?. Journal of Clinical Medicine, 2014, 3, 663-678.	1.0	13
462	Prenatal management, pregnancy and pediatric outcomes in fetuses with septated cystic hygroma. Brazilian Journal of Medical and Biological Research, 2014, 47, 799-803.	0.7	18
463	Exonic deletion of OPHN1 resulting in seizures, intellectual disability, and brain malformations. Advances in Genomics and Genetics, 2014, , 107.	0.8	0
464	Genetic Aspects of Human Epilepsy. , 2014, , .		0
465	Impact of NGS in the medical sciences: genetic syndromes with an increased risk of developing cancer as an example of the use of new technologies. Genetics and Molecular Biology, 2014, 37, 241-249.	0.6	12
469	Deletions and Other Structural Abnormalities of the Autosomes. , 2014, , .		0
471	Endocrinological anomalies in a patient with 12q14 microdeletion syndrome. Completing phenotype of this exceptional short stature condition. Journal of Pediatric Endocrinology and Metabolism, 2014, 27, 539-43.	0.4	4
472	A common cognitive, psychiatric, and dysmorphic phenotype in carriers of <scp>NRXN1</scp> deletion. Molecular Genetics & Genomic Medicine, 2014, 2, 512-521.	0.6	32
475	A de novo 2.9ÂMb interstitial deletion at 13q12.11 in a child with developmental delay accompanied by mild dysmorphic characteristics. Molecular Cytogenetics, 2014, 7, 92.	0.4	2
476	Clinical Genomics: When Whole Genome Sequencing Is like a Whole-body CT Scan. Clinical Chemistry, 2014, 60, 1390-1392.	1.5	6
477	Familial 1q22 microduplication associated with psychiatric disorders, intellectual disability and late-onset autoimmune inflammatory response. Molecular Cytogenetics, 2014, 7, 90.	0.4	5
478	SNP arrays: comparing diagnostic yields for four platforms in children with developmental delay. BMC Medical Genomics, 2014, 7, 70.	0.7	13
479	Subjectivity in chromosome bandâ€“level estimation: a multicenter study. Genetics in Medicine, 2014, 16, 170-175.	1.1	3
480	The utility of the traditional medical genetics diagnostic evaluation in the context of next-generation sequencing for undiagnosed genetic disorders. Genetics in Medicine, 2014, 16, 176-182.	1.1	239
481	Comprehensive Evaluation of the Child With Intellectual Disability or Global Developmental Delays. Pediatrics, 2014, 134, e903-e918.	1.0	412

#	Article	IF	Citations
482	Molecular and Cytogenetic Analysis in Stillbirth: Results from 481 Consecutive Cases. Fetal Diagnosis and Therapy, 2014, 36, 326-332.	0.6	22
483	Exome sequencing and diffusion tensor imaging in developmental disabilities. Pediatric Research, 2014, 75, 443-447.	1.1	8
484	Clinical interpretation of CNVs with cross-species phenotype data. Journal of Medical Genetics, 2014, 51, 766-772.	1.5	23
485	Autism spectrum mixed neurodevelopmental disorder associated with 6q27 deletion and multiple copies within 20q11.23: a case study. Advances in Mental Health and Intellectual Disabilities, 2014, 8, 210-215.	0.7	0
486	Developments in renal pharmacogenomics and applications in chronic kidney disease. Pharmacogenomics and Personalized Medicine, 2014, 7, 251.	0.4	7
487	A New Case of 13q12.2q13.1 Microdeletion Syndrome Contributes to Phenotype Delineation. Case Reports in Genetics, 2014, 2014, 1-5.	0.1	3
488	The trading zone of autism genetics: Examining the intersection of genomic and psychiatric classification. BioSocieties, 2014, 9, 329-352.	0.8	15
489	Prenatal Chromosomal Microarray for the Catholic Physician. Linacre quarterly, The, 2014, 81, 162-171.	0.1	0
490	Homozygous Truncating Intragenic Duplication in TUSC3 Responsible for Rare Autosomal Recessive Nonsyndromic Intellectual Disability with No Clinical or Biochemical Metabolic Markers. JIMD Reports, 2014, 20, 45-55.	0.7	12
491	Genetic testing and corresponding services among individuals with autism spectrum disorder (ASD). American Journal of Medical Genetics, Part A, 2014, 164, 2592-2600.	0.7	30
492	Perceived burden and neuropsychiatric morbidities in adults with 22q11.2 deletion syndrome. Journal of Intellectual Disability Research, 2014, 58, 198-210.	1.2	23
493	BACs-on-beads: a new robust and rapid detection method for prenatal diagnosis. Expert Review of Molecular Diagnostics, 2014, 14, 273-280.	1.5	14
494	Mutation Screening of the Neurexin 1 Gene in Thai Patients with Intellectual Disability and Autism Spectrum Disorder. Genetic Testing and Molecular Biomarkers, 2014, 18, 510-515.	0.3	14
495	Confined placental mosaicism: implications for fetal chromosomal analysis using microarray comparative genomic hybridization. Prenatal Diagnosis, 2014, 34, 98-101.	1.1	14
496	Advanced bone age in a girl with Wiedemannâ€“Steiner syndrome and an exonic deletion in KMT2A (MLL). American Journal of Medical Genetics, Part A, 2014, 164, 2079-2083.	0.7	35
497	A Diagnostic Approach for Cerebral Palsy in the Genomic Era. NeuroMolecular Medicine, 2014, 16, 821-844.	1.8	89
498	Ethical Issues in DNA Sequencing in the Neonate. Clinics in Perinatology, 2014, 41, 993-1000.	0.8	5
499	19q13.32 microdeletion syndrome: Three new cases. European Journal of Medical Genetics, 2014, 57, 654-658.	0.7	17

#	Article	IF	Citations
500	Diagnostic whole genome sequencing and splitâ€read mapping for nucleotide resolution breakpoint identification in CNTNAP2 deficiency syndrome. American Journal of Medical Genetics, Part A, 2014, 164, 2649-2655.	0.7	29
501	Controversies in prenatal diagnosis 3: should everyone undergoing invasive testing have a microarray?. Prenatal Diagnosis, 2014, 34, 18-22.	1.1	18
502	Chromosomal microarray analysis as a firstâ€tier clinical diagnostic test: E stonian experience. Molecular Genetics & Genomic Medicine, 2014, 2, 166-175.	0.6	22
503	Implementation of highâ€resolution <scp>SNP</scp> arrays in the investigation of fetuses with ultrasound malformations: 5â€‰years of clinical experience. Clinical Genetics, 2014, 86, 264-269.	1.0	25
504	Novel interstitial 2.6â€‰Mb deletion on 9q21 associated with multiple congenital anomalies. American Journal of Medical Genetics, Part A, 2014, 164, 237-242.	0.7	13
505	Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing. JAMA - Journal of the American Medical Association, 2014, 312, 1870.	3.8	1,171
506	Molecular Mechanisms Underpinning the Development of Obesity. , 2014, , .		6
507	Is high fetal nuchal translucency associated with submicroscopic chromosomal abnormalities on array <scp>CGH</scp>?. Ultrasound in Obstetrics and Gynecology, 2014, 43, 620-624.	0.9	49
508	Diagnostic Yield of Clinical Next-Generation Sequencing Panels for Epilepsy. JAMA Neurology, 2014, 71, 650.	4.5	54
509	Genome-wide screening of copy number variants in children born small for gestational age reveals several candidate genes involved in growth pathways. European Journal of Endocrinology, 2014, 171, 253-262.	1.9	36
510	A Case with a Ring Chromosome 13 in a Cohort of 203 Children with Non-Syndromic Autism and Review of the Cytogenetic Literature. Cytogenetic and Genome Research, 2014, 144, 1-8.	0.6	20
511	Chromosome microarray testing for patients with congenital heart defects reveals novel disease causing loci and high diagnostic yield. BMC Genomics, 2014, 15, 1127.	1.2	74
512	De Novo variants in the KMT2A (MLL) gene causing atypical Wiedemann-Steiner syndrome in two unrelated individuals identified by clinical exome sequencing. BMC Medical Genetics, 2014, 15, 49.	2.1	49
513	Array-CGH analysis in Rwandan patients presenting development delay/intellectual disability with multiple congenital anomalies. BMC Medical Genetics, 2014, 15, 79.	2.1	11
514	Copy number variants (CNVs) analysis in a deeply phenotyped cohort of individuals with intellectual disability (ID). BMC Medical Genetics, 2014, 15, 82.	2.1	12
515	Rescue karyotyping: a case series of array-based comparative genomic hybridization evaluation of archival conceptual tissue. Reproductive Biology and Endocrinology, 2014, 12, 19.	1.4	21
516	Phelan-McDermid syndrome: a review of the literature and practice parameters for medical assessment and monitoring. Journal of Neurodevelopmental Disorders, 2014, 6, 39.	1.5	122
517	Large cryptic genomic rearrangements with apparently normal karyotypes detected by array-CGH. Molecular Cytogenetics, 2014, 7, 82.	0.4	25

#	Article	IF	Citations
518	Multiplex ligation-dependent probe amplification and array comparative genomic hybridization analyses for prenatal diagnosis of cytogenomic abnormalities. Molecular Cytogenetics, 2014, 7, 84.	0.4	18
519	Clinical utility of chromosomal microarray analysis in the diagnosis and management of monosomy 7 mosaicism. Molecular Cytogenetics, 2014, 7, 93.	0.4	2
520	Deciphering the pathogenic consequences of chromosomal aberrations in human genetic disease. Molecular Cytogenetics, 2014, 7, 100.	0.4	30
521	Effectiveness of exome and genome sequencing guided by acuity of illness for diagnosis of neurodevelopmental disorders. Science Translational Medicine, 2014, 6, 265ra168.	5.8	440
522	An approach to pediatric exome and genome sequencing. Current Opinion in Pediatrics, 2014, 26, 639-645.	1.0	23
523	Cognitive deficit and autism spectrum disorders: prospective diagnosis by array CGH. Pathology, 2014, 46, 41-45.	0.3	18
524	The Correct Genetic Diagnosis Has Already Been Determined More Often Than We Think, Letâ€™s Report It to the Parents!. Clinical Pediatrics, 2014, 53, 8-10.	0.4	1
525	A Summary of Current Understanding Regarding Children with Autism Spectrum Disorder Who Are Deaf or Hard of Hearing. Seminars in Speech and Language, 2014, 35, 241-259.	0.5	22
526	Secondary Complex Chromosome Rearrangement Identified by Chromosome Analysis and FISH Subsequent to Detection of an Unbalanced Derivative Chromosome 12 by SNP Array Analysis. Cytogenetic and Genome Research, 2014, 142, 129-133.	0.6	2
527	The clinical significance of small copy number variants in neurodevelopmental disorders. Journal of Medical Genetics, 2014, 51, 677-688.	1.5	72
528	The Autism Simplex Collection: an international, expertly phenotyped autism sample for genetic and phenotypic analyses. Molecular Autism, 2014, 5, 34.	2.6	31
529	Whole-Genome Single Nucleotide Polymorphism Array Analysis Is Complementary to Classical Cytogenetic Analysis in the Evaluation of Lymphoid Proliferations. American Journal of Clinical Pathology, 2014, 141, 247-255.	0.4	7
530	Monosomy 20 Mosaicism Revealed by Extensive Karyotyping in Blood and Skin Cells: Case Report and Review of the Literature. Cytogenetic and Genome Research, 2014, 144, 155-162.	0.6	7
531	Copy number variations and human genetic disease. Current Opinion in Pediatrics, 2014, 26, 646-652.	1.0	36
532	Advances in genetic prenatal diagnosis and screening. Current Opinion in Pediatrics, 2014, 26, 634-638.	1.0	16
533	22q11.2 deletion syndrome as a risk factor for aortic root dilation in tetralogy of Fallot. Cardiology in the Young, 2014, 24, 303-310.	0.4	26
534	Autism. Lancet, The, 2014, 383, 896-910.	6.3	1,719
535	A Refined Approach to Evaluating Global Developmental Delay for the International Medical Community. Pediatric Neurology, 2014, 51, 198-206.	1.0	25

#	Article	IF	Citations
537	Genetic testing utilization and the role of the laboratory genetic counselor. Clinica Chimica Acta, 2014, 427, 193-195.	0.5	32
538	Copy number variation and autism: New insights and clinical implications. Journal of the Formosan Medical Association, 2014, 113, 400-408.	0.8	36
539	Disruption of MBD5 contributes to a spectrum of psychopathology and neurodevelopmental abnormalities. Molecular Psychiatry, 2014, 19, 368-379.	4.1	54
540	Making headway with genetic diagnostics of intellectual disabilities. Clinical Genetics, 2014, 85, 101-110.	1.0	42
541	Diagnostic yield of array comparative genomic hybridization in adults with autism spectrum disorders. Genetics in Medicine, 2014, 16, 70-77.	1.1	19
542	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	1.4	112
543	Towards a safety net for management of 22q11.2 deletion syndrome: guidelines for our times. European Journal of Pediatrics, 2014, 173, 757-765.	1.3	62
544	An Interstitial 4q31.21q31.22 Microdeletion Associated with Developmental Delay: Case Report and Literature Review. Cytogenetic and Genome Research, 2014, 142, 227-238.	0.6	8
545	9q31.1q31.3 deletion in two patients with similar clinical features: A newly recognized microdeletion syndrome?. American Journal of Medical Genetics, Part A, 2014, 164, 685-690.	0.7	9
546	Microarrays as a diagnostic tool in prenatal screening strategies: ethical reflection. Human Genetics, 2014, 133, 163-172.	1.8	35
547	An Exploration of Genetic Counselorsâ€™ Needs and Experiences with Prenatal Chromosomal Microarray Testing. Journal of Genetic Counseling, 2014, 23, 938-947.	0.9	45
548	Neuregulin 1-Induced AKT and ERK Phosphorylation in Patients with Fragile X Syndrome (FXS) and Intellectual Disability Associated with Obstetric Complications. Journal of Molecular Neuroscience, 2014, 54, 119-124.	1.1	5
549	Considering specific clinical features as evidence of pathogenic copy number variants. Journal of Applied Genetics, 2014, 55, 189-196.	1.0	17
550	Cytogenomic mapping and bioinformatic mining reveal interacting brain expressed genes for intellectual disability. Molecular Cytogenetics, 2014, 7, 4.	0.4	19
551	Clinical and genomic evaluation of 201 patients with Phelanâ€“McDermid syndrome. Human Genetics, 2014, 133, 847-859.	1.8	142
552	Duplication at Xq13.3â€“q21.1 with syndromic intellectual disability, a probable role for the ATRX gene. American Journal of Medical Genetics, Part A, 2014, 164, 918-923.	0.7	10
553	A prospective study of the clinical utility of prenatal chromosomal microarray analysis in fetuses with ultrasound abnormalities and an exploration of a framework for reporting unclassified variants and risk factors. Genetics in Medicine, 2014, 16, 469-476.	1.1	66
554	Changing interpretation of chromosomal microarray over time in a community cohort with intellectual disability. American Journal of Medical Genetics, Part A, 2014, 164, 377-385.	0.7	37

#	Article	IF	Citations
555	The communication of secondary variants: interviews with parents whose children have undergone arrayâ€<scp>CGH</scp> testing. Clinical Genetics, 2014, 86, 207-216.	1.0	20
556	Practice Parameter for the Assessment and Treatment of Children and Adolescents With Autism Spectrum Disorder. Journal of the American Academy of Child and Adolescent Psychiatry, 2014, 53, 237-257.	0.3	558
557	Diagnostic approach to microcephaly in childhood: a twoâ€center study and review of the literature. Developmental Medicine and Child Neurology, 2014, 56, 732-741.	1.1	176
558	Prenatal diagnosis of chromosomal mosaicism in over 1600 cases using array comparative genomic hybridization as a first line test. Prenatal Diagnosis, 2014, 34, 478-486.	1.1	21
559	The metabolic evaluation of the child with an intellectual developmental disorder: Diagnostic algorithm for identification of treatable causes and new digital resource. Molecular Genetics and Metabolism, 2014, 111, 428-438.	0.5	110
560	An economic analysis of prenatal cytogenetic technologies for sonographically detected fetal anomalies. American Journal of Medical Genetics, Part A, 2014, 164, 1192-1197.	0.7	14
561	A newly recognized 13q12.3 microdeletion syndrome characterized by intellectual disability, microcephaly, and eczema/atopic dermatitis encompassing the HMGB1 and KATNAL1 genes. American Journal of Medical Genetics, Part A, 2014, 164, 1277-1283.	0.7	25
562	Copy number variation plays an important role in clinical epilepsy. Annals of Neurology, 2014, 75, 943-958.	2.8	147
563	The Database of Genomic Variants: a curated collection of structural variation in the human genome. Nucleic Acids Research, 2014, 42, D986-D992.	6.5	1,033
564	Single exon-resolution targeted chromosomal microarray analysis of known and candidate intellectual disability genes. European Journal of Human Genetics, 2014, 22, 792-800.	1.4	35
566	A prenatal case of split-hand malformation associated with 17p13.3 triplication â€“ A dilemma in genetic counseling. European Journal of Medical Genetics, 2014, 57, 81-84.	0.7	11
567	Chromosomal microarray analysis of consecutive individuals with autism spectrum disorders or learning disability presenting for genetic services. Gene, 2014, 535, 70-78.	1.0	94
568	Genomics, Personalized Medicine, and Pediatrics. Academic Pediatrics, 2014, 14, 14-22.	1.0	29
569	Application of array comparative genomic hybridization in 256 patients with developmental delay or intellectual disability. Journal of Applied Genetics, 2014, 55, 125-144.	1.0	37
571	Genetic testing and genetic counseling among medicaid-enrolled children with autism spectrum disorder in 2001 and 2007. Human Genetics, 2014, 133, 111-116.	1.8	23
572	Genetic Testing Practices in Infants with Congenital Heart Disease. Congenital Heart Disease, 2014, 9, 158-167.	0.0	46
573	Highâ€resolution microarray in the assessment of fetal anomalies detected by ultrasound. Australian and New Zealand Journal of Obstetrics and Gynaecology, 2014, 54, 46-52.	0.4	14
574	Adult neuropsychiatric expression and familial segregation of 2q13 duplications. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 337-344.	1.1	23

#	Article	IF	Citations
575	Clinical utility of chromosomal microarray analysis in prenatal diagnosis: report of first 6 months in clinical practice. Journal of Maternal-Fetal and Neonatal Medicine, 2014, 27, 1333-1338.	0.7	20
576	Detectie van submicroscopische chromosomale afwijkingen door middel van array-diagnostiek. Tijdschrift Voor Kindergeneeskunde, 2014, 82, 3-18.	0.0	0
577	Exoom-sequencing in de diagnostiek van ontwikkelingsachterstand/verstandelijke beperking. Tijdschrift Voor Kindergeneeskunde, 2014, 82, 35-44.	0.0	0
578	Developing patient-friendly genetic and genomic test reports: formats to promote patient engagement and understanding. Genome Medicine, 2014, 6, 58.	3.6	71
579	Cryptic and Complex Chromosomal Aberrations in Early-Onset Neuropsychiatric Disorders. American Journal of Human Genetics, 2014, 95, 454-461.	2.6	45
580	Genomic Analyses of Patients With Unexplained Early-Onset Scoliosis. Spine Deformity, 2014, 2, 324-332.	0.7	9
581	Case 24-2014. New England Journal of Medicine, 2014, 371, 465-472.	13.9	9
582	Copy Number Variation in Obsessive-Compulsive Disorder and Tourette Syndrome: A Cross-Disorder Study. Journal of the American Academy of Child and Adolescent Psychiatry, 2014, 53, 910-919.	0.3	111
584	CNVs in Epilepsy. Current Genetic Medicine Reports, 2014, 2, 162-167.	1.9	28
585	Genomic Variation: Lessons Learned from Whole-Genome CNV Analysis. Current Genetic Medicine Reports, 2014, 2, 146-150.	1.9	16
586	A new case of de novo 19p13.2p13.12 deletion in a girl with overgrowth and severe developmental delay. Molecular Cytogenetics, 2014, 7, 40.	0.4	14
587	Severe growth hormone deficiency and pituitary malformation in a patient with chromosome 2p25 duplication and 2q37 deletion. Molecular Cytogenetics, 2014, 7, 41.	0.4	11
588	A non-syndromic intellectual disability associated with a de novo microdeletion at 7q and 18p, microduplication at Xp, and 18q partial trisomy detected using chromosomal microarray analysis approach. Molecular Cytogenetics, 2014, 7, 44.	0.4	8
589	Copy number variation in Han Chinese individuals with autism spectrum disorder. Journal of Neurodevelopmental Disorders, 2014, 6, 34.	1.5	55
590	New technologies in molecular genetics. Progress in Brain Research, 2014, 213, 253-278.	0.9	6
591	Three Clinical Experiences with SNP Array Results Consistent with Parental Incest: A Narrative with Lessons Learned. Journal of Genetic Counseling, 2014, 23, 489-495.	0.9	4
592	Chromosomal microarray analysisâ€"a routine clinical genetic test for patients with schizophrenia. Lancet Psychiatry,the, 2014, 1, 329-331.	3.7	22
595	A Qualitative Study of Healthcare Providersâ€™ Perspectives on the Implications of Genomeâ€Wide Testing in Pediatric Clinical Practice. Journal of Genetic Counseling, 2014, 23, 474-488.	0.9	17

#	Article	IF	Citations
596	Predictive diagnostic value for the clinical features accompanying intellectual disability in children with pathogenic copy number variations: a multivariate analysis. Italian Journal of Pediatrics, 2014, 40, 39.	1.0	10
597	A novel microdeletion involving the 13q31.3â€“q32.1 region in a patient with normal intelligence. European Journal of Medical Genetics, 2014, 57, 60-64.	0.7	8
598	The impact of chromosomal microarray on clinical management: a retrospective analysis. Genetics in Medicine, 2014, 16, 657-664.	1.1	66
599	Mate pair sequencing for the detection of chromosomal aberrations in patients with intellectual disability and congenital malformations. European Journal of Human Genetics, 2014, 22, 652-659.	1.4	32
600	Advances in Genetic Diagnosis of Autism Spectrum Disorders. Current Pediatrics Reports, 2014, 2, 71-81.	1.7	3
601	Corticosteroid therapy in regressive autism: a retrospective study of effects on the Frequency Modulated Auditory Evoked Response (FMAER), language, and behavior. BMC Neurology, 2014, 14, 70.	0.8	36
602	Adult expression of a 3q13.31 microdeletion. Molecular Cytogenetics, 2014, 7, 23.	0.4	10
603	Performance of chromosomal microarray for patients with intellectual disabilities/developmental delay, autism, and multiple congenital anomalies in a Chinese cohort. Molecular Cytogenetics, 2014, 7, 34.	0.4	33
604	The Genetics of Microdeletion and Microduplication Syndromes: An Update. Annual Review of Genomics and Human Genetics, 2014, 15, 215-244.	2.5	145
605	Genome-wide UPD screening in patients with intellectual disability. European Journal of Human Genetics, 2014, 22, 1233-1235.	1.4	3
606	Development and Developmental Disorders of the Cerebral Cortex. , 2014, , 523-642.		5
607	Detection and Reporting of Homozygosity Associated with Consanguinity in the Clinical Laboratory. Human Heredity, 2014, 77, 217-224.	0.4	25
608	De novo interstitial deletion 2q14.1q22.1: Is there a recognizable phenotype?. American Journal of Medical Genetics, Part A, 2014, 164, 3194-3202.	0.7	9
609	Autism spectrum disorder: advances in evidence-based practice. Cmaj, 2014, 186, 509-519.	0.9	158
610	De novo 393kb microdeletion of 7p11.2 characterized by aCGH in a boy with psychomotor retardation and dysmorphic features. Meta Gene, 2014, 2, 274-282.	0.3	6
611	Genetic diagnosis of autism spectrum disorders: The opportunity and challenge in the genomics era. Critical Reviews in Clinical Laboratory Sciences, 2014, 51, 249-262.	2.7	38
612	Development of a high-resolution Y-chromosome microarray for improved male infertility diagnosis. Fertility and Sterility, 2014, 101, 1079-1085.e3.	0.5	30
613	Etiological yield of SNP microarrays in idiopathic intellectual disability. European Journal of Paediatric Neurology, 2014, 18, 327-337.	0.7	26

#	Article	IF	Citations
614	Confirmation and further delineation of the 3q26.33â€“3q27.2 microdeletion syndrome. European Journal of Medical Genetics, 2014, 57, 76-80.	0.7	18
615	Phenotype profiling of patients with intellectual disability and copy number variations. European Journal of Paediatric Neurology, 2014, 18, 558-566.	0.7	18
616	Structural genomic variation in childhood epilepsies with complex phenotypes. European Journal of Human Genetics, 2014, 22, 896-901.	1.4	28
617	New candidate loci identified by arrayâ€CGH in a cohort of 100 children presenting with syndromic obesity. American Journal of Medical Genetics, Part A, 2014, 164, 1965-1975.	0.7	49
618	Diagnostic accuracy of the <scp>BAC</scp>sâ€onâ€Beads^{â„¢} assay versus karyotyping for prenatal detection of chromosomal abnormalities: a retrospective consecutive case series. BJOG: an International Journal of Obstetrics and Gynaecology, 2014, 121, 1245-1252.	1.1	26
619	Highâ€resolution chromosomal microarrays in prenatal diagnosis significantly increase diagnostic power. Prenatal Diagnosis, 2014, 34, 525-533.	1.1	42
620	Chromosomal microarray impacts clinical management. Clinical Genetics, 2014, 85, 147-153.	1.0	71
621	Human molecular cytogenetics: from cells to nucleotides. Genetics and Molecular Biology, 2014, 37, 194-209.	0.6	55
622	Analysis of chromosomal structural variation in patients with congenital leftâ€sided cardiac lesions. Birth Defects Research Part A: Clinical and Molecular Teratology, 2014, 100, 951-964.	1.6	12
623	Chromosome Microarray and Undiagnosed Seizures in a Pediatric Patient. Canadian Journal of Neurological Sciences, 2014, 41, 281-283.	0.3	0
624	Structural genomic variants in pediatric seizure disorders. Journal of Pediatric Epilepsy, 2015, 01, 161-169.	0.1	0
625	Genetic Advances in Intellectual Disability. Journal of Pediatric Genetics, 2015, 04, 125-127.	0.3	10
626	Risk assessment models in genetics clinic for array comparative genomic hybridization: Clinical information can be used to predict the likelihood of an abnormal result in patients. Journal of Pediatric Genetics, 2015, 02, 025-031.	0.3	2
627	Copy number variant analysis from exome data in 349 patients with epileptic encephalopathy. Annals of Neurology, 2015, 78, 323-328.	2.8	59
628	A 6q14.1â€q15 microdeletion in a male patient with severe autistic disorder, lack of oral language, and dysmorphic features with concomitant presence of a maternally inherited Xp22.31 copy number gain. Clinical Case Reports (discontinued), 2015, 3, 415-423.	0.2	4
629	The 22q11.2 Deletion Syndrome. , 0, , 100-111.		1
630	Clinical and Molecular Cytogenetic Characterisation of Children with Developmental Delay and Dysmorphic Features / KliniÄna in Molekularna Citogenetska Obravnava Otrok Z Razvojnim Zaostankom in DisplastiÄnimi Znaki. Zdravstveno Varstvo, 2015, 54, 69-73.	0.6	0
631	Williams-Beuren Syndrome: Experience of 43 Patients and a Report of an Atypical Case from a Tertiary Care Center in India. Cytogenetic and Genome Research, 2015, 146, 187-194.	0.6	8

#	Article	IF	Citations
632	Array Comparative Genomic Hybridization (Array CGH) for Detection of Genomic Copy Number Variants. Journal of Visualized Experiments, 2015, , e51718.	0.2	4
633	Endocrinopathies in a boy with cryptic copy-number variations on 4q, 7q and Xp. Human Genome Variation, 2015, 2, 15020.	0.4	0
634	WspÃ³Å‚praca genetyka klinicznego i biologa molekularnego â€“ wczoraj i dziÅ›. Pediatria Polska, 2015, 90, 171-180.	0.1	0
635	Copy number loss upstream of RAI1 uncovers gene expression regulatory region that may impact Potockiâ€“Lupski syndrome diagnosis. Molecular Cytogenetics, 2015, 8, 75.	0.4	4
636	Next-generation sequencing using a pre-designed gene panel for the molecular diagnosis of congenital disorders in pediatric patients. Human Genomics, 2015, 9, 33.	1.4	26
637	Contribution of copy number variants (CNVs) to congenital, unexplained intellectual and developmental disabilities in Lebanese patients. Molecular Cytogenetics, 2015, 8, 26.	0.4	5
638	Routine use of array comparative genomic hybridization (aCGH) as standard approach for prenatal diagnosis of chromosomal abnormalities. Clinical experience of 1763 prenatal cases. Prenatal Diagnosis, 2015, 35, 1269-1277.	1.1	27
639	Clinical characterization of a male patient with the recently described 8q21.11 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1369-1373.	0.7	6
640	Deletion of 15q11.2(BP1â€BP2) region: Further evidence for lack of phenotypic specificity in a pediatric population. American Journal of Medical Genetics, Part A, 2015, 167, 2098-2102.	0.7	33
641	A Novel 1.0 Mb Duplication of Chromosome 8p22-21.3 in a Patient With Autism Spectrum Disorder. Child Neurology Open, 2015, 2, 1-6.	0.5	6
642	At the cross-roads of participatory research and biomarker discovery in autism: the need for empirical data. BMC Medical Ethics, 2015, 16, 88.	1.0	14
643	3p22.1p21.31 microdeletion identifies CCK as Asperger syndrome candidate gene and shows the way for therapeutic strategies in chromosome imbalances. Molecular Cytogenetics, 2015, 8, 82.	0.4	27
644	Copy number variants prioritization after array-CGH analysis â€“ a cohort of 1000 patients. Molecular Cytogenetics, 2015, 8, 103.	0.4	17
645	De Novo interstitial deletion 13q33.3q34 in a male patient with double outlet right ventricle, microcephaly, dysmorphic craniofacial findings, and motor and developmental delay. American Journal of Medical Genetics, Part A, 2015, 167, 1134-1141.	0.7	18
646	The national birth defects prevention study: A review of the methods. Birth Defects Research Part A: Clinical and Molecular Teratology, 2015, 103, 656-669.	1.6	188
647	Mothers' appreciation of chromosomal microarray analysis for autism spectrum disorder. Journal for Specialists in Pediatric Nursing, 2015, 20, 244-258.	0.6	17
648	5p deletions: Current knowledge and future directions. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2015, 169, 224-238.	0.7	67
649	A novel maternally inherited 8q24.3 and a rare paternally inherited 14q23.3 CNVs in a family with neurodevelopmental disorders. American Journal of Medical Genetics, Part A, 2015, 167, 1921-1926.	0.7	18

#	Article	IF	Citations
650	Potockiâ€“Lupski syndrome in conjunction with bilateral clubfoot. Journal of Pediatric Orthopaedics Part B, 2015, 24, 373-376.	0.3	6
651	Comparing genetic counselor's and patient's perceptions of needs in prenatal chromosomal microarray testing. Prenatal Diagnosis, 2015, 35, 870-878.	1.1	38
652	Low Rates of Genetic Testing in Children With Developmental Delays, Intellectual Disability, and Autism Spectrum Disorders. Global Pediatric Health, 2015, 2, 2333794X1562371.	0.3	12
653	First Korean Case of SATB2-Associated 2q32-q33 Microdeletion Syndrome. Annals of Laboratory Medicine, 2015, 35, 275-278.	1.2	6
654	Genomics in medicine: From promise to practice. South African Medical Journal, 2015, 105, 545.	0.2	5
655	Cerebral Malformations. , 2015, , 137-150.		2
656	Routine Chromosomal Microarray Analysis is Necessary in Korean Patients With Unexplained Developmental Delay/Mental Retardation/Autism Spectrum Disorder. Annals of Laboratory Medicine, 2015, 35, 510-518.	1.2	16
657	Rationale for the Cytogenomics of Cardiovascular Malformations Consortium: A Phenotype Intensive Registry Based Approach. Journal of Cardiovascular Development and Disease, 2015, 2, 76-92.	0.8	7
658	Clinical Impact and Cost-Effectiveness of Whole Exome Sequencing as a Diagnostic Tool: A Pediatric Centerâ€™s Experience. Frontiers in Pediatrics, 2015, 3, 67.	0.9	159
659	Prenatal Diagnosis of Central Nervous System Anomalies by High-Resolution Chromosomal Microarray Analysis. BioMed Research International, 2015, 2015, 1-9.	0.9	35
660	Precision Medicine for Continuing Phenotype Expansion of Human Genetic Diseases. BioMed Research International, 2015, 2015, 1-4.	0.9	17
662	Genetic testing in patients with global developmental delay/intellectual disabilities. A review. Medicine and Pharmacy Reports, 2015, 88, 288-292.	0.2	17
663	Integration of microarray analysis into the clinical diagnosis of hematological malignancies: How much can we improve cytogenetic testing?. Oncotarget, 2015, 6, 18845-18862.	0.8	23
664	Discovery of a novel cystathionine-beta-synthase mutation and diagnosis of homocystinuria by whole exome sequencing in a family from rural Honduras. Case Reports in Clinical Pathology, 2015, 2, .	0.0	0
665	Array comparative genomic hybridisation testing in CHD. Cardiology in the Young, 2015, 25, 1155-1172.	0.4	7
666	Detection and correction of artefacts in estimation of rare copy number variants and analysis of rare deletions in type 1 diabetes. Human Molecular Genetics, 2015, 24, 1774-1790.	1.4	20
667	Identifying Human Genome-Wide CNV, LOH and UPD by Targeted Sequencing of Selected Regions. PLoS ONE, 2015, 10, e0123081.	1.1	29
669	ClinGen â€” The Clinical Genome Resource. New England Journal of Medicine, 2015, 372, 2235-2242.	13.9	1,016

#	Article	IF	Citations
670	Cost Effectiveness of Using Array-CGH for Diagnosing Learning Disability. Applied Health Economics and Health Policy, 2015, 13, 421-432.	1.0	14
671	Gamut of Genetic Testing for Neonatal Care. Clinics in Perinatology, 2015, 42, 217-226.	0.8	6
672	Array-based DNA methylation analysis in individuals with developmental delay/intellectual disability and normal molecular karyotype. European Journal of Medical Genetics, 2015, 58, 419-425.	0.7	26
673	Evaluation and Diagnosis of the Dysmorphic Infant. Clinics in Perinatology, 2015, 42, 243-261.	0.8	27
674	A French Approach to Test Fetuses with Ultrasound Abnormalities Using a Customized Microarray as First-Tier Genetic Test. Cytogenetic and Genome Research, 2015, 147, 103-110.	0.6	17
675	Distal 10q trisomy with copy number gain in chromosome region 10q23.1â€“10q25.1: the Wnt signaling pathway is the most pertinent to the gene content in the region of copy number gain: a case report. BMC Research Notes, 2015, 8, 250.	0.6	5
676	The Diagnostic Utility of Single Long Contiguous Stretches of Homozygosity in Patients without Parental Consanguinity. Molecular Syndromology, 2015, 6, 135-140.	0.3	11
677	Sociology in an Age of Genomic Instability: Copy Number Variation, Somatic Mosaicism, and the Fallen Genome. Advances in Medical Sociology, 2015, , 157-186.	0.1	12
679	Array-Comparative Genomic Hybridization Analysis in Fetuses with Major Congenital Malformations Reveals that 24% of Cases Have Pathogenic Deletions/Duplications. Cytogenetic and Genome Research, 2015, 147, 10-16.	0.6	4
680	Mechanisms of Origin, Phenotypic Effects and Diagnostic Implications of Complex Chromosome Rearrangements. Molecular Syndromology, 2015, 6, 110-134.	0.3	73
681	22q11.21 Deletion Syndromes: A Review of Proximal, Central, and Distal Deletions and Their Associated Features. Cytogenetic and Genome Research, 2015, 146, 89-99.	0.6	1,385
682	Evaluation of the Affymetrix CytoScan^{Â®}Dx Assay for developmental delay. Expert Review of Molecular Diagnostics, 2015, 15, 185-192.	1.5	9
683	Capturing the clinical utility of genomic testing: medical recommendations following pediatric microarray. European Journal of Human Genetics, 2015, 23, 1135-1141.	1.4	17
685	Accelerating Novel Candidate Gene Discovery in Neurogenetic Disorders via Whole-Exome Sequencing of Prescreened Multiplex Consanguineous Families. Cell Reports, 2015, 10, 148-161.	2.9	375
686	A genome-wide screen for copy number alterations in an adolescent pilot cohort with mÃ¼llerian anomalies. Fertility and Sterility, 2015, 103, 487-493.	0.5	8
687	Mosaic structural variation in children with developmental disorders. Human Molecular Genetics, 2015, 24, 2733-2745.	1.4	54
688	Assessing the utility of confirmatory studies following identification of large-scale genomic imbalances by microarray. Genetics in Medicine, 2015, 17, 875-879.	1.1	7
689	Current controversies in prenatal diagnosis 3: the ethical and counseling implications of new genomic technologies: all pregnant women should be offered prenatal diagnostic genome-wide testing for prenatally identified fetal congenital anomalies. Prenatal Diagnosis, 2015, 35, 19-22.	1.1	8

#	Article	IF	Citations
690	Genetic Evaluation of Children with Global Developmental Delayâ€"Current Status of Network Systems in Taiwan. Pediatrics and Neonatology, 2015, 56, 213-219.	0.3	2
691	Molecular Cytogenetic Analysis of Telomere Rearrangements. Current Protocols in Human Genetics, 2015, 84, 8.11.1-8.11.15.	3.5	3
692	A copy number variation map of the human genome. Nature Reviews Genetics, 2015, 16, 172-183.	7.7	707
693	A high-resolution copy-number variation resource for clinical and population genetics. Genetics in Medicine, 2015, 17, 747-752.	1.1	73
694	Reference Databases for Disease Associations. , 2015, , 191-216.		0
695	Inherited 15q24 microdeletion syndrome in twins and their father with phenotypic variability. European Journal of Medical Genetics, 2015, 58, 111-115.	0.7	12
696	Wholeâ€genome array as a firstâ€line cytogenetic test in prenatal diagnosis. Ultrasound in Obstetrics and Gynecology, 2015, 45, 363-372.	0.9	26
697	Identical deletion at 14q13.3 including PAX9 and NKX2-1 in siblings from mosaicism of unaffected parent. Journal of Human Genetics, 2015, 60, 203-206.	1.1	13
698	The phenotype spectrum of X-linked ichthyosis identified by chromosomal microarray. Journal of the American Academy of Dermatology, 2015, 72, 617-627.	0.6	29
699	Diagnostic Yield of Chromosomal Microarray Analysis in a Cohort of Patients with Autism Spectrum Disorders from a Highly Consanguineous Population. Journal of Autism and Developmental Disorders, 2015, 45, 2323-2328.	1.7	8
700	Ethical signposts for clinical geneticists in secondary variant and incidental finding disclosure discussions. Medicine, Health Care and Philosophy, 2015, 18, 361-370.	0.9	9
701	Multidisciplinary Assessment and Treatment of Self-Injurious Behavior in Autism Spectrum Disorder and Intellectual Disability: Integration of Psychological and Biological Theory and Approach. Journal of Autism and Developmental Disorders, 2015, 45, 1541-1568.	1.7	66
702	Paired-Duplication Signatures Mark Cryptic Inversions and Other Complex Structural Variation. American Journal of Human Genetics, 2015, 97, 170-176.	2.6	45
703	High diagnostic yield of clinical exome sequencing in Middle Eastern patients with Mendelian disorders. Human Genetics, 2015, 134, 967-980.	1.8	168
704	DiagnÃ³stico de las anomalÃas cromosÃ³micas en patologÃa constitucional. EMC - Tratado De Medicina, 2015, 19, 1-8.	0.0	0
705	Brain malformations in a patient with deletion 2p16.1: A refinement ofÂthe phenotype to BCL11A. European Journal of Medical Genetics, 2015, 58, 351-354.	0.7	24
706	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
707	Strategy of probe selection for studying mRNAs that participate in receptor-mediated apoptosis signaling. Molecular Biology, 2015, 49, 457-465.	0.4	2

#	Article	IF	Citations
708	Mosaicism in Traditional Mendelian Diseases. Current Genetic Medicine Reports, 2015, 3, 101-109.	1.9	2
709	The Clinical Utility of a Single-Nucleotide Polymorphism Microarray in Patients With Epilepsy at a Tertiary Medical Center. Journal of Child Neurology, 2015, 30, 1770-1777.	0.7	8
710	The value of microarray-based comparative genomic hybridisation (aCGH) testing in the paediatric clinic. Archives of Disease in Childhood, 2015, 100, 728-731.	1.0	4
711	Genetic architecture of colorectal cancer. Gut, 2015, 64, 1623-1636.	6.1	152
712	Human Structural Variation: Mechanisms of Chromosome Rearrangements. Trends in Genetics, 2015, 31, 587-599.	2.9	192
714	Exome sequencing and whole genome sequencing for the detection of copy number variation. Expert Review of Molecular Diagnostics, 2015, 15, 1023-1032.	1.5	73
716	Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents. American Journal of Human Genetics, 2015, 97, 6-21.	2.6	453
717	Discovery of Rare Mutations in Autism: Elucidating Neurodevelopmental Mechanisms. Neurotherapeutics, 2015, 12, 553-571.	2.1	21
718	Copy Number Variants, Aneuploidies, and Human Disease. Clinics in Perinatology, 2015, 42, 227-242.	0.8	51
719	The clustering of functionally related genes contributes to CNV-mediated disease. Genome Research, 2015, 25, 802-813.	2.4	31
720	Lymphovascular invasion and histologic grade are associated with specific genomic profiles in invasive carcinomas of the breast. Tumor Biology, 2015, 36, 1835-1848.	0.8	21
721	Phelanâ€“McDermid Syndrome and SHANK3: Implications for Treatment. Neurotherapeutics, 2015, 12, 620-630.	2.1	83
722	Genetics of Neuromuscular Disorders. , 2015, , 17-31.		1
723	Analyzing the most frequent disease loci in targeted patient categories optimizes disease gene identification and test accuracy worldwide. Journal of Translational Medicine, 2015, 13, 16.	1.8	6
724	Functional consequences of copy number variants in miscarriage. Molecular Cytogenetics, 2015, 8, 6.	0.4	17
725	p600/UBR4 in the central nervous system. Cellular and Molecular Life Sciences, 2015, 72, 1149-1160.	2.4	63
726	Connecting the CNTNAP2 Networks with Neurodevelopmental Disorders. Molecular Syndromology, 2015, 6, 7-22.	0.3	89
727	Gene discovery and functional assessment of rare copy-number variants in neurodevelopmental disorders. Briefings in Functional Genomics, 2015, 14, 315-328.	1.3	24

#	Article	IF	Citations
728	Metabolic and Genetic Causes of Autism. , 2015, , 209-217.		0
729	Array CGH as a first-tier test for neonates with congenital heart disease. Cardiology in the Young, 2015, 25, 115-122.	0.4	17
730	Variants of unknown significance on chromosomal microarray analysis: parental perspectives. Journal of Community Genetics, 2015, 6, 343-349.	0.5	28
731	Navigating the current landscape of clinical genetic testing for inherited retinal dystrophies. Genetics in Medicine, 2015, 17, 245-252.	1.1	54
732	Spectrum of Neurodevelopmental Disabilities. Journal of Child Neurology, 2015, 30, 344-356.	0.7	7
733	Genomic imbalances in pediatric patients with chronic kidney disease. Journal of Clinical Investigation, 2015, 125, 2171-2178.	3.9	68
735	Genomic Variants and Variations in Malformations of Cortical Development. Pediatric Clinics of North America, 2015, 62, 571-585.	0.9	32
736	PIAS4 is associated with macro/microcephaly in the novel interstitial 19p13.3 microdeletion/microduplication syndrome. European Journal of Human Genetics, 2015, 23, 1615-1626.	1.4	29
737	Autism spectrum disorder and epilepsy: Disorders with a shared biology. Epilepsy and Behavior, 2015, 47, 191-201.	0.9	129
738	Detection of Chromosomal Aberrations in Clinical Practice: From Karyotype to Genome Sequence. Annual Review of Genomics and Human Genetics, 2015, 16, 309-326.	2.5	52
739	A 5-Month-Old Boy with Delay in Growth and Development and Decreased Muscle Tone. Clinical Chemistry, 2015, 61, 50-54.	1.5	0
740	An efficient screening method for simultaneous detection of recurrent copy number variants associated with psychiatric disorders. Clinica Chimica Acta, 2015, 445, 34-40.	0.5	7
741	Genetics in child and adolescent psychiatry: methodological advances and conceptual issues. European Child and Adolescent Psychiatry, 2015, 24, 619-634.	2.8	9
742	Retrospective analysis supports algorithm as efficient diagnostic approach to treatable intellectual developmental disabilities. Molecular Genetics and Metabolism, 2015, 115, 1-9.	0.5	13
743	The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. Genetics in Medicine, 2015, 17, 719-725.	1.1	19
744	22q11.2 Deletion Syndrome: Laboratory Diagnosis and TBX1 and FGF8 Mutation Screening. Journal of Pediatric Genetics, 2015, 04, 017-022.	0.3	9
745	Parentsâ€™ Perceptions of the Usefulness of Chromosomal Microarray Analysis for Children with Autism Spectrum Disorders. Journal of Autism and Developmental Disorders, 2015, 45, 3262-3275.	1.7	67
746	Current Tools for Interpretation of Genomic Copy Number Variants. Current Genetic Medicine Reports, 2015, 3, 202-208.	1.9	0

#	Article	IF	Citations
747	Child with a Heterozygous Deletion of 2 Megabases that Includes the DAG1 Gene, Presenting as Developmental Delay. Journal of Pediatric Neurology, 2015, 13, 116-120.	0.0	1
748	Parental expression is overvalued in the interpretation of rare inherited variants. European Journal of Human Genetics, 2015, 23, 4-7.	1.4	8
749	Rapid Diagnosis of Imprinting Disorders Involving Copy Number Variation and Uniparental Disomy Using Genome-Wide SNP Microarrays. Cytogenetic and Genome Research, 2015, 146, 9-18.	0.6	14
750	Precision medicine is here, break out your wallet. Applied & Translational Genomics, 2015, 4, 14-15.	2.1	0
751	Intragenic <scp>CAMTA1</scp> deletions are associated with a spectrum of neurobehavioral phenotypes. Clinical Genetics, 2015, 87, 478-482.	1.0	18
752	Investigating the genetic basis of feverâ€associated syndromic epilepsies using copy number variation analysis. Epilepsia, 2015, 56, e26-32.	2.6	16
753	Future of whole genome sequencing. Journal of Paediatrics and Child Health, 2015, 51, 251-254.	0.4	10
754	Clinically relevant copy number variations detected in cerebral palsy. Nature Communications, 2015, 6, 7949.	5.8	120
755	Chromosomal microarrays testing in children with developmental disabilities and congenital anomalies. Jornal De Pediatria (VersÃ£o Em PortuguÃªs), 2015, 91, 189-195.	0.2	0
756	Building the foundation for genomics in precision medicine. Nature, 2015, 526, 336-342.	13.7	376
758	Chromosomal microarray in unexplained severe early onset epilepsy â€“ A single centre cohort. European Journal of Paediatric Neurology, 2015, 19, 390-394.	0.7	14
760	Complex Autism Spectrum Disorders and Cutting-Edge Molecular Diagnostic Tests. JAMA - Journal of the American Medical Association, 2015, 314, 879.	3.8	16
761	Molecular Diagnostic Yield of Chromosomal Microarray Analysis and Whole-Exome Sequencing in Children With Autism Spectrum Disorder. JAMA - Journal of the American Medical Association, 2015, 314, 895.	3.8	352
762	A multicenter, cross-platform clinical validation study of cancer cytogenomic arrays. Cancer Genetics, 2015, 208, 525-536.	0.2	12
763	Chromosomal microarrays testing in children with developmental disabilities and congenital anomalies. Jornal De Pediatria, 2015, 91, 189-195.	0.9	11
766	Prenatal diagnosis of complete maternal uniparental isodisomy of chromosome 4 in a fetus without congenital abnormality or inherited disease-associated variations. Molecular Cytogenetics, 2015, 8, 85.	0.4	5
767	Noninvasive detection of fetal subchromosomal abnormalities by semiconductor sequencing of maternal plasma DNA. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 14670-14675.	3.3	104
768	Asymmetric Aneuploidy in Mesenchymal Stromal Cells Detected by In Situ Karyotyping and Fluorescence In Situ Hybridization: Suggestions for Reference Values for Stem Cells. Stem Cells and Development, 2015, 24, 77-92.	1.1	17

#	Article	IF	Citations
769	Recent advances of genomic testing in perinatal medicine. Seminars in Perinatology, 2015, 39, 44-54.	1.1	31
770	FISH for 22q11.2 Deletion Not Cost-Effective for Infants with Congenital Heart Disease with Microarray. Pediatric Cardiology, 2015, 36, 531-536.	0.6	12
771	A new syndrome of intellectual disability with dysmorphism due to TBL1XR1 deletion. American Journal of Medical Genetics, Part A, 2015, 167, 164-168.	0.7	37
772	Circulating Nucleic Acids in Early Diagnosis, Prognosis and Treatment Monitoring. Advances in Predictive, Preventive and Personalised Medicine, 2015, , .	0.6	8
773	Noncardiac DiGeorge syndrome diagnosed with multiplex ligation-dependent probe amplification: A case report. Journal of the Formosan Medical Association, 2015, 114, 769-773.	0.8	5
774	Assessing copy number from exome sequencing and exome array CGH based on CNV spectrum in a large clinical cohort. Genetics in Medicine, 2015, 17, 623-629.	1.1	106
775	Karyotype versus genomic hybridization for the prenatal diagnosis of chromosomal abnormalities: a metaanalysis. American Journal of Obstetrics and Gynecology, 2015, 212, 330.e1-330.e10.	0.7	34
776	Constitutional Disorders. , 2015, , 271-296.		0
777	Array-CGH in children with mild intellectual disability: a population-based study. European Journal of Pediatrics, 2015, 174, 75-83.	1.3	16
778	How to useâ€¦ microarray comparative genomic hybridisation to investigate developmental disorders. Archives of Disease in Childhood: Education and Practice Edition, 2015, 100, 24-29.	0.3	23
779	Delineating the 15q13.3 microdeletion phenotype: a case series and comprehensive review of the literature. Genetics in Medicine, 2015, 17, 149-157.	1.1	103
780	Preferences for results from genomic microarrays: comparing parents and health care providers. Clinical Genetics, 2015, 87, 21-29.	1.0	18
781	Detecting somatic mosaicism: considerations and clinical implications. Clinical Genetics, 2015, 87, 554-562.	1.0	40
782	Genomic Applications in Pathology. , 2015, , .		1
783	Genomewide Array Comparative Genomic Hybridization in 55 JapaneseNormokaryotypic Patients with Non-Syndromic Intellectual Disability. Journal of Pediatric Neurological Disorders, 2016, 02, .	0.0	1
784	Genetics of Parkinsonâ€™s Disease: The Role of Copy Number Variations. , 2016, , .		2
785	Cytogenetics at the University of Cape Town: A 45-year journey. South African Medical Journal, 2016, 106, 29.	0.2	0
786	Chromosomal Microarray Genetic Testing â€“ A Qualitative Investigation among Parents of Children with Autism Spectrum Disorders in Rural North Carolina. Journal of Clinical & Medical Genomics, 2016, 04, .	0.1	1

#	Article	IF	Citations
787	Utilization of genetic testing among children with developmental disabilities in the United States. The Application of Clinical Genetics, 2016, Volume 9, 93-100.	1.4	26
788	Recent advances in prenatal genetic screening and testing. F1000Research, 2016, 5, 2591.	0.8	51
789	Experimental Tools for the Identification ofÂSpecific Genes in Autism Spectrum DisordersÂand Intellectual Disability. , 2016, , 3-12.		1
790	What Do Parents Think about Chromosomal Microarray Testing? A Qualitative Report from Parents of Children with Autism Spectrum Disorders. Autism Research & Treatment, 2016, 2016, 1-9.	0.1	9
791	Clinical Performance of an Ultrahigh Resolution Chromosomal Microarray Optimized for Neurodevelopmental Disorders. BioMed Research International, 2016, 2016, 1-7.	0.9	16
792	SNP Microarray in FISH Negative Clinically Suspected 22q11.2 Microdeletion Syndrome. Scientifica, 2016, 2016, 1-18.	0.6	6
793	Retrospective karyotype study in mentally retarded patients. Revista Da AssociaÃ§Ã£o MÃ©dica Brasileira, 2016, 62, 262-268.	0.3	3
794	Analysis of chromosomal abnormalities by CGH-array in patients with dysmorphic and intellectual disability with normal karyotype. Einstein (Sao Paulo, Brazil), 2016, 14, 30-34.	0.3	9
795	Fluorescence In situ Hybridization: Cell-Based Genetic Diagnostic and Research Applications. Frontiers in Cell and Developmental Biology, 2016, 4, 89.	1.8	150
796	Genetic Evaluation and Use of Chromosome Microarray in Patients with Isolated Heart Defects: Benefits and Challenges of a New Model in Cardiovascular Care. Frontiers in Cardiovascular Medicine, 2016, 3, 19.	1.1	12
797	The Current Landscape of Genetic Testing in Cardiovascular Malformations: Opportunities and Challenges. Frontiers in Cardiovascular Medicine, 2016, 3, 22.	1.1	19
798	Chromosomal Microarray Analysis of Consecutive Individuals with Autism Spectrum Disorders Using an Ultra-High Resolution Chromosomal Microarray Optimized for Neurodevelopmental Disorders. International Journal of Molecular Sciences, 2016, 17, 2070.	1.8	56
799	Large Genomic Imbalances in Brugada Syndrome. PLoS ONE, 2016, 11, e0163514.	1.1	23
800	Clinical Utility of a Comprehensive, Whole Genome CMA Testing Platform in Pediatrics: A Prospective Randomized Controlled Trial of Simulated Patients in Physician Practices. PLoS ONE, 2016, 11, e0169064.	1.1	5
801	Array-CGH analysis in patients with intellectual disability and/or congenital malformations in Brazil. Genetics and Molecular Research, 2016, 15, .	0.3	10
802	Genetic Counseling for a Prenatal Diagnosis of Structural Chromosomal Abnormality with High-Resolution Analysis Using a Single Nucleotide Polymorphism Microarray. Clinics and Practice, 2016, 6, 55-56.	0.6	1
803	A novel de novo microdeletion at 17q11.2 adjacent to NF1 gene associated with developmental delay, short stature, microcephaly and dysmorphic features. Molecular Cytogenetics, 2016, 9, 41.	0.4	15
804	Copy number variation in ALOX5 and PTGER1 is associated with NSAIDs-induced urticaria and/or angioedema. Pharmacogenetics and Genomics, 2016, 26, 280-287.	0.7	15

#	Article	IF	Citations
805	Detecting Copy Number Variation via Next Generation Technology. Current Genetic Medicine Reports, 2016, 4, 74-85.	1.9	20
806	Clinical exome sequencing in neurogenetic and neuropsychiatric disorders. Annals of the New York Academy of Sciences, 2016, 1366, 49-60.	1.8	23
807	What results to disclose, when, and who decides? Healthcare professionals' views on prenatal chromosomal microarray analysis. Prenatal Diagnosis, 2016, 36, 252-259.	1.1	28
808	DICER1 deletion and 14q32 microdeletion syndrome. Clinical Dysmorphology, 2016, 25, 37-40.	0.1	6
809	Is one diagnosis the whole story? patients with double diagnoses. American Journal of Medical Genetics, Part A, 2016, 170, 2338-2348.	0.7	20
810	Autism and chromosome abnormalitiesâ€"A review. Clinical Anatomy, 2016, 29, 620-627.	1.5	12
811	Novel copy number variants and major limb reduction malformation: Report of three cases. American Journal of Medical Genetics, Part A, 2016, 170, 1245-1250.	0.7	8
812	Detection of 1p36 deletion by clinical exome-first diagnostic approach. Human Genome Variation, 2016, 3, 16006.	0.4	20
813	Clinical utility of array comparative genomic hybridisation in prenatal setting. BMC Medical Genetics, 2016, 17, 81.	2.1	16
814	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	1.7	295
815	Diagnostic yield of array CGH in patients with autism spectrum disorder in Hong Kong. Clinical and Translational Medicine, 2016, 5, 18.	1.7	11
816	Chromosome 10q26 deletion syndrome: Two new cases and a review of the literature. Molecular Medicine Reports, 2016, 14, 5134-5140.	1.1	17
817	Assessment of copy number variations in 120 patients with Poland syndrome. BMC Medical Genetics, 2016, 17, 89.	2.1	20
818	Challenges in detecting genomic copy number aberrations using next-generation sequencing data and the eXome Hidden Markov Model: a clinical exome-first diagnostic approach. Human Genome Variation, 2016, 3, 16025.	0.4	38
819	Mutations inHECW2are associated with intellectual disability and epilepsy. Journal of Medical Genetics, 2016, 53, 697-704.	1.5	55
820	CNV analysis in Chinese children of mental retardation highlights a sex differentiation in parental contribution to de novo and inherited mutational burdens. Scientific Reports, 2016, 6, 25954.	1.6	19
821	The importance of copy number variation in congenital heart disease. Npj Genomic Medicine, 2016, 1, 16031.	1.7	62
822	Prenatal diagnosis of fetal multicystic dysplastic kidney via high-resolution whole-genome array. Nephrology Dialysis Transplantation, 2016, 31, 1693-1698.	0.4	31

#	Article	IF	Citations
824	Novel bioinformatic developments for exome sequencing. Human Genetics, 2016, 135, 603-614.	1.8	37
825	Chromosomal microarray in prenatal diagnosis: case studies and clinical challenges. Personalized Medicine, 2016, 13, 249-255.	0.8	4
826	A homozygous truncating mutation in PUS3 expands the role of tRNA modification in normal cognition. Human Genetics, 2016, 135, 707-713.	1.8	112
827	A null mutation in TNIK defines a novel locus for intellectual disability. Human Genetics, 2016, 135, 773-778.	1.8	23
828	DNA copy number variants: A potentially useful predictor of early onset renal failure in boys with posterior urethral valves. Journal of Pediatric Urology, 2016, 12, 227.e1-227.e7.	0.6	17
829	Chromosomal microarray analysis in clinical evaluation of neurodevelopmental disorders-reporting a novel deletion of SETDB1 and illustration of counseling challenge. Pediatric Research, 2016, 80, 371-381.	1.1	16
830	What Is New in Genetics and Genomics?. , 2016, , 703-712.		0
831	A Clinicianâ€™s perspective on clinical exome sequencing. Human Genetics, 2016, 135, 643-654.	1.8	33
832	Noninvasive prenatal screening or advanced diagnostic testing: caveat emptor. American Journal of Obstetrics and Gynecology, 2016, 215, 298-305.	0.7	82
833	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	1.1	125
835	Idiopathic focal epilepsies: the â€œlost tribeâ€• Epileptic Disorders, 2016, 18, 252-288.	0.7	65
836	An 8.4â€Mb 3q26.33â€3q28 microdeletion in a patient with blepharophimosisâ€“intellectual disability syndrome and a review of the literature. Clinical Case Reports (discontinued), 2016, 4, 824-830.	0.2	5
837	Chromosome microarray proficiency testing and analysis of quality metric data trends through an external quality assessment program for Australasian laboratories. Pathology, 2016, 48, 586-596.	0.3	0
838	Copy number variations in 375 patients with oesophageal atresia and/or tracheoesophageal fistula. European Journal of Human Genetics, 2016, 24, 1715-1723.	1.4	27
839	Validation of a Chromosomal Microarray for Prenatal Diagnosis Using a Prospective Cohort of Pregnancies with Increased Risk for Chromosome Abnormalities. Genetic Testing and Molecular Biomarkers, 2016, 20, 791-798.	0.3	2
841	The use of chromosomal microarray forÂprenatalÂdiagnosis. American Journal of Obstetrics and Gynecology, 2016, 215, B2-B9.	0.7	131
842	Inherited ichthyosis: Nonâ€syndromic forms. Journal of Dermatology, 2016, 43, 242-251.	0.6	147
843	Tentative clinical diagnosis of Lujanâ€Fryns syndromeâ€”A conglomeration of different genetic entities?. American Journal of Medical Genetics, Part A, 2016, 170, 94-102.	0.7	11

#	Article	IF	Citations
844	Genetic Counseling for Autism Spectrum Disorder in an Evolving Theoretical Landscape. Current Genetic Medicine Reports, 2016, 4, 147-153.	1.9	11
845	Preclinical development and qualification of ZFN-mediated CCR5 disruption in human hematopoietic stem/progenitor cells. Molecular Therapy - Methods and Clinical Development, 2016, 3, 16067.	1.8	91
846	Genetic Approach to Diagnosis of Intellectual Disability. Indian Journal of Pediatrics, 2016, 83, 1141-1149.	0.3	8
847	Interstitial Chromosome 3p14.1 Deletion due to a Maternal Insertion: Phenotype and Association with Balanced Parental Rearrangement. Molecular Syndromology, 2016, 7, 43-48.	0.3	3
848	X-Linked Candidate Genes for a Ciliopathy-Like Disorder. Molecular Syndromology, 2016, 7, 37-42.	0.3	1
851	Somatic Mosaicism and Neurological Diseases. , 2016, , 179-199.		7
852	Neurodevelopmental Disorders, Causes, and Consequences. , 2016, , 587-599.		1
853	3Disease Browser: A Web server for integrating 3D genome and disease-associated chromosome rearrangement data. Scientific Reports, 2016, 6, 34651.	1.6	32
854	Comprehensive computational analysis of chromosome 11. , 2016, , .		0
855	Genetics of Combined Pituitary Hormone Deficiency: Roadmap into the Genome Era. Endocrine Reviews, 2016, 37, 636-675.	8.9	147
856	Subtelomeric Copy Number Variations: The Importance of 4p/4q Deletions in Patients with Congenital Anomalies and Developmental Disability. Cytogenetic and Genome Research, 2016, 149, 241-246.	0.6	6
857	High-resolution SNP genotyping platform identified recurrent and novel CNVs in autism multiplex families. Neuroscience, 2016, 339, 561-570.	1.1	17
858	Causal variants screened by whole exome sequencing in a patient with maternal uniparental isodisomy of chromosome 10 and a complicated phenotype. Experimental and Therapeutic Medicine, 2016, 11, 2247-2253.	0.8	7
859	â€œIt wasn't a disaster or anythingâ€: Parentsâ€™ experiences of their child's uncertain chromosomal microarray result. American Journal of Medical Genetics, Part A, 2016, 170, 2895-2904.	0.7	24
860	Associated anomalies in cases with anotia and microtia. European Journal of Medical Genetics, 2016, 59, 607-614.	0.7	22
861	Indexing Effects of Copy Number Variation on Genes Involved in Developmental Delay. Scientific Reports, 2016, 6, 28663.	1.6	35
862	RYR2, PTDSS1 and AREG genes are implicated in a Lebanese population-based study of copy number variation in autism. Scientific Reports, 2016, 6, 19088.	1.6	27
863	Intragenic CNVs for epigenetic regulatory genes in intellectual disability: Survey identifies pathogenic and benign single exon changes. American Journal of Medical Genetics, Part A, 2016, 170, 2916-2926.	0.7	14

#	Article	IF	Citations
864	Testing the Complex Child: CGH Array, WES, Clinical Exome, WGS. Current Pediatrics Reports, 2016, 4, 155-163.	1.7	0
865	Importance of genetic testing in global health during the evaluation of familial microcephaly. Clinical Case Reports (discontinued), 2016, 4, 968-971.	0.2	0
866	Genetic Counseling in Developmental Disability. International Journal of Human Genetics, 2016, 16, 89-97.	0.1	1
867	Attenuated Phenotype in a Patient with Prader-Willi Syndrome and Duplication 16P11.2 Detected by Chromosomal Microarray. AACE Clinical Case Reports, 2016, 2, e329-e332.	0.4	0
868	Chromosomal microarray analysis as the first-tier test for the identification of pathogenic copy number variants in chromosome 9 pericentric regions and its challenge. Molecular Cytogenetics, 2016, 9, 64.	0.4	3
869	The genetic assessment of looked after children: common reasons for referral and recent advances. Archives of Disease in Childhood, 2016, 101, 581-584.	1.0	4
870	Chromosome Microarray. MCN the American Journal of Maternal Child Nursing, 2016, 41, 272-279.	0.3	1
871	Chromosomal abnormalities and copy number variations in fetal leftâ€sided congenital heart defects. Prenatal Diagnosis, 2016, 36, 177-185.	1.1	12
872	Chromosomal microarray use among women undergoing invasive prenatal diagnosis. Prenatal Diagnosis, 2016, 36, 656-661.	1.1	13
873	Whole exome sequencing is necessary to clarify ID/DD cases with de novo copy number variants of uncertain significance: Two proofâ€ofâ€concept examples. American Journal of Medical Genetics, Part A, 2016, 170, 1772-1779.	0.7	26
874	The prevalence of chromosomal aberrations associated with myelodysplastic syndromes in China. Annals of Hematology, 2016, 95, 1241-1248.	0.8	7
875	Genomic Sequencing Procedure Microcosting Analysis and Health Economic Cost-Impact Analysis. Journal of Molecular Diagnostics, 2016, 18, 319-328.	1.2	77
877	Chromosomal microarray analysis, or comparative genomic hybridization: A high throughput approach. MethodsX, 2016, 3, 8-18.	0.7	6
878	Coupleâ€™s Narratives of Communion and Isolation Following Abnormal Prenatal Microarray Testing Results. Qualitative Health Research, 2016, 26, 1975-1987.	1.0	44
879	Pathogenic copy number variants and SCN1A mutations in patients with intellectual disability and childhood-onset epilepsy. BMC Medical Genetics, 2016, 17, 34.	2.1	23
880	Overview: referrals for genetic evaluation from child psychiatrists. Child and Adolescent Psychiatry and Mental Health, 2016, 10, 7.	1.2	3
881	Chromosomal copy number analysis on chorionic villus samples from early spontaneous miscarriages by high throughput genetic technology. Molecular Cytogenetics, 2016, 9, 7.	0.4	39
882	Copy number variations in 119 Chinese children with idiopathic short stature identified by the custom genome-wide microarray. Molecular Cytogenetics, 2016, 9, 16.	0.4	12

#	Article	IF	Citations
883	POGZ truncating alleles cause syndromic intellectual disability. Genome Medicine, 2016, 8, 3.	3.6	78
884	Application of chromosomal microarrays in the evaluation of intellectual disability/global developmental delay patients â€“ A study from a tertiary care genetic centre in India. Gene, 2016, 590, 109-119.	1.0	11
885	New insights in the interpretation of array-CGH: autism spectrum disorder and positive family history for intellectual disability predict the detection of pathogenic variants. Italian Journal of Pediatrics, 2016, 42, 39.	1.0	21
887	Overview of Clinical Cytogenetics. Current Protocols in Human Genetics, 2016, 89, 8.1.1-8.1.13.	3.5	7
888	Genetic testing of aetiology of intellectual disability in a dedicated physical healthcare outpatient clinic for adults with intellectual disability. Internal Medicine Journal, 2016, 46, 177-185.	0.5	6
889	Second Pallisterâ€Opitz Genetics Symposium, Helena, Montana, July 2015. American Journal of Medical Genetics, Part A, 2016, 170, 1405-1421.	0.7	0
890	Homozygous single base deletion in TUSC3 causes intellectual disability with developmental delay in an Omani family. American Journal of Medical Genetics, Part A, 2016, 170, 1826-1831.	0.7	14
891	Obtaining a genetic diagnosis in a child with disability: impact on parental quality of life. Clinical Genetics, 2016, 89, 258-266.	1.0	50
892	Recurrent copy number variations as risk factors for autism spectrum disorders: analysis of the clinical implications. Clinical Genetics, 2016, 89, 708-718.	1.0	29
893	Parentsâ€™ Perspectives on Variants of Uncertain Significance from Chromosome Microarray Analysis. Journal of Genetic Counseling, 2016, 25, 101-111.	0.9	42
894	Rare copy number variations in an adult with transposition of the great arteries emphasize the importance of updated genetic assessments in syndromic congenital cardiac disease. International Journal of Cardiology, 2016, 203, 516-518.	0.8	10
895	Pre- and post-test genetic counseling for chromosomal and Mendelian disorders. Seminars in Perinatology, 2016, 40, 44-55.	1.1	34
896	The importance of de novo mutations for pediatric neurological diseaseâ€”It is not all in utero or birth trauma. Mutation Research - Reviews in Mutation Research, 2016, 767, 42-58.	2.4	7
897	Autism spectrum disorders: Integration of the genome, transcriptome and the environment. Journal of the Neurological Sciences, 2016, 364, 167-176.	0.3	49
898	Cytogenomic Evaluation of Children with Congenital Anomalies: Critical Implications for Diagnostic Testing and Genetic Counseling. Advances in Experimental Medicine and Biology, 2016, 912, 11-19.	0.8	1
899	Application of Array Comparative Genomic Hybridization in Newborns with Multiple Congenital Anomalies. Advances in Experimental Medicine and Biology, 2016, 912, 1-9.	0.8	6
900	Clinical exome sequencing in neurologic disease. Neurology: Clinical Practice, 2016, 6, 164-176.	0.8	56
901	Whole Exome Sequencing in Pediatric Neurology Patients. Journal of Child Neurology, 2016, 31, 887-894.	0.7	61

#	Article	IF	Citations
902	X-linked ichthyosis and Crigler-Najjar syndrome I: Coexistence in a male patient with two copy number variable regions of 2q37.1 and Xp22.3. Molecular Medicine Reports, 2016, 13, 1135-1140.	1.1	2
903	Comprehensive molecular testing in patients with high functioning autism spectrum disorder. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2016, 784-785, 46-52.	0.4	24
904	Spectrum of Cytogenomic Abnormalities Revealed by Array Comparative Genomic Hybridization on Products of Conception Culture Failure and Normal Karyotype Samples. Journal of Genetics and Genomics, 2016, 43, 121-131.	1.7	35
905	From Genetics to Genomics: A Short Introduction for Pediatric Neurologists. Neuropediatrics, 2016, 47, 005-011.	0.3	1
906	SNP array screening of cryptic genomic imbalances in 450 Japanese subjects with intellectual disability and multiple congenital anomalies previously negative for large rearrangements. Journal of Human Genetics, 2016, 61, 335-343.	1.1	7
907	Changing indications for invasive testing in an era of improved screening. Seminars in Perinatology, 2016, 40, 56-66.	1.1	24
908	Karyotype is not dead (yet)!. European Journal of Medical Genetics, 2016, 59, 11-15.	0.7	19
909	Next Generation Sequencing in Dysmorphology. , 2016, , 137-151.		1
910	Prevalence of Creatine Deficiency Syndromes in Children With Nonsyndromic Autism. Pediatrics, 2016, 137, .	1.0	13
911	Microarray Technology. Methods in Molecular Biology, 2016, , .	0.4	4
912	Application of DNA Microarray to Clinical Diagnostics. Methods in Molecular Biology, 2016, 1368, 111-132.	0.4	10
913	Genomic intensive care: should we perform genome testing in critically ill newborns?: TableÂ1. Archives of Disease in Childhood: Fetal and Neonatal Edition, 2016, 101, F94-F98.	1.4	23
914	Fetal growth and gestational factors as predictors of schizophrenia in 22q11.2 deletion syndrome. Genetics in Medicine, 2016, 18, 350-355.	1.1	29
915	Recurrent copy number variations as risk factors for neurodevelopmental disorders: critical overview and analysis of clinical implications. Journal of Medical Genetics, 2016, 53, 73-90.	1.5	90
916	The Diagnostic Yield of Array Comparative Genomic Hybridization Is High Regardless of Severity of Intellectual Disability/Developmental Delay in Children. Journal of Child Neurology, 2016, 31, 691-699.	0.7	37
917	Genetic studies in intellectual disability and related disorders. Nature Reviews Genetics, 2016, 17, 9-18.	7.7	614
918	Comparison of two subtelomeric assays for the screening of chromosomal rearrangements: analysis of 383 patients, literature review and further recommendations. Journal of Applied Genetics, 2016, 57, 63-69.	1.0	1
919	Xp11.2 microduplications including IQSEC2, TSPYL2 and KDM5C genes in patients with neurodevelopmental disorders. European Journal of Human Genetics, 2016, 24, 373-380.	1.4	43

#	Article	IF	Citations
920	Unexplained developmental delay/learning disability: guidelines for best practice protocol for first line assessment and genetic/metabolic/radiological investigations. Irish Journal of Medical Science, 2016, 185, 241-248.	0.8	21
921	Phenotypic Association Analyses With Copy Number Variation in Recurrent Depressive Disorder. Biological Psychiatry, 2016, 79, 329-336.	0.7	21
922	Clinical performance of the CytoScan Dx Assay in diagnosing developmental delay/intellectual disability. Genetics in Medicine, 2016, 18, 168-173.	1.1	9
923	Fenotipo en pacientes con discapacidad intelectual y array-CGH patolÃ³gico. NeurologÃa, 2017, 32, 568-578.	0.3	6
924	Experiences in microarrayâ€based evaluation of developmental disabilities and congenital anomalies. Clinical Genetics, 2017, 92, 372-379.	1.0	9
925	Clinical and molecular cytogenetic analyses of four families with 1q21.1 microdeletion or microduplication. Journal of Gene Medicine, 2017, 19, e2948.	1.4	18
926	â€œSet in Stoneâ€•or â€œRay of Hopeâ€: Parentsâ€™ Beliefs About Cause and Prognosis After Genomic Testing of Children Diagnosed with ASD. Journal of Autism and Developmental Disorders, 2017, 47, 1453-1463.	1.7	18
927	Microarrays in prenatal diagnosis. Best Practice and Research in Clinical Obstetrics and Gynaecology, 2017, 42, 53-63.	1.4	43
928	Exome analysis of Smithâ€“Magenis-like syndrome cohort identifies de novo likely pathogenic variants. Human Genetics, 2017, 136, 409-420.	1.8	22
929	Large patent ductus arteriosus and left ventricular non-compaction as cardiac manifestations of 1p36 monosomy. Pediatria Polska, 2017, 92, 321-324.	0.1	0
930	The Use of Next-Generation Sequencing for Research and Diagnostics for Intellectual Disability. Cold Spring Harbor Perspectives in Medicine, 2017, 7, a026864.	2.9	53
931	Genetic advances in craniosynostosis. American Journal of Medical Genetics, Part A, 2017, 173, 1406-1429.	0.7	84
933	Prenatal microarray comparative genomic hybridization: Experience from the two first years of activity at the Lyon university-hospital. Journal of Gynecology Obstetrics and Human Reproduction, 2017, 46, 275-283.	0.6	3
934	Autism genetics: opportunities and challenges for clinical translation. Nature Reviews Genetics, 2017, 18, 362-376.	7.7	340
935	Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder. Nature Neuroscience, 2017, 20, 602-611.	7.1	691
936	Fading competency of cytogenetic diagnostic laboratories: the alarm bell has started to ring. European Journal of Human Genetics, 2017, 25, 273-274.	1.4	8
937	Continuing role for classical cytogenetics: Case report of a boy with ring syndrome caused by complete ring chromosome 4 and review of literature. American Journal of Medical Genetics, Part A, 2017, 173, 727-732.	0.7	8
938	High diagnostic yield of syndromic intellectual disability by targeted next-generation sequencing. Journal of Medical Genetics, 2017, 54, 87-92.	1.5	93

#	Article	IF	Citations
939	Guideline recommendations for diagnosis and clinical management of Ring14 syndromeâ€"first report of an ad hoc task force. Orphanet Journal of Rare Diseases, 2017, 12, 69.	1.2	18
940	The influence of SNP-based chromosomal microarray and NIPT on the diagnostic yield in 10,000 fetuses with and without fetal ultrasound anomalies. Human Mutation, 2017, 38, 880-888.	1.1	33
941	Chromosome microarray analysis in the investigation of children with congenital heart disease. BMC Pediatrics, 2017, 17, 117.	0.7	34
942	CMIP haploinsufficiency in two patients with autism spectrum disorder and coâ€occurring gastrointestinal issues. American Journal of Medical Genetics, Part A, 2017, 173, 2101-2107.	0.7	6
943	Copy number variation analysis of patients with intellectual disability from North-West Spain. Gene, 2017, 626, 189-199.	1.0	20
944	Medical genetics in Israel's diverse population. Lancet, The, 2017, 389, 2453-2455.	6.3	9
945	Homozygous microdeletion of the ERI1 and MFHAS1 genes in a patient with intellectual disability, limb abnormalities, and cardiac malformation. American Journal of Medical Genetics, Part A, 2017, 173, 1955-1960.	0.7	7
946	Utilization Management in the Clinical Laboratory and Other Ancillary Services. , 2017, , .		4
947	Array-Based Comparative Genomic Hybridization (aCGH). Methods in Molecular Biology, 2017, 1541, 167-179.	0.4	6
948	Novel Causative Variants in DYRK1A, KARS, and KAT6A Associated with Intellectual Disability and Additional Phenotypic Features. Journal of Pediatric Genetics, 2017, 06, 077-083.	0.3	25
949	Salvage of fetal karyotype information from SNP array data obtained from products of conception with maternal cell contamination. Prenatal Diagnosis, 2017, 37, 781-787.	1.1	2
950	Copy Number Variation in Tourette Syndrome. Neuron, 2017, 94, 1041-1043.	3.8	6
951	Clinical and molecular characterization of three genomic rearrangements at chromosome 22q13.3 associated with autism spectrum disorder. Psychiatric Genetics, 2017, 27, 23-33.	0.6	29
952	Do the data really support ordering fragile X testing as a first-tier test without clinical features?. Genetics in Medicine, 2017, 19, 1317-1322.	1.1	20
953	Copy-number variants and candidate gene mutations in isolated split hand/foot malformation. Journal of Human Genetics, 2017, 62, 877-884.	1.1	16
954	Genetic Testing for Rare and Undiagnosed Diseases. , 2017, , 59-73.		1
955	Chromosome copy number variants in fetuses with syndromic malformations. Birth Defects Research, 2017, 109, 725-733.	0.8	7
956	Current use of chromosomal microarray by Australian paediatricians and implications for the implementation of next generation sequencing. Journal of Paediatrics and Child Health, 2017, 53, 650-656.	0.4	4

#	Article	IF	Citations
957	Retrospective Analysis of Congenital Scoliosis. Spine, 2017, 42, E841-E847.	1.0	13
958	Incidence of the 22q11.2 deletion in a large cohort of miscarriage samples. Molecular Cytogenetics, 2017, 10, 6.	0.4	27
959	A CNV Catalogue. , 2017, , 235-417.		1
960	Array-Comparative Genomic Hybridization/Microarray Analysis: Interpretation of Copy Number Variants. , 2017, , 191-234.		1
961	Two females with mutations in USP9X highlight the variable expressivity of the intellectual disability syndrome. European Journal of Medical Genetics, 2017, 60, 359-364.	0.7	12
962	HERC1 mutations in idiopathic intellectual disability. European Journal of Medical Genetics, 2017, 60, 279-283.	0.7	37
963	The clinical use of chromosomal microarray analysis in detection of fetal chromosomal rearrangements: a study from China Mainland. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2017, 212, 44-50.	0.5	11
964	A clear bias in parental origin of de novo pathogenic CNVs related to intellectual disability, developmental delay and multiple congenital anomalies. Scientific Reports, 2017, 7, 44446.	1.6	19
965	Efficient CNV breakpoint analysis reveals unexpected structural complexity and correlation of dosage-sensitive genes with clinical severity in genomic disorders. Human Molecular Genetics, 2017, 26, 1927-1941.	1.4	20
966	Genomic Disorders and Neurocognitive Impairment in Pediatric CKD. Journal of the American Society of Nephrology: JASN, 2017, 28, 2303-2309.	3.0	36
967	Important Considerations in the Management of Newborns with Cyanosis. NeoReviews, 2017, 18, e258-e264.	0.4	1
968	Copy Number Variations Found in Patients with a Corpus Callosum Abnormality and Intellectual Disability. Journal of Pediatrics, 2017, 185, 160-166.e1.	0.9	25
969	MANAGEMENT OF ENDOCRINE DISEASE: Diagnostic and therapeutic approach of tall stature. European Journal of Endocrinology, 2017, 176, R339-R353.	1.9	29
970	Copy number variants analysis in a cohort of isolated and syndromic developmental delay/intellectual disability reveals novel genomic disorders, position effects and candidate disease genes. Clinical Genetics, 2017, 92, 415-422.	1.0	43
971	Copy number variation in 19 Italian multiplex families with autism spectrum disorder: Importance of synaptic and neurite elongation genes. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2017, 174, 547-556.	1.1	7
972	Chromosomal Microarray Detection of Constitutional Copy Number Variation Using Saliva DNA. Journal of Molecular Diagnostics, 2017, 19, 397-403.	1.2	10
973	Diagnosis and clinical management of duplications and deletions. Fertility and Sterility, 2017, 107, 12-18.	0.5	28
974	Prenatal diagnosis of susceptibility loci for neurodevelopmental disorders â€“ genetic counseling and pregnancy outcome in 57 cases. Prenatal Diagnosis, 2017, 37, 73-80.	1.1	13

#	Article	IF	Citations
975	An Update on Molecular Diagnostic Testing of Human Imprinting Disorders. Journal of Pediatric Genetics, 2017, 06, 003-017.	0.3	21
976	Addition of chromosomal microarray and next generation sequencing to FISH and classical cytogenetics enhances genomic profiling of myeloid malignancies. Cancer Genetics, 2017, 216-217, 128-141.	0.2	13
977	Potential Role of Genomic Sequencing in the Early Diagnosis of Treatable Genetic Conditions. Journal of Pediatrics, 2017, 189, 222-226.e1.	0.9	7
978	Futuristic Look at Genetic and Birth Defect Diagnoses and Treatments. Clinical Obstetrics and Gynecology, 2017, 60, 867-877.	0.6	0
979	Copy Number Variation Disorders. Current Genetic Medicine Reports, 2017, 5, 183-190.	1.9	46
980	Current evidence-based recommendations on investigating children with global developmental delay. Archives of Disease in Childhood, 2017, 102, 1071-1076.	1.0	89
981	Detection of an Underlying 22q11.2 Duplication in a Female Neonate With Trisomy 18. Laboratory Medicine, 2017, 48, 372-375.	0.8	2
982	Prevalence of Pathogenic Copy Number Variation in Adults With Pediatric-Onset Epilepsy and Intellectual Disability. JAMA Neurology, 2017, 74, 1301.	4.5	72
983	Detection of structural mosaicism from targeted and whole-genome sequencing data. Genome Research, 2017, 27, 1704-1714.	2.4	44
984	Efficient detection of chromosome imbalances and single nucleotide variants using targeted sequencing in the clinical setting. European Journal of Medical Genetics, 2017, 60, 667-674.	0.7	10
985	Characteristics of rare and private deletions identified in phenotypically normal individuals. Human Genome Variation, 2017, 4, 17037.	0.4	1
986	Genomic Disorders in Psychiatryâ€"What Does the Clinician Need to Know?. Current Psychiatry Reports, 2017, 19, 82.	2.1	36
987	Is it time to retire fragile X testing as a first-tier test for developmental delay, intellectual disability, and autism spectrum disorder?. Genetics in Medicine, 2017, 19, 1380-1381.	1.1	15
988	Expanding the genetic heterogeneity of intellectual disability. Human Genetics, 2017, 136, 1419-1429.	1.8	122
989	Chromosomal microarray analysis in a cohort of underrepresented population identifies SERINC2 as a novel candidate gene for autism spectrum disorder. Scientific Reports, 2017, 7, 12096.	1.6	21
990	HDR-del: A tool based on Hamming distance for prioritizing pathogenic chromosomal deletions in exome sequencing. Human Mutation, 2017, 38, 1796-1800.	1.1	6
991	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. American Journal of Human Genetics, 2017, 101, 206-217.	2.6	51
992	Epilepsy: Transition from pediatric to adult care. Recommendations of the Ontario epilepsy implementation task force. Epilepsia, 2017, 58, 1502-1517.	2.6	74

#	Article	IF	Citations
993	Copyâ€Number Variants Detection by Lowâ€Pass Wholeâ€Genome Sequencing. Current Protocols in Human Genetics, 2017, 94, 8.17.1-8.17.16.	3.5	19
994	Rare Genome-Wide Copy Number Variation and Expression of Schizophrenia in 22q11.2 Deletion Syndrome. American Journal of Psychiatry, 2017, 174, 1054-1063.	4.0	77
995	Early-Life Epilepsies and the Emerging Role of Genetic Testing. JAMA Pediatrics, 2017, 171, 863.	3.3	125
996	Diagnostic Impact and Cost-effectiveness of Whole-Exome Sequencing for Ambulant Children With Suspected Monogenic Conditions. JAMA Pediatrics, 2017, 171, 855.	3.3	252
997	Partial microduplication in the histone acetyltransferase complex member KANSL1 is associated with congenital heart defects in 22q11.2 microdeletion syndrome patients. Scientific Reports, 2017, 7, 1795.	1.6	28
998	High-Throughput Sequencing as First-Tier Diagnostics in Congenital and Early-Onset Disorders. JAMA Pediatrics, 2017, 171, 833.	3.3	8
999	A novel approach in pediatric telegenetic services: geneticist, pediatrician and genetic counselor team. Genetics in Medicine, 2017, 19, 1260-1267.	1.1	43
1000	Intellectual Disability & Rare Disorders: A Diagnostic Challenge. Advances in Experimental Medicine and Biology, 2017, 1031, 39-54.	0.8	27
1001	The epilepsy phenotype in adult patients with intellectual disability and pathogenic copy number variants. Seizure: the Journal of the British Epilepsy Association, 2017, 53, 86-93.	0.9	4
1002	Uniparental disomy and prenatal phenotype. Medicine (United States), 2017, 96, e8474.	0.4	7
1003	Intellectual Disabilities and Global Developmental Delay. , 2017, , 19-55.		3
1004	Anomalies chromosomiques. Journal De Pediatrie Et De Puericulture, 2017, 30, 249-270.	0.0	1
1005	Definition of a putative pathological region in PARK2 associated with autism spectrum disorder through in silico analysis of its functional structure. Psychiatric Genetics, 2017, 27, 54-61.	0.6	11
1006	Chromosomal Microarray Testing for Children With Unexplained Neurodevelopmental Disorders. JAMA - Journal of the American Medical Association, 2017, 317, 2545.	3.8	17
1007	Clinical mutation assay of tumors. Anti-Cancer Drugs, 2017, 28, 1-10.	0.7	7
1008	Genetic Testing in a Cohort of Complex Esophageal Atresia. Molecular Syndromology, 2017, 8, 236-243.	0.3	7
1009	Uniparental Disomy of Chromosome 15 in Two Cases by Chromosome Microarray: A Lesson Worth Thinking. Cytogenetic and Genome Research, 2017, 152, 1-8.	0.6	5
1010	Mov10 suppresses retroelements and regulates neuronal development and function in the developing brain. BMC Biology, 2017, 15, 54.	1.7	40

#	Article	IF	Citations
1011	Use of clinical chromosomal microarray in Chinese patients with autism spectrum disorderâ€"implications of a copy number variation involving DPP10. Molecular Autism, 2017, 8, 31.	2.6	16
1012	Fifteen years of genetic testing from a London developmental clinic. Archives of Disease in Childhood, 2017, 102, 1014-1018.	1.0	5
1013	MLPA analysis in a cohort of patients with autism. Molecular Cytogenetics, 2017, 10, 2.	0.4	1
1014	Molecular dissection of germline chromothripsis in a developmental context using patient-derived iPS cells. Genome Medicine, 2017, 9, 9.	3.6	25
1016	Current Genetic Testing Tools in Neonatal Medicine. Pediatrics and Neonatology, 2017, 58, 111-121.	0.3	21
1017	Fluorescence In Situ Hybridization (FISH). Springer Protocols, 2017, , .	0.1	47
1018	Array CGH. Springer Protocols, 2017, , 567-586.	0.1	1
1020	Chromosomal Microarrays: Understanding Genetics of Neurodevelopmental Disorders and Congenital Anomalies. Journal of Pediatric Genetics, 2017, 06, 042-050.	0.3	24
1021	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. European Journal of Human Genetics, 2017, 25, 176-182.	1.4	291
1022	Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders. Genetics in Medicine, 2017, 19, 667-675.	1.1	143
1023	Universal Prenatal Chromosomal Microarray Analysis: Additive Value and Clinical Dilemmas in Fetuses with a Normal Karyotype. American Journal of Perinatology, 2017, 34, 340-348.	0.6	21
1024	Prenatal genetic counselling for psychiatric disorders. Prenatal Diagnosis, 2017, 37, 6-13.	1.1	17
1025	Chromosomal microarray testing in adults with intellectual disability presenting with comorbid psychiatric disorders. European Journal of Human Genetics, 2017, 25, 66-72.	1.4	30
1026	Genetic studies in a patient with X-linked retinoschisis coexisting with developmental delay and sensorineural hearing loss. Ophthalmic Genetics, 2017, 38, 260-266.	0.5	5
1027	Chromosomal microarray in a highly consanguineous population: diagnostic yield, utility of regions of homozygosity, and novel mutations. Clinical Genetics, 2017, 91, 616-622.	1.0	39
1028	Nonâ€invasive prenatal screening for chromosomal abnormalities using circulating cell-free fetal DNA in maternal plasma: Current applications, limitations and prospects. Egyptian Journal of Medical Human Genetics, 2017, 18, 1-7.	0.5	4
1029	Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. Molecular Psychiatry, 2017, 22, 615-624.	4.1	187
1030	Array comparative genomic hybridization and genomic sequencing in the diagnostics of the causes of congenital anomalies. Journal of Applied Genetics, 2017, 58, 185-198.	1.0	6

#	Article	IF	Citations
1031	Communicating microarray results of uncertain clinical significance in consultation summary letters and implications for practice. European Journal of Human Genetics, 2017, 25, 22-30.	1.4	4
1032	Genomic findings in patients with clinical suspicion of 22q11.2 deletion syndrome. Journal of Applied Genetics, 2017, 58, 93-98.	1.0	21
1033	Clinical relevance of small copy-number variants in chromosomal microarray clinical testing. Genetics in Medicine, 2017, 19, 377-385.	1.1	24
1034	Mycophenolate mofetil embryopathy: A newly recognized teratogenic syndrome. European Journal of Medical Genetics, 2017, 60, 16-21.	0.7	71
1035	Bridging the gap from prenatal karyotyping to whole-genome array comparative genomic hybridization in Hong Kong: survey on knowledge and acceptance of health-care providers and pregnant women. Archives of Gynecology and Obstetrics, 2017, 296, 1109-1116.	0.8	5
1036	Phenotype in patients with intellectual disability and pathological results in array CGH. NeurologÃa (English Edition), 2017, 32, 568-578.	0.2	3
1037	Distal 22q11.2 Microduplication. Child Neurology Open, 2017, 4, 2329048X1773765.	0.5	13
1038	Whole-Exome Sequencing Identifies a de novo AHDC1 Mutation in a Colombian Patient with Xia-Gibbs Syndrome. Molecular Syndromology, 2017, 8, 308-312.	0.3	21
1039	Applying Multivariate Adaptive Splines to Identify Genes With Expressions Varying After Diagnosis in Microarray Experiments. Cancer Informatics, 2017, 16, 117693511770538.	0.9	3
1040	Introduction to Human Genetics â^—. , 2017, , 281-311.		0
1041	Cri-du-Chat syndrome diagnosed in a 21-year-old woman by means of comparative genomic hybridization. Revista Facultad De Medicina, 2017, 65, 525-529.	0.0	1
1042	Genomewide Array Comparative Genomic Hybridization in 55 JapaneseNormokaryotypic Patients with Non-Syndromic Intellectual Disability. Neonatal and Pediatric Medicine, 2017, s1, .	0.1	0
1043	DNA Microarrays and Genetic Testing. , 2017, , 327-337.		1
1044	Cytogenomic assessment of the diagnosis of 93 patients with developmental delay and multiple congenital abnormalities: The Brazilian experience. Clinics, 2017, 72, 526-537.	0.6	10
1045	Global Developmental Delay and Intellectual Disability. , 2017, , 418-423.		1
1046	Partial monosomy 8p and trisomy 16q in two children with developmental delay detected by array comparative genomic hybridization. Molecular Medicine Reports, 2017, 16, 8808-8818.	1.1	3
1047	Application of array comparative genomic hybridization in Korean children under 6 years old with global developmental delay. Korean Journal of Pediatrics, 2017, 60, 282.	1.9	2
1048	Reclassifying Anaphylaxis to Neuromuscular Blocking Agents Based on the Presumed Patho-Mechanism: IgE-Mediated, Pharmacological Adverse Reaction or â€œInnate Hypersensitivityâ€?. International Journal of Molecular Sciences, 2017, 18, 1223.	1.8	73

#	Article	IF	Citations
1049	Molecular Genetic Analysis of Human Endometrial Mesenchymal Stem Cells That Survived Sublethal Heat Shock. Stem Cells International, 2017, 2017, 1-14.	1.2	10
1050	Cognitive and Motor Regression. , 2017, , 424-430.		3
1051	Molecular Pathology and Testing in Melanocytic Tumors. , 2017, , 321-337.		1
1052	Future Diagnostic Strategiesâ€"Pediatric. , 2017, , 361-379.		0
1053	Improving molecular diagnosis of aniridia and WAGR syndrome using customized targeted array-based CGH. PLoS ONE, 2017, 12, e0172363.	1.1	26
1054	17q23.2q23.3 de novo duplication in association with speech and language disorder, learning difficulties, incoordination, motor skill impairment, and behavioral disturbances: a case report. BMC Medical Genetics, 2017, 18, 119.	2.1	9
1055	De novo chromosome 7q36.1q36.2 triplication in a child with developmental delay, growth failure, distinctive facial features, and multiple congenital anomalies: a case report. BMC Medical Genetics, 2017, 18, 118.	2.1	11
1056	Genotype-phenotype correlations and expansion of the molecular spectrum of AP4M1-related hereditary spastic paraplegia. Orphanet Journal of Rare Diseases, 2017, 12, 172.	1.2	17
1057	Advances in paediatrics in 2016: current practices and challenges in allergy, autoimmune diseases, cardiology, endocrinology, gastroenterology, infectious diseases, neonatology, nephrology, neurology, nutrition, pulmonology. Italian Journal of Pediatrics, 2017, 43, 80.	1.0	2
1058	Intersociety policy statement on the use of whole-exome sequencing in the critically ill newborn infant. Italian Journal of Pediatrics, 2017, 43, 100.	1.0	51
1059	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	3.6	50
1060	Impact of IQ on the diagnostic yield of chromosomal microarray in a community sample of adults with schizophrenia. Genome Medicine, 2017, 9, 105.	3.6	30
1061	Epileptic Seizures Associated with Chromosomal Abnormalities Detected by Array Comparative Genomic Hybridization in Five Albanian Children. Journal of Pediatric Epilepsy, 2017, 06, 156-160.	0.1	0
1062	Clinical Lung Cancer Mutation Detection. , 0, , .		0
1064	Automation of the Molecular Diagnostic Laboratory. , 2017, , 35-46.		3
1065	Chromosomal microarray in clinical diagnosis: a study of 337 patients with congenital anomalies and developmental delays or intellectual disability. Croatian Medical Journal, 2017, 58, 231-238.	0.2	14
1066	Bannayan-Riley-Ruvalcaba Syndrome in a Patient with aPTENMutation Identified by Chromosomal Microarray Analysis: A Case Report. Pediatric Gastroenterology, Hepatology and Nutrition, 2017, 20, 65.	0.4	4
1067	Genetic Testing among Children in a Complex Care Program. Children, 2017, 4, 42.	0.6	14

#	Article	IF	Citations
1069	Chromosomes and Chromosomal Abnormalities. , 2017, , 268-276.		6
1070	Techniques for detecting chromosomal aberrations in myelodysplastic syndromes. Oncotarget, 2017, 8, 62716-62729.	0.8	6
1071	Genetics, Risks of Recurrence, and Genetic Counseling. , 0, , 54-84.		1
1072	The Genetic and Epigenetic Basis Involved in the Pathophysiology of ASD: Therapeutic Implications. , 0, , .		1
1073	Use of Array Comparative Genomic Hybridization for the Diagnosis of DiGeorge Syndrome in Saudi Arabian Population. Cytogenetic and Genome Research, 2018, 154, 20-29.	0.6	6
1074	Role of copy number variants in sudden cardiac death and related diseases: genetic analysis and translation into clinical practice. European Journal of Human Genetics, 2018, 26, 1014-1025.	1.4	26
1075	Application of highâ€resolution array platform for genomeâ€wide copy number variation analysis in patients with nonsyndromic cleft lip and palate. Journal of Clinical Laboratory Analysis, 2018, 32, e22428.	0.9	12
1076	Yield of karyotyping in children with developmental delay and/or dysmorphic features in Sohag University Hospital, Upper Egypt. Egyptian Journal of Medical Human Genetics, 2018, 19, 253-259.	0.5	5
1077	Autism spectrum disorder and genetic testing: Parental perceptions and decisionâ€making. Journal for Specialists in Pediatric Nursing, 2018, 23, e12211.	0.6	9
1078	Practice guideline: joint CCMG-SOGC recommendations for the use of chromosomal microarray analysis for prenatal diagnosis and assessment of fetal loss in Canada. Journal of Medical Genetics, 2018, 55, 215-221.	1.5	84
1079	Use of Genetic Testing for Primary Immunodeficiency Patients. Journal of Clinical Immunology, 2018, 38, 320-329.	2.0	88
1080	An estimation of the prevalence of genomic disorders using chromosomal microarray data. Journal of Human Genetics, 2018, 63, 795-801.	1.1	49
1081	Reply to letter: â€œBrain MRI abnormalities resembling unidentified bright objects in a patient with Phelan- McDermid syndromeâ€. European Journal of Paediatric Neurology, 2018, 22, 570.	0.7	0
1082	Cutis laxa in a patient with 1p36 deletion syndrome. Journal of Dermatology, 2018, 45, 871-873.	0.6	7
1083	Progress in the genetics of autism spectrum disorder. Developmental Medicine and Child Neurology, 2018, 60, 445-451.	1.1	116
1084	Getting serious about the early-life epilepsies. Neurology, 2018, 90, 842-848.	1.5	8
1085	Elucidating the diagnostic odyssey of 22q11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 936-944.	0.7	45
1086	Phase determination using chromosomal microarray and fluorescence inÂsitu hybridization in a patient with early onset Parkinson disease and two deletions in PRKN. Molecular Genetics & Genomic Medicine, 2018, 6, 457-462.	0.6	7

#	Article	IF	Citations
1087	TASP1 is deleted in an infant with developmental delay, microcephaly, distinctive facial features, and multiple congenital anomalies. Clinical Genetics, 2018, 94, 170-173.	1.0	9
1088	Genetic disorders and mortality in infancy and early childhood: delayed diagnoses and missed opportunities. Genetics in Medicine, 2018, 20, 1396-1404.	1.1	58
1089	Interstitial Chromosome 3p13p14 Deletions: An Update and Review. Molecular Syndromology, 2018, 9, 122-133.	0.3	3
1090	Cost-effectiveness of Genome and Exome Sequencing in Children Diagnosed with Autism Spectrum Disorder. Applied Health Economics and Health Policy, 2018, 16, 481-493.	1.0	20
1091	When ultrasound anomalies are present: An estimation of the frequency of chromosome abnormalities not detected by cellâ€free DNA aneuploidy screens. Prenatal Diagnosis, 2018, 38, 250-257.	1.1	13
1092	A genetic approach to evaluation of short stature of undetermined cause. Lancet Diabetes and Endocrinology,the, 2018, 6, 564-574.	5.5	54
1093	Paediatric genomics: diagnosing rare disease in children. Nature Reviews Genetics, 2018, 19, 253-268.	7.7	369
1094	Utility of Genetic Testing in Fetal Alcohol Spectrum Disorder. Journal of Pediatrics, 2018, 196, 270-274.e1.	0.9	10
1095	Prenatal diagnosis by chromosomal microarray analysis. Fertility and Sterility, 2018, 109, 201-212.	0.5	194
1097	Client-centred clinical genetic diagnostics. Advances in Mental Health and Intellectual Disabilities, 2018, 12, 1-10.	0.7	3
1098	Current controversies in prenatal diagnosis 2: Cellâ€free DNA prenatal screening should be used to identify all chromosome abnormalities. Prenatal Diagnosis, 2018, 38, 160-165.	1.1	29
1099	Clinical approach to the patient with neurogenetic disease. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 3-9.	1.0	7
1100	Novel contiguous gene deletion in peruvian girl with Trichothiodystrophy type 4 and glutaric aciduria type 3. European Journal of Medical Genetics, 2018, 61, 388-392.	0.7	7
1101	Intellectual disability and epilepsy due to the K/Lâ€mediated Xq28 duplication: Further evidence of a distinct, dosageâ€dependent phenotype. American Journal of Medical Genetics, Part A, 2018, 176, 551-559.	0.7	9
1102	Copy number variations and fetal ventriculomegaly. Current Opinion in Obstetrics and Gynecology, 2018, 30, 104-110.	0.9	13
1103	Pathogenic copy number variants in patients with congenital hypopituitarism associated with complex phenotypes. Clinical Endocrinology, 2018, 88, 425-431.	1.2	11
1104	The Use of Chromosomal Microarray Analysis in Prenatal Diagnosis. Obstetrics and Gynecology Clinics of North America, 2018, 45, 55-68.	0.7	49
1105	Genetic short stature. Growth Hormone and IGF Research, 2018, 38, 29-33.	0.5	36

#	Article	IF	Citations
1106	Genomic medicine for kidney disease. Nature Reviews Nephrology, 2018, 14, 83-104.	4.1	102
1107	Monogenic Obesity. Contemporary Endocrinology, 2018, , 135-152.	0.3	12
1108	Genome-wide sequencing technologies: A primer for paediatricians. Paediatrics and Child Health, 2018, 23, 191-197.	0.3	10
1109	Utility of chromosomal microarray in anomalous fetuses. Prenatal Diagnosis, 2018, 38, 140-147.	1.1	9
1110	A Comprehensive Workflow for Read Depth-Based Identification of Copy-Number Variation from Whole-Genome Sequence Data. American Journal of Human Genetics, 2018, 102, 142-155.	2.6	156
1111	Role of Striatal Direct Pathway 2-Arachidonoylglycerol Signaling in Sociability and Repetitive Behavior. Biological Psychiatry, 2018, 84, 304-315.	0.7	36
1112	Developmental Defects Associated With DNA Copy Number Gain of Chromosome 2q33.1: A Case Report and Review of Literature. Laboratory Medicine, 2018, 49, 160-164.	0.8	0
1113	Disruptive Behavior, Global Developmental Delay, and Obesity in a 5-Year-Old Boy with a Chromosome Microduplication. Journal of Developmental and Behavioral Pediatrics, 2018, 39, 81-84.	0.6	4
1114	Prematurity, ventricular septal defect and dysmorphisms are independent predictors of pathogenic copy number variants: a retrospective study on array-CGH results and phenotypical features of 293 children with neurodevelopmental disorders and/or multiple congenital anomalies. Italian Journal of Pediatrics, 2018, 44, 34.	1.0	7
1115	Small de novo CNVs as biomarkers of parental exposure to low doses of ionizing radiation of caesium-137. Scientific Reports, 2018, 8, 5914.	1.6	18
1116	Neurodevelopmental risk copy number variants in adults with intellectual disabilities and comorbid psychiatric disorders. British Journal of Psychiatry, 2018, 212, 287-294.	1.7	30
1117	Measuring and Estimating the Effect Sizes of Copy Number Variants on General Intelligence in Community-Based Samples. JAMA Psychiatry, 2018, 75, 447.	6.0	77
1118	Patterns of homozygosity in patients with uniparental disomy: detection rate and suggested reporting thresholds for SNP microarrays. Genetics in Medicine, 2018, 20, 1522-1527.	1.1	35
1119	Diagnostic efficacy and new variants in isolated and complex autism spectrum disorder using molecular karyotyping. Journal of Applied Genetics, 2018, 59, 179-185.	1.0	12
1120	Is prenatal genomic testing ready for prime time?. Genetics in Medicine, 2018, 20, 695-696.	1.1	1
1121	Chromosomal microarray as primary diagnostic genomic tool for pregnancies at increased risk within a populationâ€based combined firstâ€trimester screening program. Ultrasound in Obstetrics and Gynecology, 2018, 51, 480-486.	0.9	46
1122	Diagnostic application of a capture based <scp>NGS</scp> test for the concurrent detection of variants in sequence and copy number as well as <scp>LOH</scp>. Clinical Genetics, 2018, 93, 545-556.	1.0	12
1123	Genomic diagnostics within a medically underserved population: efficacy and implications. Genetics in Medicine, 2018, 20, 31-41.	1.1	47

#	Article	IF	Citations
1124	When genotype is not predictive of phenotype: implications for genetic counseling based on 21,594 chromosomal microarray analysis examinations. Genetics in Medicine, 2018, 20, 128-131.	1.1	47
1125	Using medical exome sequencing to identify the causes of neurodevelopmental disorders: Experience of 2 clinical units and 216 patients. Clinical Genetics, 2018, 93, 567-576.	1.0	82
1126	Towards precision nephrology: the opportunities and challenges of genomic medicine. Journal of Nephrology, 2018, 31, 47-60.	0.9	13
1127	Pathways from autism spectrum disorder diagnosis to genetic testing. Genetics in Medicine, 2018, 20, 737-744.	1.1	29
1129	A de novo mutation in RPL10 causes a rare X-linked ribosomopathy characterized by syndromic intellectual disability and epilepsy: A new case and review of the literature. European Journal of Medical Genetics, 2018, 61, 89-93.	0.7	22
1130	Ethical considerations in prenatal testing: Genomic testing and medical uncertainty. Seminars in Fetal and Neonatal Medicine, 2018, 23, 1-6.	1.1	35
1131	Exome sequencing reveals NAA15 and PUF60 as candidate genes associated with intellectual disability. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 10-20.	1.1	32
1132	Genomic disorders 20 years onâ€"mechanisms for clinical manifestations. Clinical Genetics, 2018, 93, 439-449.	1.0	102
1133	Genetic testing in intellectual disability psychiatry: Opinions and practices of <scp>UK</scp> child and intellectual disability psychiatrists. Journal of Applied Research in Intellectual Disabilities, 2018, 31, 273-284.	1.3	16
1134	CNVs affecting cancer predisposing genes (CPGs) detected as incidental findings in routine germline diagnostic chromosomal microarray (CMA) testing. Journal of Medical Genetics, 2018, 55, 89-96.	1.5	7
1135	Prenatal diagnosis of posterior fossa anomalies: Additional value of chromosomal microarray analysis in fetuses with cerebellar hypoplasia. Prenatal Diagnosis, 2018, 38, 91-98.	1.1	17
1136	Cost Effectiveness of Karyotyping, Chromosomal Microarray Analysis, and Targeted Next-Generation Sequencing of Patients with Unexplained Global Developmental Delay or Intellectual Disability. Molecular Diagnosis and Therapy, 2018, 22, 129-138.	1.6	23
1137	The role of CNVs in the etiology of rare autosomal recessive disorders: the example of TRAPPC9-associated intellectual disability. European Journal of Human Genetics, 2018, 26, 143-148.	1.4	26
1138	19p13 microduplications encompassing NFIX are responsible for intellectual disability, short stature and small head circumference. European Journal of Human Genetics, 2018, 26, 85-93.	1.4	7
1139	Array-CGH Analysis in a Cohort of Phenotypically Well-Characterized Individuals with â€œEssentialâ€• Autism Spectrum Disorders. Journal of Autism and Developmental Disorders, 2018, 48, 442-449.	1.7	19
1140	Screening for fetal chromosomal and subchromosomal disorders. Seminars in Fetal and Neonatal Medicine, 2018, 23, 85-93.	1.1	10
1141	Interstitial microdeletion of 17q11.2 is associated with hypotonia, fatigue, intellectual disability, and a subtle facial phenotype in three unrelated patients. American Journal of Medical Genetics, Part A, 2018, 176, 209-213.	0.7	2
1142	Recurrent Copy Number Variants Associated with Syndromic Short Stature of Unknown Cause. Hormone Research in Paediatrics, 2018, 89, 13-21.	0.8	29

#	Article	IF	Citations
1143	Genetic evaluation of patients with congenital heart disease. Current Opinion in Pediatrics, 2018, 30, 707-713.	1.0	19
1144	Presentation of a recurrent FMR1 missense mutation (R138Q) in an affected female. Translational Science of Rare Diseases, 2018, 3, 139-144.	1.6	7
1145	Prenatal diagnosis and pregnancy outcome analysis of thickened nuchal fold in the second trimester. Medicine (United States), 2018, 97, e13334.	0.4	12
1146	Targeted Next-Generation Sequencing of Korean Patients With Developmental Delay and/or Intellectual Disability. Frontiers in Pediatrics, 2018, 6, 391.	0.9	36
1147	Genetic testing in children and adolescents with intellectual disability. Current Opinion in Psychiatry, 2018, 31, 490-495.	3.1	15
1148	Is It Time for Genome Sequencing to Replace Exome Sequencing?. Journal of Molecular and Genetic Medicine: an International Journal of Biomedical Research, 2018, 12, .	0.1	0
1150	Chromosomal Microarray Analysis Results From Pregnancies With Various Ultrasonographic Anomalies. Obstetrics and Gynecology, 2018, 132, 1368-1375.	1.2	29
1151	From bedside to bench and back: Translating ASD models. Progress in Brain Research, 2018, 241, 113-158.	0.9	2
1152	Diagnosis of Smith-Magenis Syndrome in a Patient with Mental Retardation and Sleep Disturbance Confirmed by Multiplex Ligation-dependent Probe Amplification. Laboratory Medicine Online, 2018, 8, 71.	0.0	0
1153	Increasing the diagnostic yield of exome sequencing by copy number variant analysis. PLoS ONE, 2018, 13, e0209185.	1.1	58
1154	The etiology of VACTERL association: Current knowledge and hypotheses. , 2018, 178, 440-446.		41
1155	Extending the Phenotype and Identification of a Novel Candidate Gene for Immunodeficiency in 5q11 Microdeletion Syndrome. Molecular Syndromology, 2018, 9, 312-318.	0.3	2
1156	Complex structural variants in Mendelian disorders: identification and breakpoint resolution using short- and long-read genome sequencing. Genome Medicine, 2018, 10, 95.	3.6	111
1157	Genetics and genomics in Peru: Clinical and research perspective. Molecular Genetics & Genomic Medicine, 2018, 6, 873-886.	0.6	12
1158	Rare Copy Number Variations in a Chinese Cohort of Autism Spectrum Disorder. Frontiers in Genetics, 2018, 9, 665.	1.1	10
1159	Associated anomalies in cases with anorectal anomalies. American Journal of Medical Genetics, Part A, 2018, 176, 2646-2660.	0.7	10
1160	Rare Copy Number Variations and Predictors in Children With Intellectual Disability and Epilepsy. Frontiers in Neurology, 2018, 9, 947.	1.1	23
1161	13q mosaic deletion including RB1 associated to mild phenotype and no cancer outcome â€“ case report and review of the literature. Molecular Cytogenetics, 2018, 11, 53.	0.4	2

#	Article	IF	Citations
1162	Heterogeneity in clinical sequencing tests marketed for autism spectrum disorders. Npj Genomic Medicine, 2018, 3, 27.	1.7	26
1163	Chromosomal Abnormalities in Syndromic Orofacial Clefts: Report of Three Children. Case Reports in Genetics, 2018, 2018, 1-5.	0.1	1
1164	Genome-Wide Array Analysis Reveals Novel Genomic Regions and Candidate Gene for Intellectual Disability. Molecular Diagnosis and Therapy, 2018, 22, 749-757.	1.6	5
1165	Importance and usage of chromosomal microarray analysis in diagnosing intellectual disability, global developmental delay, and autism; and discovering new loci for these disorders. Molecular Cytogenetics, 2018, 11, 54.	0.4	9
1166	Investigating the child with intellectual disability. Journal of Paediatrics and Child Health, 2018, 54, 1154-1158.	0.4	12
1167	Clinical analysis of germline copy number variation in DMD using a non-conjugate hierarchical Bayesian model. BMC Medical Genomics, 2018, 11, 91.	0.7	3
1168	Parental Knowledge and Perceptions of Pediatric Genomic Testing for Autism Spectrum Disorders in Rural Settings. Advances in Neurodevelopmental Disorders, 2018, 2, 402-411.	0.7	5
1169	Interest of chromosomal microarray analysis in the prenatal diagnosis of fetal intrauterine growth restriction. Prenatal Diagnosis, 2018, 38, 1111-1119.	1.1	10
1170	Assessing copy number abnormalities and copy-neutral loss-of-heterozygosity across the genome as best practice in diagnostic evaluation of acute myeloid leukemia: An evidence-based review from the cancer genomics consortium (CGC) myeloid neoplasms working group. Cancer Genetics, 2018, 228-229, 218-235.	0.2	21
1171	Quality control guidelines for clinical-grade human induced pluripotent stem cell lines. Regenerative Medicine, 2018, 13, 859-866.	0.8	147
1172	Multicolor Fluorescence In Situ Hybridization (FISH) Approaches for Simultaneous Analysis of the Entire Human Genome. Current Protocols in Human Genetics, 2018, 99, e70.	3.5	4
1173	Case Definitions for Conditions Identified by Newborn Screening Public Health Surveillance. International Journal of Neonatal Screening, 2018, 4, 16.	1.2	17
1174	Effect of assisted reproductive technology on the molecular karyotype of missed abortion tissues. Bioscience Reports, 2018, 38, .	1.1	12
1175	Diagnostic Testing in Epilepsy Genetics Clinical Practice. , 2018, , .		0
1176	A Multicentric Brazilian Investigative Study of Copy Number Variations in Patients with Congenital Anomalies and Intellectual Disability. Scientific Reports, 2018, 8, 13382.	1.6	1
1177	The clinical implementation of copy number detection in the age of next-generation sequencing. Expert Review of Molecular Diagnostics, 2018, 18, 907-915.	1.5	8
1178	The Cytoscan HD Array in the Diagnosis of Neurodevelopmental Disorders. High-Throughput, 2018, 7, 28.	4.4	19
1179	Intellectual and Developmental Disability. , 2018, , 367-392.e2.		1

#	Article	IF	Citations
1180	Whole-Genome Sequencing as a Method of Prenatal Genetic Diagnosis. , 2018, , 263-291.		0
1181	Mosaic Tetrasomy of 9p24.3q21.11 postnatally identified in an infant born with multiple congenital malformations: a case report. BMC Pediatrics, 2018, 18, 298.	0.7	4
1182	Molecular Karyotyping in Children and Adolescents with Gender Dysphoria. Transgender Health, 2018, 3, 147-153.	1.2	8
1183	CGH analysis in Colombian patients: findings of 1374 arrays in a seven-year study. Molecular Cytogenetics, 2018, 11, 46.	0.4	5
1184	The Genetic Evaluation of a Child With Cancer. , 2018, , 21-31.		1
1185	Identification of rare de novo epigenetic variations in congenital disorders. Nature Communications, 2018, 9, 2064.	5.8	82
1186	Genetic Testing Techniques. , 2018, , 47-64.		18
1187	Genetic testing and autism: Tutorial for communication sciences and disorders. Journal of Communication Disorders, 2018, 74, 61-73.	0.8	3
1188	Genetics in mainstream medicine: Finally within grasp to influence healthcare globally. Molecular Genetics & Genomic Medicine, 2018, 6, 473-480.	0.6	8
1189	Novel applications of array comparative genomic hybridization in molecular diagnostics. Expert Review of Molecular Diagnostics, 2018, 18, 531-542.	1.5	28
1190	Enrichment of rare copy number variation in children with developmental language disorder. Clinical Genetics, 2018, 94, 313-320.	1.0	19
1191	Genome-wide copy number variation analysis identifies novel candidate loci associated with pediatric obesity. European Journal of Human Genetics, 2018, 26, 1588-1596.	1.4	23
1192	Clinical Application of Chromosome Microarray Analysis in Han Chinese Children with Neurodevelopmental Disorders. Neuroscience Bulletin, 2018, 34, 981-991.	1.5	7
1193	Rare structural variants in the DOCK8 gene identified in a cohort of 439 patients with neurodevelopmental disorders. Scientific Reports, 2018, 8, 9449.	1.6	13
1194	Phenotypic Analysis of Korean Patients with Abnormal Chromosomal Microarray in Patients with Unexplained Developmental Delay/Intellectual Disability. Yonsei Medical Journal, 2018, 59, 431.	0.9	4
1195	Chromosomal microarray analysis in developmental delay and intellectual disability with comorbid conditions. BMC Medical Genomics, 2018, 11, 49.	0.7	26
1196	A Pure 2-Mb 3q26.2 Duplication Proximal to the Critical Region of 3q Duplication Syndrome. Molecular Syndromology, 2018, 9, 197-204.	0.3	2
1197	Peri-mortem evaluation of infants who die without a diagnosis: focus on advances in genomic technology. Journal of Perinatology, 2018, 38, 1125-1134.	0.9	9

#	Article	IF	Citations
1198	Advancing Stem Cell Models of Alpha-Synuclein Gene Regulation in Neurodegenerative Disease. Frontiers in Neuroscience, 2018, 12, 199.	1.4	19
1199	Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. Npj Genomic Medicine, 2018, 3, 16.	1.7	420
1200	Rothmund-Thomson Syndrome: Insights from New Patients on the Genetic Variability Underpinning Clinical Presentation and Cancer Outcome. International Journal of Molecular Sciences, 2018, 19, 1103.	1.8	20
1201	Application of chromosomal microarray to investigate genetic causes of isolated fetal growth restriction. Molecular Cytogenetics, 2018, 11, 33.	0.4	16
1202	The coexistence of copy number variations (CNVs) and single nucleotide polymorphisms (SNPs) at a locus can result in distorted calculations of the significance in associating SNPs to disease. Human Genetics, 2018, 137, 553-567.	1.8	57
1203	Forging the path for PRecISion Medicine in Autism: The PRISMA Neurogenetic Psychiatry Consultation Service. The Brown University Child and Adolescent Behavior Letter, 2018, 34, 1-7.	0.0	0
1204	Identification of novel genomic imbalances in Saudi patients with congenital heart disease. Molecular Cytogenetics, 2018, 11, 9.	0.4	4
1205	A feasible diagnostic approach for the translocation carrier from the indication of products of conception. Molecular Cytogenetics, 2018, 11, 12.	0.4	10
1206	Collaborations in medical genetics: 10â€Year history of an ongoing Vietnameseâ€North American Collaboration. Molecular Genetics & Genomic Medicine, 2018, 6, 129-133.	0.6	2
1207	Classical fragile-X phenotype in a female infant disclosed by comprehensive genomic studies. BMC Medical Genetics, 2018, 19, 74.	2.1	6
1208	Derivative chromosomes involving 5p large rearranged segments went unnoticed with the use of conventional cytogenetics. Molecular Cytogenetics, 2018, 11, 30.	0.4	5
1209	Chromosomal Microarray: Application for Congenital Heart Diseases. Korean Circulation Journal, 2018, 48, 233.	0.7	2
1210	Copy number variant discrepancy resolution using the ClinGen dosage sensitivity map results in updated clinical interpretations in ClinVar. Human Mutation, 2018, 39, 1650-1659.	1.1	23
1211	How Cytogenetics Paradigms Shape Decision Making in Translational Genomics. , 2018, , 45-59.		0
1212	All Along the Watchtower: a Case of Long QT Syndrome Misdiagnosis Secondary to Genetic Testing Misinterpretation. Journal of Genetic Counseling, 2018, 27, 1515-1522.	0.9	6
1213	Chromosomal Microarray With Clinical Diagnostic Utility in Children With Developmental Delay or Intellectual Disability. Annals of Laboratory Medicine, 2018, 38, 473-480.	1.2	27
1214	Microduplication in the 2p16.1p15 chromosomal region linked to developmental delay and intellectual disability. Molecular Cytogenetics, 2018, 11, 39.	0.4	4
1215	A comprehensive clinical and genetic study in 127 patients with ID in Kinshasa, DR Congo. American Journal of Medical Genetics, Part A, 2018, 176, 1897-1909.	0.7	7

#	Article	IF	Citations
1216	Chromosome Disorders. , 2018, , 211-223.e2.		1
1217	Adults With Congenital Heart Disease. , 2018, , 24-30.		0
1218	Genetic and genomic testing for neurologic disease in clinical practice. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 147, 11-22.	1.0	20
1219	A streamlined workflow for single-cells genome-wide copy-number profiling by low-pass sequencing of LM-PCR whole-genome amplification products. PLoS ONE, 2018, 13, e0193689.	1.1	20
1220	The Human Genome and Neonatal Care. , 2018, , 180-189.e2.		0
1222	Yield of additional genetic testing after chromosomal microarray for diagnosis of neurodevelopmental disability and congenital anomalies: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2018, 20, 1105-1113.	1.1	57
1223	Genome-wide sequencing in acutely ill infants: genomic medicineâ€™s critical application?. Genetics in Medicine, 2019, 21, 498-504.	1.1	42
1224	Detection of copy number variants using chromosomal microarray analysis for the prenatal diagnosis of congenital heart defects with normal karyotype. Journal of Clinical Laboratory Analysis, 2019, 33, e22630.	0.9	18
1225	Basic Genetics and Epigenetics of Childhood Lung Disease. , 2019, , 40-48.e2.		1
1226	Novel biallelic PCNT deletion causing microcephalic osteodysplastic primordial dwarfism type II with congenital heart defect. Science China Life Sciences, 2019, 62, 144-147.	2.3	4
1227	Epidemiology of fetal cerebral ventriculomegaly and evaluation of chromosomal microarray analysis versus karyotyping for prenatal diagnosis in a Chinese hospital. Journal of International Medical Research, 2019, 47, 5508-5517.	0.4	7
1228	Cognitive and Behavioral Functioning Among Fraternal Twins with an Unbalanced Translocation of Chromosomes 10q and 12p: A Case Report. Journal of Developmental and Physical Disabilities, 2019, 31, 635-651.	1.0	0
1229	Inherited and De Novo Genetic Risk for Autism Impacts Shared Networks. Cell, 2019, 178, 850-866.e26.	13.5	326
1230	A comparison of genomic laboratory reports and observations that may enhance their clinical utility for providers and patients. Molecular Genetics & Genomic Medicine, 2019, 7, e00551.	0.6	12
1231	Familial inheritance of the 3q29 microdeletion syndrome: case report and review. BMC Medical Genomics, 2019, 12, 51.	0.7	7
1232	Trastorno del espectro autista: DiagnÃ³stico clÃnico y test ADOS. Revista Chilena De Pediatria, 2019, 90, 485-491.	0.4	2
1233	Duplications in 19p13.3 are associated with male infertility. Journal of Assisted Reproduction and Genetics, 2019, 36, 2171-2179.	1.2	19
1234	Identification and In Silico Characterization of a Novel Point Mutation within the Phosphatidylinositol Glycan Anchor Biosynthesis Class G Gene in an Iranian Family with Intellectual Disability. Journal of Molecular Neuroscience, 2019, 69, 538-545.	1.1	9

#	Article	IF	Citations
1235	Wholeâ€genome sequencing revealed an interstitial deletion encompassing OCRL and SMARCA1 gene in a patient with Lowe syndrome. Molecular Genetics & Genomic Medicine, 2019, 7, e876.	0.6	7
1236	Investigating Potential Biomarkers in Autism Spectrum Disorder. Frontiers in Integrative Neuroscience, 2019, 13, 31.	1.0	29
1237	Homozygous Deletion of the CFTR Gene Caused by Interstitial Maternal Isodisomy in a Peruvian Child with Cystic Fibrosis. Journal of Pediatric Genetics, 2019, 08, 147-152.	0.3	2
1238	A clinically validated whole genome pipeline for structural variant detection and analysis. BMC Genomics, 2019, 20, 545.	1.2	15
1239	The rare 13q33â€“q34 microdeletions: eight new patients and review of the literature. Human Genetics, 2019, 138, 1145-1153.	1.8	11
1240	PTENÎ± regulates endocytosis and modulates olfactory function. FASEB Journal, 2019, 33, 11148-11162.	0.2	3
1241	Genomic imbalances defining novel intellectual disability associated loci. Orphanet Journal of Rare Diseases, 2019, 14, 164.	1.2	3
1242	Expanding phenotype with severe midline brain anomalies and missense variant supports a causal role for FOXA2 in 20p11.2 deletion syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 1783-1790.	0.7	10
1243	Quantitative PCR evaluation of deletions/duplications identified by array CGH. Molecular and Cellular Probes, 2019, 46, 101421.	0.9	3
1244	Primary gonadal failure. Best Practice and Research in Clinical Endocrinology and Metabolism, 2019, 33, 101295.	2.2	15
1246	The clinical benefit of array-based comparative genomic hybridization for detection of copy number variants in Czech children with intellectual disability and developmental delay. BMC Medical Genomics, 2019, 12, 111.	0.7	13
1247	Structural variation at the CYP2C locus: Characterization of deletion and duplication alleles. Human Mutation, 2019, 40, e37-e51.	1.1	15
1248	Actions and Uncertainty: How Prenatally Diagnosed Variants of Uncertain Significance Become Actionable. Hastings Center Report, 2019, 49, S61-S71.	0.7	19
1249	Chromosomal Microarray Analysis. , 2019, , 125-136.		0
1250	Pitfalls in molecular diagnostics. Seminars in Diagnostic Pathology, 2019, 36, 342-354.	1.0	10
1251	High resolution Chromosomal Microarray Analysis (CMA) enhances the genetic profile of pediatric B-cell Acute Lymphoblastic Leukemia patients. Leukemia Research, 2019, 83, 106177.	0.4	6
1252	The Genetics of Sudden Infant Death Syndrome. , 2019, , 159-171.		1
1253	Intracranial calcifications and dystonia associated with a novel deletion of chromosome 8p11.2 encompassing SLC20A2 and THAP1. BMJ Case Reports, 2019, 12, e228782.	0.2	7

#	Article	IF	Citations
1254	Genomic Sequencing for Infants and Children in Intensive Care Units. Current Pediatrics Reports, 2019, 7, 78-82.	1.7	1
1255	Model system identification of novel congenital heart disease gene candidates: focus on RPL13. Human Molecular Genetics, 2019, 28, 3954-3969.	1.4	19
1256	Hypogonadotrophic hypogonadism, delayed puberty and risk for neurodevelopmental disorders. Journal of Neuroendocrinology, 2019, 31, e12803.	1.2	13
1257	Genotypic and phenotypic variability of 22q11.2 microduplications: An institutional experience. American Journal of Medical Genetics, Part A, 2019, 179, 2178-2189.	0.7	25
1258	Integration of Large-Scale Genomic Data Sources With Evolutionary History Reveals Novel Genetic Loci for Congenital Heart Disease. Circulation Genomic and Precision Medicine, 2019, 12, 442-451.	1.6	19
1259	A large data resource of genomic copy number variation across neurodevelopmental disorders. Npj Genomic Medicine, 2019, 4, 26.	1.7	118
1260	Paternal impacts on development: identification of genomic regions vulnerable to oxidative DNA damage in human spermatozoa. Human Reproduction, 2019, 34, 1876-1890.	0.4	43
1261	Influence of validating the parental origin on the clinical interpretation of fetal copy number variations in 141 core family cases. Molecular Genetics & Genomic Medicine, 2019, 7, e00944.	0.6	9
1262	De novo Mutations From Whole Exome Sequencing in Neurodevelopmental and Psychiatric Disorders: From Discovery to Application. Frontiers in Genetics, 2019, 10, 258.	1.1	49
1263	Standards of diagnostic assessment for autism spectrum disorder. Paediatrics and Child Health, 2019, 24, 444-451.	0.3	60
1264	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. Genome Medicine, 2019, 11, 68.	3.6	88
1265	Primary care physiciansâ€™ understanding and utilization of pediatric exome sequencing results. Journal of Genetic Counseling, 2019, 28, 1130-1138.	0.9	6
1266	RBV: Read balance validator, a tool for prioritising copy number variations in germline conditions. Scientific Reports, 2019, 9, 16934.	1.6	0
1267	Detection of Point Mutations and Chromosomal Translocations Based on Massive Parallel Sequencing of Enriched 3C Libraries. Russian Journal of Genetics, 2019, 55, 1273-1281.	0.2	7
1268	Subtelomeric Rearrangements: Presentation of 21 Probands with Emphasis on Familial Cases. Acta Medica Portuguesa, 2019, 32, 529.	0.2	1
1269	Genetics and epigenetics of autism spectrum disorderâ€”current evidence in the field. Journal of Applied Genetics, 2019, 60, 37-47.	1.0	139
1271	Associated anomalies in cases with agenesis of the corpus callosum. American Journal of Medical Genetics, Part A, 2019, 179, 2101-2111.	0.7	15
1272	The Road to Diagnosis: Shortening the Diagnostic Odyssey in Epilepsy. Epilepsy Currents, 2019, 19, 307-309.	0.4	10

#	Article	IF	Citations
1273	A Novel Homozygous Deletion within the <i>FRY</i> Gene Associated with Nonsyndromic Developmental Delay. Cytogenetic and Genome Research, 2019, 159, 19-25.	0.6	5
1274	Syndromic Oral Clefts: Challenges of Genetic Assessment in Brazil and Suggestions to Improve Health Policies. Public Health Genomics, 2019, 22, 69-76.	0.6	3
1275	Investigating developmental delay in South Africa: A pragmatic approach. South African Medical Journal, 2019, 109, 210.	0.2	4
1276	Translational Medicine in Neurological Disorders: A Genomic Perspective. Current Genomics, 2019, 20, 151-153.	0.7	1
1277	Identification of Structural Variation from NGS-Based Non-Invasive Prenatal Testing. International Journal of Molecular Sciences, 2019, 20, 4403.	1.8	20
1278	FADD in Cancer: Mechanisms of Altered Expression and Function, and Clinical Implications. Cancers, 2019, 11, 1462.	1.7	31
1279	Association between distress and knowledge among parents of autistic children. PLoS ONE, 2019, 14, e0223119.	1.1	5
1280	Genetic Testing Experiences Among Parents of Children with Autism Spectrum Disorder in the United States. Journal of Autism and Developmental Disorders, 2019, 49, 4821-4833.	1.7	18
1281	Chromosomal Microarray Analysis in Children with Unexplained Developmental Delay/Intellectual Disability. Journal of Pediatric Genetics, 2019, 08, 001-009.	0.3	5
1282	Introduction of genomics into prenatal diagnostics. Lancet, The, 2019, 393, 719-721.	6.3	13
1283	Diagnosis and management in Pittâ€Hopkins syndrome: First international consensus statement. Clinical Genetics, 2019, 95, 462-478.	1.0	63
1284	Genomic Copy Number Variations in the Autism Clinicâ€"Work in Progress. Frontiers in Cellular Neuroscience, 2019, 13, 57.	1.8	24
1285	Chromosomal microarray vs. NIPS: analysis of 5541 low-risk pregnancies. Genetics in Medicine, 2019, 21, 2462-2467.	1.1	55
1286	Importance of patient selection criteria in determining diagnostic copy number variations in patients with multiple congenital anomaly/mental retardation. Molecular Cytogenetics, 2019, 12, 23.	0.4	2
1287	Possibility of early diagnosis in a fetus affected by Praderâ€'Willi syndrome with maternal heteroâ€'UPD15: A lesson to be learned. Molecular Medicine Reports, 2019, 20, 95-102.	1.1	5
1288	9q34.3 microduplications lead to neurodevelopmental disorders through EHMT1 overexpression. Neurogenetics, 2019, 20, 145-154.	0.7	12
1289	Genomics of disease risk in globally diverse populations. Nature Reviews Genetics, 2019, 20, 520-535.	7.7	217
1290	Testing criteria for 22q11.2 deletion syndrome: preliminary results of a low cost strategy for public health. Orphanet Journal of Rare Diseases, 2019, 14, 123.	1.2	11

#	Article	IF	Citations
1291	A machine-learning approach for accurate detection of copy number variants from exome sequencing. Genome Research, 2019, 29, 1134-1143.	2.4	41
1292	A Review of the Genomic Analysis of Children Presenting with Developmental Delay/Intellectual Disability and Associated Dysmorphic Features. Cureus, 2019, 11, e3873.	0.2	11
1293	Potential genetic causes of miscarriage in euploid pregnancies: a systematic review. Human Reproduction Update, 2019, 25, 452-472.	5.2	95
1294	Analytical validation and chromosomal distribution of regions of homozygosity by oligonucleotide array comparative genomic hybridization from normal prenatal and postnatal case series. Molecular Cytogenetics, 2019, 12, 12.	0.4	5
1295	Needs assessment in genetic testing education: A survey of parents of children with autism spectrum disorder in the united states. Autism Research, 2019, 12, 1162-1170.	2.1	15
1296	Meta-analysis and multidisciplinary consensus statement: exome sequencing is a first-tier clinical diagnostic test for individuals with neurodevelopmental disorders. Genetics in Medicine, 2019, 21, 2413-2421.	1.1	378
1297	Familial segregation of a 5q15â€q21.2 deletion associated with facial dysmorphism and speech delay. Clinical Case Reports (discontinued), 2019, 7, 1154-1160.	0.2	4
1298	Towards a Rational and Efficient Diagnostic Approach in Children Referred for Growth Failure to the General Paediatrician. Hormone Research in Paediatrics, 2019, 91, 223-240.	0.8	37
1299	Molecular Characterization of a Familial 13.6-Mb 20p11.1p12.1 Duplication without Clinical Consequence. Cytogenetic and Genome Research, 2019, 157, 141-147.	0.6	0
1300	New genetic testing technologies: Advantages and limitations. South African Medical Journal, 2019, 109, 207.	0.2	6
1301	Multi-gene testing in neurological disorders showed an improved diagnostic yield: data from over 1000 Indian patients. Journal of Neurology, 2019, 266, 1919-1926.	1.8	26
1302	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	3.6	42
1303	Genotypeâ€“phenotype associations in children with copy number variants associated with high neuropsychiatric risk in the UK (IMAGINE-ID): a case-control cohort study. Lancet Psychiatry,the, 2019, 6, 493-505.	3.7	87
1304	Challenges in the clinical interpretation of small de novo copy number variants in neurodevelopmental disorders. Gene, 2019, 706, 162-171.	1.0	9
1305	Fifteen-minute consultation: Efficient investigation of the child with early developmental impairment in the era of genomic sequencing. Archives of Disease in Childhood: Education and Practice Edition, 2019, 105, edpract-2018-315123.	0.3	2
1306	Intellectual disability and autism spectrum disorders â€˜on the flyâ€™: insights from Drosophila. DMM Disease Models and Mechanisms, 2019, 12, .	1.2	38
1307	Improving access to genetic testing for adults with intellectual disability: A literature review and lessons from a quality improvement project in East London. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2019, 180, 566-575.	1.1	6
1308	The frequency of CNV s in a cohort population of consecutive fetuses with congenital anomalies after the termination of pregnancy. Molecular Genetics & Genomic Medicine, 2019, 7, e658.	0.6	3

#	Article	IF	Citations
1309	Costâ€effectiveness of five prenatal screening strategies for trisomies and other unbalanced chromosomal abnormalities: modelâ€based analysis. Ultrasound in Obstetrics and Gynecology, 2019, 54, 596-603.	0.9	2
1310	Mutations in PIEZO2 contribute to Gordon syndrome, Mardenâ€Walker syndrome and distal arthrogryposis: A bioinformatics analysis of mechanisms. Experimental and Therapeutic Medicine, 2019, 17, 3518-3524.	0.8	10
1311	What do parents expect from a genetic diagnosis of their child with intellectual disability?. Journal of Applied Research in Intellectual Disabilities, 2019, 32, 1129-1137.	1.3	8
1312	Copy Number Variations Causing Epilepsy. , 2019, , 398-405.		0
1313	GATAD2B Gene Microdeletion Causing Intellectual Disability Autosomal Dominant Type 18: Case Report and Review of the Literature. Molecular Syndromology, 2019, 10, 186-194.	0.3	2
1314	Case for genome sequencing in infants and children with rare, undiagnosed or genetic diseases. Journal of Medical Genetics, 2019, 56, 783-791.	1.5	93
1315	Truncating biallelic variant in DNAJA1, encoding the co-chaperone Hsp40, is associated with intellectual disability and seizures. Neurogenetics, 2019, 20, 109-115.	0.7	3
1316	Phenotype comparison among individuals with developmental delay/intellectual disability with or without genomic imbalances. Journal of Intellectual Disability Research, 2019, 63, 1379-1389.	1.2	4
1317	Experience of chromosomal microarray applied in prenatal and postnatal settings in Hong Kong. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 196-207.	0.7	29
1318	Schizophrenia is a laterâ€onset feature of <scp>PCDH</scp>19 Girls Clustering Epilepsy. Epilepsia, 2019, 60, 429-440.	2.6	23
1319	Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly. Genetics in Medicine, 2019, 21, 2043-2058.	1.1	57
1320	Advances in identification of genes involved in autosomal recessive intellectual disability: a brief review. Journal of Medical Genetics, 2019, 56, 567-573.	1.5	15
1321	Molecular cytogenetic identification of small supernumerary marker chromosomes using chromosome microarray analysis. Molecular Cytogenetics, 2019, 12, 13.	0.4	9
1322	Refining the Phenotype of Recurrent Rearrangements of Chromosome 16. International Journal of Molecular Sciences, 2019, 20, 1095.	1.8	34
1323	Pretransplant HLA typing revealed loss of heterozygosity in the major histocompatibility complex in a patient with acute myeloid leukemia. Human Immunology, 2019, 80, 257-262.	1.2	9
1324	Diagnostic implications of genetic copy number variation in epilepsy plus. Epilepsia, 2019, 60, 689-706.	2.6	61
1325	Consensus Statement of the Indian Academy of Pediatrics on Diagnosis and Management of Fragile X Syndrome in India. Indian Pediatrics, 2019, 56, 221-228.	0.2	5
1326	Single Molecule and Single Cell Sequencing. Advances in Experimental Medicine and Biology, 2019, , .	0.8	7

#	Article	IF	Citations
1327	Bionano Genome Mapping: High-Throughput, Ultra-Long Molecule Genome Analysis System for Precision Genome Assembly and Haploid-Resolved Structural Variation Discovery. Advances in Experimental Medicine and Biology, 2019, 1129, 97-118.	0.8	41
1328	Clinical application of next-generation sequencing to the practice of neurology. Lancet Neurology, The, 2019, 18, 492-503.	4.9	76
1329	Diagnostic Utility of Genome-wide DNA Methylation Testing in Genetically Unsolved Individuals with Suspected Hereditary Conditions. American Journal of Human Genetics, 2019, 104, 685-700.	2.6	125
1331	Neurodevelopmental Disorders and Array-Based Comparative Genomic Hybridization: Sensitivity and Specificity using a Criteria Checklist for Genetic Test Performance. Neuropediatrics, 2019, 50, 164-169.	0.3	2
1332	The utilization of prenatal microarray: A survey of current genetic counseling practices and barriers. Prenatal Diagnosis, 2019, 39, 351-360.	1.1	9
1333	Rare copy number variations affecting the synaptic gene DMXL2 in neurodevelopmental disorders. Journal of Neurodevelopmental Disorders, 2019, 11, 3.	1.5	6
1334	Altered Urinary Amino Acids in Children With Autism Spectrum Disorders. Frontiers in Cellular Neuroscience, 2019, 13, 7.	1.8	40
1335	Autism, spectrum or clusters? An EEG coherence study. BMC Neurology, 2019, 19, 27.	0.8	15
1336	Genetics of Intellectual Disability. , 2019, , 12-27.		1
1337	Complex Physical Health Issues in People with Intellectual Disability. , 2019, , 83-101.		1
1338	The Genetics of Autism Spectrum Disorders. , 2019, , 112-128.		14
1339	Diagnostic and service impact of genomic testing technologies in a neonatal intensive care unit. Journal of Paediatrics and Child Health, 2019, 55, 1309-1314.	0.4	11
1340	Cytogenomic Microarray Testing. , 2019, , .		0
1341	The Mouse, a Model Organism for Biomedical Research. , 2019, , 119-140.		6
1342	Estimating the effect size of the 15Q11.2 BP1â€“BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. Journal of Medical Genetics, 2019, 56, 701-710.	1.5	43
1343	ANALYSIS OF GENE COPY NUMBER VARIATIONS IN PATIENTS WITH CARDIAC SEPTAL DEFECTS USING MULTIPLEX LIGATION-DEPENDENT PROBE AMPLIFICATION. Asian Journal of Pharmaceutical and Clinical Research, 2019, , 100-103.	0.3	0
1344	Duplication of 1q31.3q41 in two affected siblings due to paternal insertional translocation. BMJ Case Reports, 2019, 12, e230941.	0.2	1
1345	Critical exon indexing improves clinical interpretation of copy number variants in neurodevelopmental disorders. Neurology: Genetics, 2019, 5, e378.	0.9	4

#	Article	IF	Citations
1346	Les normes de lâ€™Ã©valuation diagnostique du trouble du spectre de lâ€™autisme. Paediatrics and Child Health, 2019, 24, 452-460.	0.3	2
1347	Evaluation of the Genetic Association Between Adult Obesity and Neuropsychiatric Disease. Diabetes, 2019, 68, 2235-2246.	0.3	7
1348	Noninvasive prenatal testing for chromosome aneuploidies and subchromosomal microdeletions/microduplications in a cohort of 42,910 single pregnancies with different clinical features. Human Genomics, 2019, 13, 60.	1.4	78
1349	Chromosomal Aberrations in Pediatric Patients with Developmental Delay/Intellectual Disability: A Single-Center Clinical Investigation. BioMed Research International, 2019, 2019, 1-16.	0.9	18
1350	Molecular Etiology Disclosed by Array CGH in Patients With Silverâ€“Russell Syndrome or Similar Phenotypes. Frontiers in Genetics, 2019, 10, 955.	1.1	11
1351	Rethinking what constitutes a diagnosis in the genomics era: a critical illness perspective. Current Opinion in Pediatrics, 2019, 31, 317-321.	1.0	3
1352	A Retrospective Analysis of 10-Year Data Assessed the Diagnostic Accuracy and Efficacy of Cytogenomic Abnormalities in Current Prenatal and Pediatric Settings. Frontiers in Genetics, 2019, 10, 1162.	1.1	15
1353	Predictive impact of rare genomic copy number variations in siblings of individuals with autism spectrum disorders. Nature Communications, 2019, 10, 5519.	5.8	28
1354	Diagnosis of intellectual disability/global developmental delay via genetic analysis in a central region of China. Chinese Medical Journal, 2019, 132, 1533-1540.	0.9	8
1355	Application of next-generation sequencing for the diagnosis of fetuses with congenital heart defects. Current Opinion in Obstetrics and Gynecology, 2019, 31, 132-138.	0.9	3
1356	Prenatal diagnosis of a 5q35.3 microduplication involving part of the ADAMTS2 locus: a likely benign variant without apparent phenotypic abnormality. Medicine (United States), 2019, 98, e18258.	0.4	1
1357	Clinical utility of genomic sequencing. Current Opinion in Pediatrics, 2019, 31, 732-738.	1.0	14
1358	Role of late amniocentesis in the era of modern genomic technologies. Ultrasound in Obstetrics and Gynecology, 2019, 53, 676-685.	0.9	22
1359	Persistent Lin28 Expression Impairs Neurite Outgrowth and Cognitive Function in the Developing Mouse Neocortex. Molecular Neurobiology, 2019, 56, 3780-3795.	1.9	9
1360	Views from the clinic: Healthcare provider perspectives on whole genome sequencing in paediatrics. European Journal of Medical Genetics, 2019, 62, 350-356.	0.7	11
1361	Estimating the burden and economic impact of pediatric genetic disease. Genetics in Medicine, 2019, 21, 1781-1789.	1.1	59
1362	Next Generation Sequencing of Prenatal Structural Chromosomal Rearrangements Using Large-Insert Libraries. Methods in Molecular Biology, 2019, 1885, 251-265.	0.4	0
1363	Growth hormone therapy in children; research and practice â€“ A review. Growth Hormone and IGF Research, 2019, 44, 20-32.	0.5	52

#	Article	IF	Citations
1364	Analysis of chromosome microstructures in products of conception associated with recurrent miscarriage. Reproductive BioMedicine Online, 2019, 38, 787-795.	1.1	12
1365	How many copies of GSTM1 and GSTT1 are associated with head and neck cancer risk?. Biomarkers, 2019, 24, 262-267.	0.9	2
1366	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. Nature Genetics, 2019, 51, 117-127.	9.4	144
1367	Diagnostic approach with genetic tests for global developmental delay and/or intellectual disability: Single tertiary center experience. Annals of Human Genetics, 2019, 83, 115-123.	0.3	14
1368	Development of Criteria for Epilepsy Genetic Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2019, 46, 7-13.	0.3	18
1369	Mutations in glucokinase and other genes detected in neonatal and type 1B diabetes patient using whole exome sequencing may lead to disease-causing changes in protein activity. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 428-433.	1.8	7
1370	Chromosomal Basis of Inheritance. , 2019, , 237-265.		0
1371	Diagnostic Yield of Intellectual Disability Gene Panels. Pediatric Neurology, 2019, 92, 32-36.	1.0	21
1372	Evaluation for Genetic Disorders in the Absence of a Clinical Indication for Testing. Journal of Molecular Diagnostics, 2019, 21, 3-12.	1.2	21
1373	Chromosomal microarray analysis in fetuses with congenital anomalies of the kidney and urinary tract: A prospective cohort study and metaâ€analysis. Prenatal Diagnosis, 2019, 39, 165-174.	1.1	18
1374	Autism spectrum disorders: autistic phenotypes and complicated mechanisms. World Journal of Pediatrics, 2019, 15, 17-25.	0.8	12
1375	Array-CGH increased the diagnostic rate of developmental delay or intellectual disability in Taiwan. Pediatrics and Neonatology, 2019, 60, 453-460.	0.3	8
1376	Diagnostic yield of genetic tests in epilepsy. Neurology, 2019, 92, .	1.5	102
1377	Genome-Wide Copy Number Variation Detection Using NGS: Data Analysis and Interpretation. Methods in Molecular Biology, 2019, 1908, 113-124.	0.4	17
1378	Blockchain, Digital Identity, E-government. , 2019, , 233-258.		15
1379	Identifying Genomic Variations in Monozygotic Twins Discordant for Autism Spectrum Disorder Using Whole-Genome Sequencing. Molecular Therapy - Nucleic Acids, 2019, 14, 204-211.	2.3	40
1381	Retrospectively investigating the 12-year experience of prenatal diagnosis of small supernumerary marker chromosomes through array comparative genomic hybridization. Taiwanese Journal of Obstetrics and Gynecology, 2019, 58, 139-144.	0.5	5
1382	Chromosomal Microarray Analysis as a First-Tier Clinical Diagnostic Test in Patients With Developmental Delay/Intellectual Disability, Autism Spectrum Disorders, and Multiple Congenital Anomalies: A Prospective Multicenter Study in Korea. Annals of Laboratory Medicine, 2019, 39, 299-310.	1.2	44

#	Article	IF	Citations
1383	Locus and allelic heterogeneity in five families with hereditary spastic paraplegia. Journal of Human Genetics, 2019, 64, 17-21.	1.1	7
1384	Cytogenetic Analysis. , 2019, , 145-164.		0
1385	Nature and Frequency of Genetic Disease. , 2019, , 47-51.		7
1386	Chromosomal Microarray Analysis Using Array Comparative Genomic Hybridization on DNA from Amniotic Fluid and Chorionic Villus Sampling. Methods in Molecular Biology, 2019, 1885, 171-186.	0.4	1
1387	Neurodevelopmental Disabilities. , 2019, , 61-79.		0
1389	The Genomic Health of Human Pluripotent Stem Cells: Genomic Instability and the Consequences on Nuclear Organization. Frontiers in Genetics, 2018, 9, 623.	1.1	28
1390	Evaluation of copy number variant detection from panelâ€based nextâ€generation sequencing data. Molecular Genetics & Genomic Medicine, 2019, 7, e00513.	0.6	35
1391	Chromosomal Microarrays and Exome Sequencing for Diagnosis of Fetal Abnormalities. , 2019, , 577-595.		1
1392	Next-Generation Sequencing for Gene Panels and Clinical Exomes. , 2019, , 553-575.		1
1393	European guidelines for constitutional cytogenomic analysis. European Journal of Human Genetics, 2019, 27, 1-16.	1.4	108
1394	Application of chromosome microarray analysis in patients with unexplained developmental delay/intellectual disability in South China. Pediatrics and Neonatology, 2019, 60, 35-42.	0.3	21
1395	Establishment of a simple and rapid method to detect MECP2 duplications using digital polymerase chain reaction. Congenital Anomalies (discontinued), 2020, 60, 10-14.	0.3	4
1396	Genetic counselling and testing in adults with congenital heart disease: A consensus document of the ESC Working Group of Grown-Up Congenital Heart Disease, the ESC Working Group on Aorta and Peripheral Vascular Disease and the European Society of Human Genetics. European Journal of Preventive Cardiology, 2020, 27, 1423-1435.	0.8	38
1397	CNTN6 copy number variations: Uncertain clinical significance in individuals with neurodevelopmental disorders. European Journal of Medical Genetics, 2020, 63, 103636.	0.7	6
1398	Exome sequencing in patients with antiepileptic drug exposure and complex phenotypes. Archives of Disease in Childhood, 2020, 105, 384-389.	1.0	3
1399	Diagnostic interpretation of genetic studies in patients with primary immunodeficiency diseases: AÂworking group report of the Primary Immunodeficiency Diseases Committee of the American Academy of Allergy, Asthma & Immunology. Journal of Allergy and Clinical Immunology, 2020, 145, 46-69.	1.5	54
1400	BACs-on-Beadsâ„¢ assay, a rapid aneuploidy test, improves the diagnostic yield of conventional karyotyping. Molecular Biology Reports, 2020, 47, 169-177.	1.0	4
1401	First report of THOC6 related intellectual disability (Beaulieu Boycott Innes syndrome) in two siblings from India. European Journal of Medical Genetics, 2020, 63, 103742.	0.7	6

#	Article	IF	Citations
1402	Cytogenetics/cytogenomics. , 2020, , 525-539.		3
1403	Additive Diagnostic Yield of Homozygosity Regions Identified During Chromosomal microarray Testing in Children with Developmental Delay, Dysmorphic Features or Congenital Anomalies. Biochemical Genetics, 2020, 58, 74-101.	0.8	5
1404	Genome-Wide Meta-Analyses of FTND and TTFC Phenotypes. Nicotine and Tobacco Research, 2020, 22, 900-909.	1.4	17
1405	Novel missense variant in TTN cosegregating with familial atrioventricular block. European Journal of Medical Genetics, 2020, 63, 103752.	0.7	6
1406	Low-pass genome sequencing versus chromosomal microarray analysis: implementation in prenatal diagnosis. Genetics in Medicine, 2020, 22, 500-510.	1.1	64
1408	Metaâ€Analyses Support Previous and Novel Autism Candidate Genes: Outcomes of an Unexplored Brazilian Cohort. Autism Research, 2020, 13, 199-206.	2.1	25
1409	Modulation of hippocampal neuronal resilience during aging by the Hsp70/Hsp90 coâ€chaperone STI1. Journal of Neurochemistry, 2020, 153, 727-758.	2.1	16
1410	Diagnostic and Clinical Utility of Clinical Exome Sequencing in Children With Moderate and Severe Global Developmental Delay / Intellectual Disability. Journal of Child Neurology, 2020, 35, 116-131.	0.7	22
1411	Testing single/combined clinical categories on 5110 Italian patients with developmental phenotypes to improve arrayâ€based detection rate. Molecular Genetics & Genomic Medicine, 2020, 8, e1056.	0.6	6
1412	Evidence-Based Genetic Counseling for Psychiatric Disorders: A Road Map. Cold Spring Harbor Perspectives in Medicine, 2020, 10, a036608.	2.9	61
1413	Copy number variants in psychiatric disorders. , 2020, , 357-362.		1
1414	The genetic workup for structural congenital heart disease. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 178-186.	0.7	13
1415	Identification, Evaluation, and Management of Children With Autism Spectrum Disorder. Pediatrics, 2020, 145, .	1.0	621
1416	Chromosomal microarray analysis in prenatal diagnosis: ethical considerations of the Belgian approach. Journal of Medical Ethics, 2020, 46, 104-109.	1.0	11
1417	Muscular, Ocular and Brain Involvement Associated with a De Novo 11q13.2q14.1 Duplication: Contribution to the Differential Diagnosis of Muscle-Eye-Brain Congenital Muscular Dystrophy. Journal of Neuromuscular Diseases, 2020, 7, 69-76.	1.1	0
1418	Practice Parameter for the Assessment and Treatment of Psychiatric Disorders in Children and Adolescents With Intellectual Disability (Intellectual Developmental Disorder). Journal of the American Academy of Child and Adolescent Psychiatry, 2020, 59, 468-496.	0.3	25
1419	Variant interpretation is a component of clinical practice among genetic counselors in multiple specialties. Genetics in Medicine, 2020, 22, 785-792.	1.1	14
1421	Copy number alterations involving 59 <scp>ACMG</scp>â€recommended secondary findings genes. Clinical Genetics, 2020, 98, 577-588.	1.0	2

#	Article	IF	Citations
1422	EpiSigns. Advances in Molecular Pathology, 2020, 3, 29-39.	0.2	3
1423	Single nucleotide polymorphism array analysis of 102 patients with developmental delay and/or intellectual disability from Fujian, China. Clinica Chimica Acta, 2020, 510, 638-643.	0.5	2
1424	<scp>PPP1R21</scp>â€related syndromic intellectual disability: Report of an adult patient and review. American Journal of Medical Genetics, Part A, 2020, 182, 3014-3022.	0.7	8
1425	How geneticists think about Differences/Disorders of Sexual Development (DSD): A conversation. Journal of Pediatric Urology, 2020, 16, 760-767.	0.6	7
1426	Clinical features and genetic analysis of two Chinese families with X-linked ichthyosis. Journal of International Medical Research, 2020, 48, 030006052096229.	0.4	4
1427	Application of Chromosome Microarray Analysis in the Investigation of Developmental Disabilities and Congenital Anomalies: Single Center Experience and Review of <i>NRXN3</i> and <i>NEDD4L</i> Deletions. Molecular Syndromology, 2020, 11, 197-206.	0.3	6
1428	Genome Sequencing as a Diagnostic Test in Children With Unexplained Medical Complexity. JAMA Network Open, 2020, 3, e2018109.	2.8	47
1429	A six-attribute classification of geneticmosaicism. Genetics in Medicine, 2020, 22, 1743-1757.	1.1	34
1430	Clinical Genetic Screening in Adult Patients with Kidney Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2020, 15, 1497-1510.	2.2	53
1431	The Feasibility and Outcomes of Genetic Testing for Autism and Neurodevelopmental Disorders on an Inpatient Child and Adolescent Psychiatry Service. Autism Research, 2020, 13, 1450-1464.	2.1	6
1432	Polyhydramnios and abnormal foetal heart rate patterns in a foetus with Prader-Willi syndrome: A case report. Case Reports in Women's Health, 2020, 27, e00227.	0.2	4
1433	Utility of clinical exome sequencing in a complex Emirati pediatric cohort. Computational and Structural Biotechnology Journal, 2020, 18, 1020-1027.	1.9	14
1434	Genomic Diagnosis for Pediatric Disorders: Revolution and Evolution. Frontiers in Pediatrics, 2020, 8, 373.	0.9	30
1435	Global developmental delay and intellectual disability. , 2020, , 269-281.		0
1436	Loss-of-Function NUBPL Mutation May Link Parkinson's Disease to Recessive Complex I Deficiency. Frontiers in Neurology, 2020, 11, 555961.	1.1	5
1437	Genomic imbalances in craniofacial microsomia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 970-985.	0.7	8
1438	Diagnostic yield and treatment impact of wholeâ€genome sequencing in paediatric neurological disorders. Developmental Medicine and Child Neurology, 2021, 63, 934-938.	1.1	14
1439	Diagnostic power and clinical impact of exome sequencing in a cohort of 500 patients with rare diseases. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 955-964.	0.7	22

#	Article	IF	Citations
1440	Mutagenicity monitoring in humans: Global versus specific origin of mutations. Mutation Research - Reviews in Mutation Research, 2020, 786, 108341.	2.4	7
1441	Next Generation Sequencing and Bioinformatics Analysis of Family Genetic Inheritance. Frontiers in Genetics, 2020, 11, 544162.	1.1	41
1442	Mendelian, non-Mendelian, multigenic inheritance, and epigenetics. , 2020, , 3-25.		0
1443	Chorea in children: etiology, diagnostic approach and management. Journal of Neural Transmission, 2020, 127, 1323-1342.	1.4	19
1444	Utility of Chromosomal Microarray in Children with Unexplained Developmental Delay/Intellectual Disability. Fetal and Pediatric Pathology, 2022, 41, 208-218.	0.4	5
1445	SETD5 Gene Haploinsufficiency in Three Patients With Suspected KBG Syndrome. Frontiers in Neurology, 2020, 11, 631.	1.1	9
1446	Utility of the â€œomicsâ€•in kidney disease: Methods of analysis, sampling considerations, and technical approaches in renal biomarkers. , 2020, , 19-153.		0
1447	Automatic Identification of Down Syndrome Using Facial Images with Deep Convolutional Neural Network. Diagnostics, 2020, 10, 487.	1.3	26
1448	Optimizing Genetic Diagnosis of Neurodevelopmental Disorders in the Clinical Setting. Clinics in Laboratory Medicine, 2020, 40, 231-256.	0.7	11
1449	Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network. Molecular Genetics & Genomic Medicine, 2020, 8, e1397.	0.6	16
1450	A Comprehensive Clinical Genetics Approach to Critical Congenital Heart Disease in Infancy. Journal of Pediatrics, 2020, 227, 231-238.e14.	0.9	20
1451	Exon-focused targeted oligonucleotide microarray design increases detection of clinically relevant variants across multiple NHS genomic centres. Npj Genomic Medicine, 2020, 5, 28.	1.7	3
1452	Genetic background of ataxia in children younger than 5 years in Finland. Neurology: Genetics, 2020, 6, e444.	0.9	6
1453	Single-cell strand sequencing of a macaque genome reveals multiple nested inversions and breakpoint reuse during primate evolution. Genome Research, 2020, 30, 1680-1693.	2.4	16
1455	Based on a cohort of 52,879 microarrays, recurrent intragenic FBN2 deletion encompassing exons 1â€“8 does not cause Beals syndrome. European Journal of Medical Genetics, 2020, 63, 104008.	0.7	3
1456	Optic nerve coloboma as extension of the phenotype of 22q11.23 duplication syndrome: a case report. BMC Ophthalmology, 2020, 20, 333.	0.6	0
1457	Stickler Syndrome: A Review of Clinical Manifestations and the Genetics Evaluation. Journal of Personalized Medicine, 2020, 10, 105.	1.1	37
1458	Breakpoint junction analysis for complex genomic rearrangements with the caldera volcanoâ€•like pattern. Human Mutation, 2020, 41, 2119-2127.	1.1	2

#	Article	IF	Citations
1459	Communityâ€based recruitment and exome sequencing indicates high diagnostic yield in adults with intellectual disability. Molecular Genetics & Genomic Medicine, 2020, 8, e1439.	0.6	6
1460	Peruvian Newborn Male with 3p13 Deletion Syndrome Encompassing the FOXP1 Gene: Review of the Literature. Journal of Pediatric Genetics, 2020, 09, 270-278.	0.3	2
1461	Should We Report 15q11.2 BP1-BP2 Deletions and Duplications in the Prenatal Setting?. Journal of Clinical Medicine, 2020, 9, 2602.	1.0	8
1462	A diagnostic approach to syndromic retinal dystrophies with intellectual disability. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 538-570.	0.7	7
1463	Implementation of chromosomal microarrays in a cohort of patients with intellectual disability at the Argentinean public health system. Molecular Biology Reports, 2020, 47, 6863-6878.	1.0	2
1464	Relationship between Clinical Parameters and Chromosomal Microarray Data in Infants with Developmental Delay. Healthcare (Switzerland), 2020, 8, 305.	1.0	1
1465	Genomic Testing for Diagnosis of Genetic Disorders in Children: Chromosomal Microarray and Nextâ€"Generation Sequencing. Indian Pediatrics, 2020, 57, 549-554.	0.2	5
1466	Limited diagnostic impact of duplications <1 Mb of uncertain clinical significance: a 10-year retrospective analysis of reporting practices at the Mayo Clinic. Genetics in Medicine, 2020, 22, 2120-2124.	1.1	2
1467	Identification of a likely pathogenic structural variation in the LAMA1 gene by Bionano optical mapping. Npj Genomic Medicine, 2020, 5, 31.	1.7	14
1468	The Reproductive Journey in the Genomic Era: From Preconception to Childhood. Genes, 2020, 11, 1521.	1.0	7
1469	å…¨åŸºå›é«”å®šåºå°å…’ç«¥ç¥žç¶“ç–¾ç—……çš„è¨ºæ–·çŽ‡å’Œæ²»ç™‚å½±éŸ¿. Developmental Medicine and Child Neurology, 2020, 63,	1.1	
1470	Expanding clinical genetics services in a rural state in the post-genomic, technology-connected age: A dispatch from Mississippi. Translational Science of Rare Diseases, 2020, 4, 169-177.	1.6	1
1471	Identification of Neuropsychiatric Copy Number Variants in a Health Care System Population. JAMA Psychiatry, 2020, 77, 1276.	6.0	46
1472	Who ever heard of 16p11.2 deletion syndrome? Parentsâ€™ perspectives on a susceptibility copy number variation syndrome. European Journal of Human Genetics, 2020, 28, 1196-1204.	1.4	10
1473	The diagnostic yield of intellectual disability: combined whole genome low-coverage sequencing and medical exome sequencing. BMC Medical Genomics, 2020, 13, 70.	0.7	11
1474	Recent Advances in Understanding the Genetic Architecture of Autism. Annual Review of Genomics and Human Genetics, 2020, 21, 289-304.	2.5	30
1475	Established and Novel Mechanisms Leading to de novo Genomic Rearrangements in the Human Germline. Cytogenetic and Genome Research, 2020, 160, 167-176.	0.6	22
1476	Reinterpretation of Chromosomal Microarrays with Detailed Medical History. Journal of Pediatrics, 2020, 222, 180-185.e1.	0.9	4

#	Article	IF	Citations
1477	Phenotype and growth in Sotos syndrome patient from DR Congo (Central Africa). American Journal of Medical Genetics, Part A, 2020, 182, 1572-1575.	0.7	4
1478	Diagnostic Utility of Array Comparative Genomic Hybridization in Children with Neurological Diseases. Fetal and Pediatric Pathology, 2022, 41, 68-76.	0.4	0
1479	Taking consent for neonatal microarray analysis as a screen for genomic rearrangements: are paediatricians equipped for the genomic era?. Archives of Disease in Childhood, 2020, 105, 1021-1022.	1.0	1
1480	Low-Pass Genome Sequencing. Journal of Molecular Diagnostics, 2020, 22, 823-840.	1.2	35
1481	Low-pass genome sequencing: a validated method in clinical cytogenetics. Human Genetics, 2020, 139, 1403-1415.	1.8	31
1482	Chromosomal Microarray Analysis Has a Poor Diagnostic Yield in Children with Developmental Delay/Intellectual Disability When Concurrent Cerebellar Anomalies Are Present. Cerebellum, 2020, 19, 629-635.	1.4	3
1483	Genetic testing strategies in the newborn. Journal of Perinatology, 2020, 40, 1007-1016.	0.9	9
1484	A structural variation reference for medical and population genetics. Nature, 2020, 581, 444-451.	13.7	614
1485	Genetic testing on products of conception and its relationship with body mass index. Journal of Assisted Reproduction and Genetics, 2020, 37, 1853-1860.	1.2	4
1486	Phenotypes Associated with 16p11.2 Copy Number Gains and Losses at a Single Institution. Laboratory Medicine, 2020, 51, 642-648.	0.8	1
1487	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	1.1	36
1488	Management of Sleep Disturbances Associated with Smith-Magenis Syndrome. CNS Drugs, 2020, 34, 723-730.	2.7	5
1489	Pediatric Cataract Surgery and IOL Implantation. , 2020, , .		2
1491	Efficiency of noninvasive prenatal testing for the detection of fetal microdeletions and microduplications in autosomal chromosomes. Molecular Genetics & Genomic Medicine, 2020, 8, e1339.	0.6	19
1492	Genetics and pediatric hospital admissions, 1985 to 2017. Genetics in Medicine, 2020, 22, 1777-1785.	1.1	13
1493	A chromosome 1q22 microdeletion including ASH1L is associated with intellectual disability in a Chinese family. Molecular Cytogenetics, 2020, 13, 20.	0.4	8
1494	Parallel Tests of Whole Exome Sequencing and Copy Number Variant Sequencing Increase the Diagnosis Yields of Rare Pediatric Disorders. Frontiers in Genetics, 2020, 11, 473.	1.1	8
1495	Re-examining the Ethics of Genetic Counselling in the Genomic Era. Journal of Bioethical Inquiry, 2020, 17, 325-335.	0.9	11

#	Article	IF	Citations
1496	Mutated NUP188 and Other Nucleoporins as Gateways to Developmental Syndromes. Molecular Syndromology, 2020, 11, 1-3.	0.3	0
1497	â€œWhat if There's Something Wrong with Her?â€â€How Biomedical Technologies Contribute to Epistemic Injustice in Healthcare. Southern Journal of Philosophy, 2020, 58, 161-185.	0.4	5
1498	Appropriateness of arrayâ€CGH in the ADHD clinics: A comparative study. Genes, Brain and Behavior, 2020, 19, e12651.	1.1	4
1499	Whole genome analysis identifies the association of TP53 genomic deletions with lower survival in Stage III colorectal cancer. Scientific Reports, 2020, 10, 5009.	1.6	8
1500	Dysmorphology in the Era of Genomic Diagnosis. Journal of Personalized Medicine, 2020, 10, 18.	1.1	7
1501	Systematic evidence-based review: outcomes from exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability. Genetics in Medicine, 2020, 22, 986-1004.	1.1	53
1502	A minimum estimate of the prevalence of 22q11 deletion syndrome and other chromosome abnormalities in a combined prenatal and postnatal cohort. Human Reproduction, 2020, 35, 694-704.	0.4	7
1503	Linking risk factors and outcomes in autism spectrum disorder: is there evidence for resilience?. BMJ, The, 2020, 368, l6880.	3.0	45
1505	A Preterm Infant with Multiple Anomalies Diagnosed with Atypical CHARGE Syndrome after a Novel CHD7 Variant Confirmed Using Whole-Genome Sequencing. Neonatology, 2020, 117, 374-379.	0.9	0
1506	Prenatal diagnosis and molecular cytogenetic characterization of three chromosomal abnormalities with favorable outcomes. Taiwanese Journal of Obstetrics and Gynecology, 2020, 59, 338-341.	0.5	0
1507	KIAA1109 gene mutation in surviving patients with Alkuraya-KuÄinskas syndrome: a review of literature. BMC Medical Genetics, 2020, 21, 136.	2.1	10
1508	Associated anomalies in cases with congenital clubfoot. American Journal of Medical Genetics, Part A, 2020, 182, 2027-2036.	0.7	4
1509	Professionalsâ€™ accounts of genetic testing in adoption: a qualitative study. Archives of Disease in Childhood, 2020, 105, 74-79.	1.0	3
1510	A Private 16q24.2q24.3 Microduplication in a Boy with Intellectual Disability, Speech Delay and Mild Dysmorphic Features. Genes, 2020, 11, 707.	1.0	10
1511	Whole Genome Low-Coverage Sequencing Concurrently Detecting Copy Number Variations and Their Underlying Complex Chromosomal Rearrangements by Systematic Breakpoint Mapping in Intellectual Deficiency/Developmental Delay Patients. Frontiers in Genetics, 2020, 11, 616.	1.1	6
1512	Further evidence of GABRA4 and TOP3B as autism susceptibility genes. European Journal of Medical Genetics, 2020, 63, 103876.	0.7	12
1513	High Detection Rate of Copy Number Variations Using Capture Sequencing Data: A Retrospective Study. Clinical Chemistry, 2020, 66, 455-462.	1.5	16
1514	Cardiofaciocutaneous syndrome with rare structural variant in DOCK8 gene associated with neurodevelopmental disorders. Clinical Case Reports (discontinued), 2020, 8, 539-544.	0.2	1

#	Article	IF	Citations
1515	Genomic Stability Testing of Pluripotent Stem Cells. Current Protocols in Stem Cell Biology, 2020, 52, e107.	3.0	5
1516	Factors Influencing Decisions About Prenatal Genetic Testing for Autism Among Mothers of Children with Autism Spectrum Disorders. Advances in Neurodevelopmental Disorders, 2020, 4, 190-198.	0.7	0
1517	An integrated analysis of rare CNV and exome variation in Autism Spectrum Disorder using the Infinium PsychArray. Scientific Reports, 2020, 10, 3198.	1.6	42
1518	Cytogenetic and molecular diagnostic testing associated with prenatal and postnatal birth defects. Birth Defects Research, 2020, 112, 293-306.	0.8	12
1519	Perceived utility of biological testing for autism spectrum disorder is associated with child and family functioning. Research in Developmental Disabilities, 2020, 100, 103605.	1.2	7
1520	Age dependent association of inbreeding with risk for schizophrenia in Egypt. Schizophrenia Research, 2020, 216, 450-459.	1.1	1
1521	Genetic Counseling in Neurodevelopmental Disorders. Cold Spring Harbor Perspectives in Medicine, 2020, 10, a036533.	2.9	20
1522	Cytogenetic Investigation in 136 Consecutive Stillbirths: Does the Tissue Type Affect the Success Rate of Chromosomal Microarray Analysis and Karyotype?. Fetal Diagnosis and Therapy, 2020, 47, 315-320.	0.6	3
1523	Chromosomal microarray analysis of infertile men with azoospermia factor microdeletions. Gene, 2020, 735, 144389.	1.0	8
1524	Identifying schizophrenia patients who carry pathogenic genetic copy number variants using standard clinical assessment: retrospective cohort study. British Journal of Psychiatry, 2020, 216, 275-279.	1.7	12
1525	Copy Number Variation and Clinical Outcomes in Patients With GermlinePTENMutations. JAMA Network Open, 2020, 3, e1920415.	2.8	19
1526	Clinical exome sequencing as the first-tier test for diagnosing developmental disorders covering both CNV and SNV: a Chinese cohort. Journal of Medical Genetics, 2020, 57, 558-566.	1.5	61
1527	Sex and genes, part 2: A biopsychosocial approach to assess and treat challenging sexual behavior in persons with intellectual disabilities including fragile X syndrome and 22q11.2 deletion syndrome. Behavioral Sciences and the Law, 2020, 38, 152-172.	0.6	4
1528	Detection of copy number variants with chromosomal microarray in 10Â377 pregnancies at a single laboratory. Acta Obstetricia Et Gynecologica Scandinavica, 2020, 99, 775-782.	1.3	9
1529	Ocular Findings in the 16p11.2 Microdeletion Syndrome: A Case Report and Literature Review. Case Reports in Pediatrics, 2020, 2020, 1-5.	0.2	4
1530	Translating insights from neuropsychiatric genetics and genomics for precision psychiatry. Genome Medicine, 2020, 12, 43.	3.6	53
1531	The phenotypeâ€driven computational analysis yields clinical diagnosis for patients with atypical manifestations of known intellectual disability syndromes. Molecular Genetics & Genomic Medicine, 2020, 8, e1263.	0.6	15
1532	Triple diagnosis of Wiedemannâ€Steiner, Waardenburg and DLG3â€related intellectual disability association found by WES: A case report. Journal of Gene Medicine, 2020, 22, e3197.	1.4	4

#	Article	IF	Citations
1533	Sudden Cardiac Death and Copy Number Variants: What Do We Know after 10 Years of Genetic Analysis?. Forensic Science International: Genetics, 2020, 47, 102281.	1.6	20
1534	Exons deletion of CNKSR2 gene identified in X-linked syndromic intellectual disability. BMC Medical Genetics, 2020, 21, 69.	2.1	6
1535	Universal chromosomal microarray analysis reveals high proportion of copyâ€number variants in lowâ€risk pregnancies. Ultrasound in Obstetrics and Gynecology, 2021, 57, 813-820.	0.9	18
1537	Adaptation and validation of the Genetic Counseling Outcome Scale for autism spectrum disorders and related conditions. Journal of Genetic Counseling, 2021, 30, 305-318.	0.9	7
1538	Diagnostic yield of additional exome sequencing after the detection of long continuous stretches of homozygosity (LCSH) in SNP arrays. Journal of Human Genetics, 2021, 66, 409-417.	1.1	3
1539	50 Years Ago in T J P. Journal of Pediatrics, 2021, 231, 109.	0.9	0
1540	The role of chromosomal microarray analysis among fetuses with normal karyotype and single system anomaly or nonspecific sonographic findings. Acta Obstetricia Et Gynecologica Scandinavica, 2021, 100, 235-243.	1.3	7
1541	Clinical Characteristics and Genotypeâ€“Phenotype Correlation in Children with KMT2E Gene-Related Neurodevelopmental Disorders: Report of Two New Cases and Review of Published Literature. Neuropediatrics, 2021, 52, 098-104.	0.3	8
1542	Prenatal chromosomal microarray analysis inÂ2466Âfetuses with ultrasonographic soft markers: aÂprospective cohort study. American Journal of Obstetrics and Gynecology, 2021, 224, 516.e1-516.e16.	0.7	35
1543	Cost-effectiveness of genome-wide sequencing for unexplained developmental disabilities and multiple congenital anomalies. Genetics in Medicine, 2021, 23, 451-460.	1.1	34
1544	Factors Affecting Family Compliance with Genetic Testing of Children Diagnosed with Autism Spectrum Disorder. Journal of Autism and Developmental Disorders, 2021, 51, 1201-1209.	1.7	10
1545	Clinical Utility of Next-Generation Sequencing for Developmental Disorders in the Rehabilitation Department: Experiences from a Single Chinese Center. Journal of Molecular Neuroscience, 2021, 71, 845-853.	1.1	5
1546	Absence of heterozygosity detected by singleâ€nucleotide polymorphism array in prenatal diagnosis. Ultrasound in Obstetrics and Gynecology, 2021, 57, 314-323.	0.9	8
1547	Medical Conditions Associated with Autism. , 2021, , 2837-2842.		0
1548	Rhabdoid Tumors. , 2021, , 55-76.		0
1549	Progestogens in Threatened Miscarriage. , 2021, , 55-68.		1
1550	Chromosomal aberrations as the cause of a complex phenotype in children with primary immunodeficiencies. Pediatric Hematology/Oncology and Immunopathology, 2021, 19, 62-67.	0.1	0
1551	Neuro-Ophthalmological Manifestations of Craniosynostosis: Current Perspectives. Eye and Brain, 2021, Volume 13, 29-40.	3.8	20

#	Article	IF	Citations
1552	Medical Evaluation in Autism. , 2021, , 2844-2847.		0
1553	Application of Next Generation Sequencing in Laboratory Medicine. Annals of Laboratory Medicine, 2021, 41, 25-43.	1.2	99
1554	SNP Array as a Tool for Prenatal Diagnosis of Congenital Heart Disease Screened by Echocardiography: Implications for Precision Assessment of Fetal Prognosis. Risk Management and Healthcare Policy, 2021, Volume 14, 345-355.	1.2	6
1555	Genetic Disease and Therapy. Annual Review of Pathology: Mechanisms of Disease, 2021, 16, 145-166.	9.6	21
1556	Karyotyping as the first genomic approach. , 2021, , 17-34.		3
1557	Prenatal Diagnosis by Chromosome Microarray Analysis, An Indian Experience. Journal of Obstetrics and Gynecology of India, 2021, 71, 156-167.	0.3	2
1559	A Genetics-First Approach to Dissecting the Heterogeneity of Autism: Phenotypic Comparison of Autism Risk Copy Number Variants. American Journal of Psychiatry, 2021, 178, 77-86.	4.0	62
1560	Chorea, Ballism, and Athetosis. , 2021, , 401-417.e9.		0
1561	Genomic disorders in the genomics era. , 2021, , 35-59.		1
1562	An 88.8-kb Novel Deletion of 19q13.2 Encompassing the ATP1A3Gene Detected by Array CGH in a Patient with Delayed Psychomotor Development, Generalized Hypotonia and Macrocephaly. Molecular Syndromology, 2021, 12, 234-239.	0.3	0
1563	Germline copy number variations in BRCA1/2 negative families: Role in the molecular etiology of hereditary breast cancer in Tunisia. PLoS ONE, 2021, 16, e0245362.	1.1	9
1564	The Role of the Reanalysis of Genetic Test Results in the Diagnosis of Dysmorphic Syndrome Caused by Inherited Xq24 Deletion including the UBE2A and CXorf56 Genes. Genes, 2021, 12, 350.	1.0	4
1565	ClinSV: clinical grade structural and copy number variant detection from whole genome sequencing data. Genome Medicine, 2021, 13, 32.	3.6	36
1566	Incidental finding of APC deletion in a child: double trouble or double chance? â€“ a case report. Italian Journal of Pediatrics, 2021, 47, 31.	1.0	5
1567	Genetic diagnoses in pediatric patients with epilepsy and comorbid intellectual disability. Epilepsy Research, 2021, 170, 106552.	0.8	5
1568	Genetic Testing in Neurodevelopmental Disorders. Frontiers in Pediatrics, 2021, 9, 526779.	0.9	90
1569	Diagnostic and clinical utility of nextâ€generation sequencing in children born with multiple congenital anomalies in the China neonatal genomes project. Human Mutation, 2021, 42, 434-444.	1.1	15
1570	Can Genomics Remove Uncertainty from Adoption? Social Workersâ€™ and Medical Advisorsâ€™ Accounts of Genetic Testing. British Journal of Social Work, 2022, 52, 719-737.	0.9	1

#	Article	IF	Citations
1572	Clinical epigenomics: genome-wide DNA methylation analysis for the diagnosis of Mendelian disorders. Genetics in Medicine, 2021, 23, 1065-1074.	1.1	88
1573	Genome sequencing broadens the range of contributing variants with clinical implications in schizophrenia. Translational Psychiatry, 2021, 11, 84.	2.4	16
1574	Chromosomal microarray detects genetic risks of neurodevelopmental disorders in newborns with congenital heart disease. Cardiology in the Young, 2021, 31, 1275-1282.	0.4	3
1575	Diagnostic yield of wholeâ€exome sequencing in nonâ€syndromic intellectual disability. Journal of Intellectual Disability Research, 2021, 65, 577-588.	1.2	11
1576	One healthy live birth after preimplantation genetic testing of a cryptic balanced translocation (9;13) in a family with cerebral palsy and glaucoma: a case report. BMC Medical Genomics, 2021, 14, 82.	0.7	1
1577	Higher chromosomal aberration frequency in products of conception from women older than 32 years old with diminished ovarian reserve undergoing IVF/ICSI. Aging, 2021, 13, 10128-10140.	1.4	8
1578	Whole genomeÂmapping and identification of single nucleotide polymorphismsÂofÂfour BangladeshiÂindividualsÂand their functional significance. BMC Research Notes, 2021, 14, 105.	0.6	1
1579	<scp>PLXNA2</scp> and <scp>LRRC40</scp> as candidate genes in autism spectrum disorder. Autism Research, 2021, 14, 1088-1100.	2.1	5
1580	CYP27A1-dependent anti-melanoma activity of limonoid natural products targets mitochondrial metabolism. Cell Chemical Biology, 2021, 28, 1407-1419.e6.	2.5	11
1582	A practical approach to the genomics of kidney disorders. Pediatric Nephrology, 2022, 37, 21-35.	0.9	11
1583	Cellâ€free DNA analysis of maternal blood in prenatal screening for chromosomal microdeletions and microduplications: a systematic review. Prenatal Diagnosis, 2021, 41, 1324-1331.	1.1	18
1584	Prenatal ultrasound findings in Koolenâ€de Vries foetuses: Central nervous system anomalies are frequent markers of this syndrome. Molecular Genetics & Genomic Medicine, 2021, 9, e1649.	0.6	2
1585	Comorbidities associated with genetic abnormalities in children with intellectual disability. Scientific Reports, 2021, 11, 6563.	1.6	8
1586	Intellectual Developmental Disabilities:. Delaware Journal of Public Health, 2021, 7, 6-8.	0.2	0
1587	Genetic and metabolic investigations for individuals with neurodevelopmental disorders: A survey of Canadian geneticists' practices. American Journal of Medical Genetics, Part A, 2021, 185, 1757-1766.	0.7	2
1588	Comparison of the diagnostic yield of aCGH and genome-wide sequencing across different neurodevelopmental disorders. Npj Genomic Medicine, 2021, 6, 25.	1.7	27
1589	Need for psychiatric phenotyping in patients with rare genetic disorders. European Child and Adolescent Psychiatry, 2021, 30, 327-329.	2.8	3
1591	Xp11.2 Duplication in Females: Unique Features of a Rare Copy Number Variation. Frontiers in Genetics, 2021, 12, 635458.	1.1	2

#	Article	IF	Citations
1592	Quando il disturbo del neurosviluppo ha un substrato genetico: un caso di sindrome di Kleefstra. Medico E Bambino Pagine Elettroniche, 2021, 24, 114-117.	0.0	0
1593	Massive parallel sequencing in a family with rectal cancer. Hereditary Cancer in Clinical Practice, 2021, 19, 23.	0.6	3
1594	Coâ€occurring nonâ€omphalocele and nonâ€gastroschisis anomalies among cases with congenital omphalocele and gastroschisis. American Journal of Medical Genetics, Part A, 2021, 185, 1954-1971.	0.7	0
1595	Towards a Change in the Diagnostic Algorithm of Autism Spectrum Disorders: Evidence Supporting Whole Exome Sequencing as a First-Tier Test. Genes, 2021, 12, 560.	1.0	18
1596	Avances genÃ³micos de la Ãºltima dÃ©cada y su influencia en el enfoque diagnÃ³stico de la discapacidad intelectual.. Revista De Neuro-psiquiatria, 2021, 84, 33-50.	0.0	1
1597	Parental stress and adjustment in the context of rare genetic syndromes: A scoping review. Journal of Intellectual Disabilities, 2022, 26, 522-544.	1.0	8
1598	Optimizing the Diagnostic Strategy to Identify Genetic Abnormalities in Miscarriage. Molecular Diagnosis and Therapy, 2021, 25, 351-359.	1.6	5
1599	The Relationship Between Clinical Phenotypes and Chromosomal Microdeletions/Duplications in Pediatric Neurology. Duzce Universitesi Tip FakÃ¼ltesi Dergisi, 0, , .	0.3	1
1600	Shared Neurodevelopmental Perturbations Can Lead to Intellectual Disability in Individuals with Distinct Rare Chromosome Duplications. Genes, 2021, 12, 632.	1.0	0
1601	Pharmacological inhibition of the primary endocannabinoid producing enzyme, DGL â€Î±, induces autism spectrum disorderâ€like and coâ€morbid phenotypes in adult C57BL/J mice. Autism Research, 2021, 14, 1375-1389.	2.1	11
1602	Population Genomic Screening for Genetic Etiologies of Neurodevelopmental/Psychiatric Disorders Demonstrates Personal Utility and Positive Participant Responses. Journal of Personalized Medicine, 2021, 11, 365.	1.1	6
1603	Detection of Disease-Causing SNVs/Indels and CNVs in Single Test Based on Whole Exome Sequencing: A Retrospective Case Study in Epileptic Encephalopathies. Frontiers in Pediatrics, 2021, 9, 635703.	0.9	9
1604	Clinical application of whole-exome sequencing: A retrospective, single-center study. Experimental and Therapeutic Medicine, 2021, 22, 753.	0.8	4
1605	Congenital chromoanagenesis in the routine postnatal chromosomal microarray analyses. American Journal of Medical Genetics, Part A, 2021, 185, 2335-2344.	0.7	1
1607	Transcriptome analysis of MBD5-associated neurodevelopmental disorder (MAND) neural progenitor cells reveals dysregulation of autism-associated genes. Scientific Reports, 2021, 11, 11295.	1.6	4
1608	Changes in empowerment and anxiety of patients and parents during genetic counselling for epilepsy. European Journal of Paediatric Neurology, 2021, 32, 128-135.	0.7	4
1609	Craniofacial features of 3q29 deletion syndrome: Application of nextâ€generation phenotyping technology. American Journal of Medical Genetics, Part A, 2021, 185, 2094-2101.	0.7	13
1610	A Simple Practical Guide to Genomic Diagnostics in a Pediatric Setting. Genes, 2021, 12, 818.	1.0	2

#	Article	IF	Citations
1611	Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey. Molecular Genetics & Genomic Medicine, 2021, 9, e1665.	0.6	11
1613	Demystifying genetic jargon in psychiatry. BJ Psych Advances, 0, , 1-14.	0.5	2
1614	Novel treatments for autism spectrum disorder based on genomics and systems biology. , 2022, 230, 107939.		19
1615	Chromosomal microarray analysis, including constitutional and neoplastic disease applications, 2021 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 1818-1829.	1.1	18
1616	Diagnostic genetic testing for neurodevelopmental psychiatric disorders: closing the gap between recommendation and clinical implementation. Current Opinion in Genetics and Development, 2021, 68, 1-8.	1.5	28
1617	A Novel Mutation in the Myosin Binding Protein C Gene in a Prader-Willi Syndrome Pedigree. Reproductive Sciences, 2021, 28, 2718-2722.	1.1	1
1618	Guidelines for genetic testing of muscle and neuromuscular junction disorders. Muscle and Nerve, 2021, 64, 255-269.	1.0	8
1619	What a finding of gene copy number variation can add to the diagnosis of developmental neuropsychiatric disorders. Current Opinion in Genetics and Development, 2021, 68, 18-25.	1.5	15
1620	The introduction of clinical genetic testing in Ethiopia: Experiences and lessons learned. American Journal of Medical Genetics, Part A, 2021, 185, 2995-3004.	0.7	0
1621	All for one and one for all: heterogeneity of genetic etiologies in neurodevelopmental psychiatric disorders. Current Opinion in Genetics and Development, 2021, 68, 71-78.	1.5	14
1622	Clinical Genetic Testing in Children with Kidney Disease. Childhood Kidney Diseases, 2021, 25, 14-21.	0.1	1
1623	Inherited unbalanced reciprocal translocation with 3q duplication and 5p deletion in a foetus revealed by cell-free foetal DNA (cffDNA) testing: a case report. European Journal of Medical Research, 2021, 26, 64.	0.9	0
1624	Novel interstitial 2q12.3q13 microdeletion predisposes to developmental delay and behavioral problems. Neurogenetics, 2021, 22, 195-206.	0.7	2
1625	A Curriculum for Genomic Education of Molecular Genetic Pathology Fellows. Journal of Molecular Diagnostics, 2021, 23, 1218-1240.	1.2	4
1627	Exome Sequencing in 200 Intellectual Disability/Autistic Patients: New Candidates and Atypical Presentations. Brain Sciences, 2021, 11, 936.	1.1	17
1628	<scp>PLXNA2</scp> as a candidate gene in patients with intellectual disability. American Journal of Medical Genetics, Part A, 2021, 185, 3859-3865.	0.7	6
1629	Genotype-Phenotype Analysis of 8q24.3 Duplication and 21q22.3 Deletion in a Chinese Patient and Literature Review. Public Health Genomics, 2021, 24, 1-11.	0.6	1
1630	Have maternal or paternal ages any impact on the prenatal incidence of genomic copy number variants associated with fetal structural anomalies?. PLoS ONE, 2021, 16, e0253866.	1.1	6

#	Article	IF	Citations
1631	Estimate of the contemporary live-birth prevalence of recurrent 22q11.2 deletions: a cross-sectional analysis from population-based newborn screening. CMAJ Open, 2021, 9, E802-E809.	1.1	65
1632	Enhancing the Impact of Genomics Research in Autism through Integration of Research Results into Routine Care Pathwaysâ€"A Case Series. Journal of Personalized Medicine, 2021, 11, 755.	1.1	0
1633	Performances of NIPT for copy number variations at different sequencing depths using the semiconductor sequencing platform. Human Genomics, 2021, 15, 41.	1.4	11
1634	Copy neutral absence of heterozygosity on chromosome 15 distal long arm: A surrogate marker for Praderâ€"Willi/Angelman syndromes?. Molecular Cytogenetics, 2021, 14, 37.	0.4	1
1635	Clinical Characteristics and Genetic Etiology of Children With Developmental Language Disorder. Frontiers in Pediatrics, 2021, 9, 651995.	0.9	8
1636	Prevalence of copy number variants (CNVs) and rhGH treatment efficacy in an Italian cohort of children born small for gestational age (SGA) with persistent short stature associated with a complex clinical phenotype. Journal of Endocrinological Investigation, 2021, , 1.	1.8	2
1637	Investigation on combined copy number variation sequencing and cytogenetic karyotyping for prenatal diagnosis. BMC Pregnancy and Childbirth, 2021, 21, 496.	0.9	7
1638	A Reassessment of Copy Number Variations in Congenital Heart Defects: Picturing the Whole Genome. Genes, 2021, 12, 1048.	1.0	6
1639	Optical genome mapping enables constitutional chromosomal aberration detection. American Journal of Human Genetics, 2021, 108, 1409-1422.	2.6	108
1640	An ethical analysis of divergent clinical approaches to the application of genetic testing for autism and schizophrenia. Human Genetics, 2021, , 1.	1.8	11
1641	A 343 Italian cohort of patients analysed with arrayâ€comparative genomic hybridization: unsolved problems and genetic counselling difficulties. Journal of Intellectual Disability Research, 2021, 65, 863-869.	1.2	1
1642	Diagnostic Usefulness of MLPA Techniques for Recurrent Copy Number Variants Detection in Global Developmental Delay/Intellectual Disability. International Journal of General Medicine, 2021, Volume 14, 4511-4515.	0.8	3
1643	Diagnosis and Workup of Intellectual Disability in Adults. Neurology: Clinical Practice, 2021, 11, 534-540.	0.8	3
1645	Clinical utility and costâ€effectiveness analysis of chromosome testing concomitant with chromosomal microarray of patients with constitutional disorders in a U.S. academic medical center. Journal of Genetic Counseling, 2021, , .	0.9	1
1646	Cytogenetics and Molecular Diagnostics. , 2021, , 25-30.		0
1647	Higher CNV Frequencies in Chromosome 14 of Girls With Turner Syndrome Phenotype. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e4935-e4955.	1.8	2
1648	Appropriate Clinical Genetic Testing of Hemochromatosis Type 2â€"4, Including Ferroportin Disease. The Application of Clinical Genetics, 2021, Volume 14, 353-361.	1.4	1
1649	Next-generation gene panel testing in adolescents and adults in a medical neuropsychiatric genetics clinic. Neurogenetics, 2021, 22, 313-322.	0.7	4

#	Article	IF	Citations
1650	Is Prenatal Diagnosis Necessary for Fetal Isolated Nasal Bone Absence or Hypoplasia?. International Journal of General Medicine, 2021, Volume 14, 4435-4441.	0.8	8
1651	Attention Deficit Hyperactivity and Autism Spectrum Disorders as the Core Symptoms of AUTS2 Syndrome: Description of Five New Patients and Update of the Frequency of Manifestations and Genotype-Phenotype Correlation. Genes, 2021, 12, 1360.	1.0	16
1652	Multiple Genetic Rare Variants in Autism Spectrum Disorders: A Single-Center Targeted NGS Study. Applied Sciences (Switzerland), 2021, 11, 8096.	1.3	0
1653	The current state of prenatal detection of genetic conditions in congenital heart defects. Translational Pediatrics, 2021, 10, 2157-2170.	0.5	6
1654	Application of Single Nucleotide Polymorphism Microarray in Prenatal Diagnosis of Fetuses with Central Nervous System Abnormalities. International Journal of General Medicine, 2021, Volume 14, 4239-4246.	0.8	4
1655	Genetic Testing for Heritable Cardiovascular Diseases in Pediatric Patients: A Scientific Statement From the American Heart Association. Circulation Genomic and Precision Medicine, 2021, 14, e000086.	1.6	43
1656	Cloud-based genomics pipelines for ophthalmology: reviewed from research to clinical practice. Modeling and Artificial Intelligence in Ophthalmology, 2021, 3, 101-140.	0.1	1
1657	Neurodevelopmental Disorders in Patients With Complex Phenotypes and Potential Complex Genetic Basis Involving Non-Coding Genes, and Double CNVs. Frontiers in Genetics, 2021, 12, 732002.	1.1	12
1658	Diagnostic yield of genetic testing in 324 infants with hypotonia. Clinical Genetics, 2021, 100, 752-757.	1.0	4
1659	Balanced at First Sight, but in Reality out of Balance. Molecular Syndromology, 2021, 12, 333-334.	0.3	2
1660	Chromosomal Microarray in Children With Developmental Delay: The Experience of a Tertiary Center in Korea. Frontiers in Pediatrics, 2021, 9, 690493.	0.9	7
1661	Case Report: An Atypical Angelman Syndrome Case With Obesity and Fulfilled Autism Spectrum Disorder Identified by Microarray. Frontiers in Genetics, 2021, 12, 755605.	1.1	0
1662	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila. American Journal of Human Genetics, 2021, 108, 1669-1691.	2.6	23
1663	Genomic Aberrations Associated with the Pathophysiological Mechanisms of Neurodevelopmental Disorders. Cells, 2021, 10, 2317.	1.8	6
1664	A Tiered Genetic Screening Strategy for the Molecular Diagnosis of Intellectual Disability in Chinese Patients. Frontiers in Genetics, 2021, 12, 669217.	1.1	3
1665	Genetic counseling for congenital heart disease â€“ Practice resource of the National Society of Genetic Counselors. Journal of Genetic Counseling, 2022, 31, 9-33.	0.9	9
1666	Diagnostic Considerations in the Epilepsiesâ€”Testing Strategies, Test Type Advantages, and Limitations. Neurotherapeutics, 2021, 18, 1468-1477.	2.1	4
1667	Developmental disabilities across the world: A scientometric review from 1936 to 2020. Research in Developmental Disabilities, 2021, 117, 104031.	1.2	20

#	Article	IF	Citations
1668	Chromosomal Microarray Analysis as First-Tier Genetic Test for Schizophrenia. Frontiers in Genetics, 2021, 12, 620496.	1.1	4
1669	Pretransplant HLA mistyping in diagnostic sample of a T-ALL patient due to loss of heterozygosity in the major histocompatibility complex. Transplant Immunology, 2021, 69, 101463.	0.6	1
1670	Reproductive genetics. , 2022, , 21-46.e3.		0
1671	Outcomes of prior authorization requests for genetic testing in outpatient pediatric genetics clinics. Genetics in Medicine, 2021, 23, 950-955.	1.1	13
1672	Genetic developmental disability diagnosed in adulthood: a case report. Journal of Medical Case Reports, 2021, 15, 28.	0.4	2
1673	Genome-wide analysis of gene dosage in 24,092 individuals estimates that 10,000 genes modulate cognitive ability. Molecular Psychiatry, 2021, 26, 2663-2676.	4.1	33
1674	Current approaches to genetic testingÂinÂpediatric disease. , 2021, , 15-36.		0
1675	First Report of a de novo 10q23.31q23.33 Microdeletion: Obesity, Intellectual Disability and Microcephaly. Molecular Syndromology, 2021, 12, 258-262.	0.3	0
1676	Artificial Intelligence in Blood Transcriptomics. , 2021, , 1-16.		0
1677	Prevalence and Phenotypic Impact of Robertsonian Translocations. Molecular Syndromology, 2021, 12, 1-11.	0.3	18
1678	Next-generation cytogenomics: High-resolution structural variation detection by optical genome mapping. , 2021, , 123-146.		1
1679	CGH Findings in Children with Complex and Essential Autistic Spectrum Disorder. Journal of Autism and Developmental Disorders, 2021, , 1.	1.7	2
1680	CNV profiles of Chinese pediatric patients with developmental disorders. Genetics in Medicine, 2021, 23, 669-678.	1.1	17
1682	DeepCNV: a deep learning approach for authenticating copy number variations. Briefings in Bioinformatics, 2021, 22, .	3.2	15
1683	Genetic Referral Patterns and Responses to Clinical Scenarios: A Survey of Primary Care Providers and Clinical Geneticists. Journal of Primary Care and Community Health, 2021, 12, 215013272110467.	1.0	11
1684	22q11.2 deletion syndrome and congenital heart disease. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 64-72.	0.7	66
1685	Prenatally detected copy number variants in a national cohort: A postnatal followâ€up study. Prenatal Diagnosis, 2020, 40, 1272-1283.	1.1	5
1686	Genomic Applications in Inherited Genetic Disorders. , 2015, , 535-551.		1

#	Article	IF	Citations
1687	Chromosome Microarrays in Diagnostic Testing: Interpreting the Genomic Data. Methods in Molecular Biology, 2014, 1168, 117-155.	0.4	4
1688	Molecular Basis of Cardiac Development and Congenital Heart Disease. , 2012, , 317-339.		2
1689	Overview and Introduction to Autism Spectrum Disorder (ASD). Advances in Neurobiology, 2020, 24, 3-42.	1.3	16
1690	Medical comorbidities in autism spectrum disorder. , 2015, , 33-41.		3
1691	Molecular Pathology Methods. , 2016, , 19-52.		3
1693	Molecular Diagnostic Techniques and Biotechnology in Plant Biosecurity. , 2014, , 375-416.		5
1694	A study of normal copy number variations in Israeli population. Human Genetics, 2021, 140, 553-563.	1.8	1
1695	Global Developmental Delay and Regression. , 2012, , 65-72.		1
1696	Clinical Neurogenetics. , 2012, , 704-734.		3
1697	Global Developmental Delay and Mental Retardation/Intellectual Disability. , 2012, , 554-574.		1
1698	State of the Art of Genetic Testing for Patients With Autism: A Practical Guide for Clinicians. Seminars in Pediatric Neurology, 2020, 34, 100804.	1.0	23
1700	A microcosting and costâ€“consequence analysis of clinical genomic testing strategies in autism spectrum disorder. Genetics in Medicine, 2017, 19, 1268-1275.	1.1	62
1701	Chromosomal microarray testing influences medical management. Genetics in Medicine, 2011, 13, 770-776.	1.1	107
1712	Diagnostic Approach to Microdeletion Syndromes Based on 22q11.2 Investigation: Challenges in Four Cases. Molecular Syndromology, 2017, 8, 244-252.	0.3	10
1713	Genetic Counselling Pitfall: Co-Occurrence of an 11.8-Mb Xp22 Duplication and an Xp21.2 Duplication Disrupting IL1RAPL1. Molecular Syndromology, 2017, 8, 325-330.	0.3	4
1714	Genetic basis of human congenital anomalies of the kidney and urinary tract. Journal of Clinical Investigation, 2018, 128, 4-15.	3.9	91
1715	A highly sensitive and specific workflow for detecting rare copy-number variants from exome sequencing data. Genome Medicine, 2020, 12, 14.	3.6	40
1716	Evaluation of the Child With Developmental Impairments. CONTINUUM Lifelong Learning in Neurology, 2018, 24, 228-247.	0.4	7

#	Article	IF	Citations
1717	Semiconductor Sequencing Analysis of Chromosomal Copy Number Variations in Spontaneous Miscarriage. Medical Science Monitor, 2017, 23, 5550-5557.	0.5	7
1718	A Key Role for the Genetic Counsellor in the Genomics Era. F1000Research, 2018, 7, 420.	0.8	2
1719	Array-Based Identification of Copy Number Changes in a Diagnostic Setting : Simultaneous Gene - Focused and Low Resolution Whole Human Genome Analysis. Sultan Qaboos University Medical Journal, 2013, 13, 69-79.	0.3	5
1720	Analytical and Clinical Validity Study of FirstStepDx PLUS: A Chromosomal Microarray Optimized for Patients with Neurodevelopmental Conditions. PLOS Currents, 2017, 9, .	1.4	9
1721	Uncovering Genomic Causes of Co-Morbidity in Epilepsy: Gene-Driven Phenotypic Characterization of Rare Microdeletions. PLoS ONE, 2011, 6, e23182.	1.1	24
1722	Genome-Wide Mapping of Copy Number Variation in Humans: Comparative Analysis of High Resolution Array Platforms. PLoS ONE, 2011, 6, e27859.	1.1	59
1723	High Proportion of 22q13 Deletions and SHANK3 Mutations in Chinese Patients with Intellectual Disability. PLoS ONE, 2012, 7, e34739.	1.1	43
1724	Identification of Rare Recurrent Copy Number Variants in High-Risk Autism Families and Their Prevalence in a Large ASD Population. PLoS ONE, 2013, 8, e52239.	1.1	69
1725	Genome Wide Analysis of Chromosomal Alterations in Oral Squamous Cell Carcinomas Revealed over Expression of MGAM and ADAM9. PLoS ONE, 2013, 8, e54705.	1.1	27
1726	Parental Imbalances Involving Chromosomes 15q and 22q May Predispose to the Formation of De Novo Pathogenic Microdeletions and Microduplications in the Offspring. PLoS ONE, 2013, 8, e57910.	1.1	7
1727	Simple, Rapid and Inexpensive Quantitative Fluorescent PCR Method for Detection of Microdeletion and Microduplication Syndromes. PLoS ONE, 2013, 8, e61328.	1.1	9
1728	Ultra High-Resolution Gene Centric Genomic Structural Analysis of a Non-Syndromic Congenital Heart Defect, Tetralogy of Fallot. PLoS ONE, 2014, 9, e87472.	1.1	16
1729	Whole-Genome Array CGH Evaluation for Replacing Prenatal Karyotyping in Hong Kong. PLoS ONE, 2014, 9, e87988.	1.1	41
1730	Detection of Chromosomal Breakpoints in Patients with Developmental Delay and Speech Disorders. PLoS ONE, 2014, 9, e90852.	1.1	41
1731	Screening for Intellectual Disability Using High-Resolution CMA Technology in a Retrospective Cohort from Central Brazil. PLoS ONE, 2014, 9, e103117.	1.1	12
1732	The Clinical Impact of Chromosomal Microarray on Paediatric Care in Hong Kong. PLoS ONE, 2014, 9, e109629.	1.1	20
1733	Copy Number Variants Associated with 14 Cases of Self-Injurious Behavior. PLoS ONE, 2016, 11, e0149646.	1.1	5
1734	Overview of Cytogenetic Technologies. Korean Journal of Clinical Laboratory Science, 2018, 50, 375-381.	0.1	4

#	Article	IF	Citations
1735	An Integrated Model of Emotions, Attitudes, and Intentions Associated with Undergoing Autism Genetic Testing. Psychology and Cognitive Sciences: Open Journal, 2015, 1, 29-38.	0.2	3
1736	Association of genes with phenotype in autism spectrum disorder. Aging, 2019, 11, 10742-10770.	1.4	23
1737	The intellectual developmental disorders Mexico study: situational diagnosis, burden, genomics and intervention proposal. Salud Publica De Mexico, 2016, 58, 694.	0.1	8
1738	NeuroArray: A Customized aCGH for the Analysis of Copy Number Variations in Neurological Disorders. Current Genomics, 2018, 19, 431-443.	0.7	3
1739	Genetic analysis of intellectual disability and autism. Acta Biomedica, 2020, 91, e2020003.	0.2	18
1740	Syndromic autism spectrum disorders: moving from a clinically defined to a molecularly defined approach. Dialogues in Clinical Neuroscience, 2017, 19, 353-371.	1.8	105
1741	Evaluation of Array Comparative genomic Hybridisation in prenatal diagnosis of fetal anomalies: a multicentre cohort study with cost analysis and assessment of patient, health professional and commissioner preferences for array comparative genomic hybridisation. Efficacy and Mechanism Evaluation, 2017, 4, 1-104.	0.9	23
1742	A scoping study to explore the cost-effectiveness of next-generation sequencing compared with traditional genetic testing for the diagnosis of learning disabilities in children. Health Technology Assessment, 2015, 19, 1-90.	1.3	23
1743	Interstitial deletion of 5q33.3q35.1 in a boy with severe mental retardation. Korean Journal of Pediatrics, 2016, 59, S19.	1.9	11
1744	Genetic tests by next-generation sequencing in children with developmental delay and/or intellectual disability. Clinical and Experimental Pediatrics, 2020, 63, 195-202.	0.9	29
1745	First Case of Peroxisomal D-bifunctional Protein Deficiency with Novel HSD17B4 Mutations and Progressive Neuropathy in Korea. Journal of Korean Medical Science, 2020, 35, e357.	1.1	6
1747	A Bibliometric Insight of Genetic Factors in ASD: Emerging Trends and New Developments. Brain Sciences, 2021, 11, 33.	1.1	11
1748	Genetics of Autism Spectrum Disorder: An Update. Psychiatric Annals, 2019, 49, 109-114.	0.1	4
1749	A novel CRX mutation by whole-exome sequencing in an autosomal dominant cone-rod dystrophy pedigree. International Journal of Ophthalmology, 2015, 8, 1112-7.	0.5	6
1750	What Should a Psychiatrist Know About Genetics?. Journal of Clinical Psychiatry, 2018, 80, .	1.1	40
1751	Recurrent benign copy number variants & issues in interpretation of variants of unknown significance identified by cytogenetic microarray in Indian patients with intellectual disability. Indian Journal of Medical Research, 2015, 142, 699.	0.4	4
1752	Application of chromosomal microarray for evaluation of idiopathic short stature in Asian Indian children: A pilot study. Indian Journal of Endocrinology and Metabolism, 2018, 22, 100.	0.2	10
1753	Clinical practice guidelines for assessment and management of intellectual disability. Indian Journal of Psychiatry, 2019, 61, 194.	0.4	20

#	Article	IF	Citations
1754	Genetic studies in chronic kidney disease: interpretation and clinical applicability. Journal of Nephrology, 2012, 25, 851-864.	0.9	7
1756	Molecular genetic methods in the diagnosis of myelodysplastic syndromes. A review. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2014, 158, 339-345.	0.2	11
1757	MEF2C-Related 5q14.3 Microdeletion Syndrome Detected by Array CGH: A Case Report. Annals of Rehabilitation Medicine, 2015, 39, 482.	0.6	6
1758	Patient With Delayed Development Resulting From De Novo Duplication of 7q36.1-q36.3 and Deletion of 9p24.3. Annals of Rehabilitation Medicine, 2017, 41, 881.	0.6	1
1759	Effects of Copy Number Variations on Developmental Aspects of Children With Delayed Development. Annals of Rehabilitation Medicine, 2019, 43, 215-223.	0.6	6
1760	Clinical Applications of Chromosomal Microarray Analysis. Journal of Genetic Medicine, 2010, 7, 111-118.	0.1	6
1761	First-Tier Array CGH in Clinically Variable Entity Diagnosis: 22q13.3 Deletion Syndrome. , 0, , .		1
1762	Chromosomal Microarray Testing in 42 Korean Patients with Unexplained Developmental Delay, Intellectual Disability, Autism Spectrum Disorders, and Multiple Congenital Anomalies. Genomics and Informatics, 2017, 15, 82-86.	0.4	4
1763	Next generation sequencing and array-based comparative genomic hybridization for molecular diagnosis of pediatric endocrine disorders. Annals of Pediatric Endocrinology and Metabolism, 2017, 22, 90.	0.8	4
1764	MLPA is a practical and complementary alternative to CMA for diagnostic testing in patients with autism spectrum disorders and identifying new candidate CNVs associated with autism. PeerJ, 2019, 6, e6183.	0.9	8
1765	Differences in the importance of microcephaly, dysmorphism, and epilepsy in the detection of pathogenic CNVs in ID and ASD patients. PeerJ, 2019, 7, e7979.	0.9	6
1766	Xia-Gibbs Syndrome: A Review of Literature. Cureus, 2020, 12, e12352.	0.2	7
1767	Compound Heterozygote of Point Mutation and Chromosomal Microdeletion Involving OTUD6B Coinciding with ZMIZ1 Variant in Syndromic Intellectual Disability. Genes, 2021, 12, 1583.	1.0	6
1768	Study protocol: childhood outcomes of fetal genomic variants: the PrenatAL Microarray (PALM) cohort study. BMC Pediatrics, 2021, 21, 447.	0.7	0
1769	Diagnostic yield of microarrays in individuals with nonâ€syndromic developmental delay and intellectual disability. Journal of Intellectual Disability Research, 2021, 65, 1033-1048.	1.2	6
1770	Identification of copy number variants in children and adolescents with autism spectrum disorder: a study from Turkey. Molecular Biology Reports, 2021, 48, 7371-7378.	1.0	1
1771	17q12 Recurrent Deletions and Duplications: Description of a Case Series with Neuropsychiatric Phenotype. Genes, 2021, 12, 1660.	1.0	6
1772	Descriptive and risk factor analysis of infantile cataracts: National Birth Defects Prevention Study, 2000â€“2011. American Journal of Medical Genetics, Part A, 2022, 188, 509-521.	0.7	2

#	Article	IF	Citations
1773	Update on Clinically Relevant Genetic Testing in Pediatric Dermatology. Dermatologic Clinics, 2021, 40, 1-8.	1.0	0
1774	Artificial intelligence enables comprehensive genome interpretation and nomination of candidate diagnoses for rare genetic diseases. Genome Medicine, 2021, 13, 153.	3.6	53
1775	Clinical and genetic profile of children with unexplained intellectual disability/developmental delay and epilepsy. Epilepsy Research, 2021, 177, 106782.	0.8	3
1777	Koolenâ€de Vries syndrome in a 63â€yearâ€old woman: Report of the oldest patient and a review of the adult phenotype. American Journal of Medical Genetics, Part A, 2021, , .	0.7	3
1778	Predictors of empowerment in parents of children with autism and related neurodevelopmental disorders who are undergoing genetic testing. Molecular Genetics & Genomic Medicine, 2021, 9, e1803.	0.6	2
1779	Applications of Noninvasive Prenatal Testing for Subchromosomal Copy Number Variations Using Cell-Free DNA. Advances in Molecular Pathology, 2021, 4, 17-25.	0.2	1
1780	Genetic Evaluation in Developmental Disabilities. , 2011, , 69-78.		0
1781	Evidence-Based Pathology and Laboratory Medicine in the Molecular Pathology Era: Transition of Tests from the Research Bench into Practice. , 2011, , 297-304.		0
1782	Evaluation for a genetic disease. , 2011, , 669-673.		0
1785	Array Comparative Genomic Hybridization in Cytogenetics and Molecular Genetics. , 2012, , 21-35.		0
1786	Development of LOINC for Integrating Constitutional Cytogenetic Test Result Reports into Electronic Health Records. European Journal for Biomedical Informatics, 0, , .	0.5	0
1788	Cytogenetic Testing and Chromosomal Disorders. , 2012, , 39-59.		0
1790	GenetinÄ—s diagnozÄ—s nustatymas esant intelektinei negaliai: phelan-mcdermid sindromo pavyzdys. Health Sciences, 2012, 22, 73-77.	0.0	0
1791	Malformation syndromes. , 2012, , 791-817.		0
1792	Array-CGH and SNP-Arrays, the New Karyotype. , 2012, , 39-52.		0
1793	Specific Chromosome Disorders in Newborns. , 2012, , 196-208.		1
1794	Fluorescence In Situ Hybridization (FISH). , 2013, , 415-439.		0
1795	Understanding the Value of Information from Pediatric Clinical Research. Paediatric Drugs, 2012, 14, 295-297.	1.3	0

#	Article	IF	Citations
1796	An Unusual Case of an Infant with Failure to Thrive. Pediatric Annals, 2012, 41, 502-505.	0.3	0
1799	The child with autism. Practical Pre-School, 2013, 2013, 16-17.	0.0	0
1801	CGH-based microarray detection of cryptic and novel copy number alterations and balanced translocations in cytogenetically abnormal cases of b-cell all. Health, 2013, 05, 23-40.	0.1	0
1803	Soto syndrome: a rare overgrowth disorder. Ceylon Medical Journal, 2013, 58, 40.	0.1	0
1804	Identifying Microdeletion Syndromes in Patients with Intellectual Disability Using Molecular Genetic Testing: An Example for the Brazilian Public Health Care System. American Journal of Public Health Research, 2013, 1, 86-92.	0.2	3
1805	Low-frequency Mosaicism of Trisomy 14, Missed by Array CGH. Journal of Genetic Medicine, 2013, 10, 52-56.	0.1	0
1807	Chromosome Microarrays. , 2014, , 149-165.		0
1808	SVEM: A Structural Variant Estimation Method Using Multi-mapped Reads on Breakpoints. Lecture Notes in Computer Science, 2014, , 208-219.	1.0	0
1809	Overgrowth Syndrome with 9q22.3 Microdeletion Detected by Microarray Comparative Genomic Hybridization. Neonatal Medicine, 2014, 21, 264.	0.1	0
1810	Detection of chromosomal imbalances using combined MLPA kits in patients with syndromic intellectual disability. Romanian Journal of Laboratory Medicine, 2014, 22, .	0.1	3
1811	Nature and Frequency of Genetic Disease. , 2014, , .		0
1813	Situations spÃ©cifiques. , 2014, , 171-242.		0
1814	Causes of Congenital Malformations. , 2014, , 105-164.		1
1818	GENETICS FOR DIAGNOSTICS IN PREVENTIVE MEDICINE. Cardiovascular Therapy and Prevention (Russian) Tj ETQq1 1 0.784314 rgBT	0.4	0
1820	Ethical Issues Raised by the Clinical Implementation of New Diagnostic Tools for Genetic Diseases in Children: Array Comparative Genomic Hybridization (aCGH) as a Case Study. Journal of Clinical Research & Bioethics, 2015, 06, .	0.2	0
1821	Copy Number Variation in Human Health, Disease and Evolution. , 2015, , 129-154.		4
1823	Genetics of Autism Spectrum Disorders: The Opportunity and Challenge in the Genetics Clinic. , 2015, , 33-66.		0
1824	Diagnostic approach for genetic causes of intellectual disability. Journal of Genetic Medicine, 2015, 12, 6-11.	0.1	1

#	Article	IF	Citations
1825	Clinical utility of next-generation sequencing in neurodevelopmental disorders: non-syndromic intellectual disability as a model. Istanbul Bilim University Florence Nightingale Journal of Medicine, 2015, 1, 52-57.	0.1	0
1829	CONTEMPORARY APPROACH TO DIAGNOSIS OF GENETIC CAUSES OF INTELLECTUAL DISABILITY. Journal of Special Education and Rehabilitation, 2016, 3-4, 62-62.	0.5	0
1830	Intellectual Disability and Developmental Delay: Cytogenetic Testing. , 2016, , 91-100.		0
1831	22q11.2 Deletion Syndrome: A Paradigmatic Copy-Number-Variant (CNV) Disorder. , 2016, , 723-730.		0
1832	Chromosomal Microarray. , 2016, , 2-6-2-9.		0
1833	Cytogenetics: Applications. , 2016, , 67-82.		0
1835	Personalized Medicine for the Treatment of Human Cancer. , 2017, , 843-855.		0
1836	Utilization Management of Genetic Testing. , 2017, , 219-233.		0
1837	Clinical Genetics and Heritable Parathyroid Disease: Monogenic Disorders. , 2017, , 423-432.		0
1838	Molecular Diagnosis of Epilepsy in Clinical Practice. Journal of the Korean Child Neurology Society, 2016, 24, 183-189.	0.0	0
1839	Oculofacial Manifestations of Chromosomal Aberrations. , 2017, , 109-170.		1
1840	In Situ Hybridization â˜†. , 2017, , .		0
1842	Genetic Diagnosis of Chromosomal Congenital Anomalies in Albanian Pediatric Patients by Array CGH. Open Access Macedonian Journal of Medical Sciences, 2017, 5, 587-591.	0.1	1
1844	Prenatal and Postnatal Genetic Testing: Why, How, and When?. Pediatric Annals, 2017, 46, e423-e427.	0.3	1
1845	Medical Conditions Associated with Autism. , 2018, , 1-5.		0
1846	Medical Evaluation in Autism. , 2018, , 1-4.		0
1848	Update in Clinical Genetics and Metabolics. , 2018, , 369-390.		0
1850	The Ethical Complexity of Using Whole-Exome Sequencing to Detect Adult-Onset Conditions in the Prenatal and Pediatric Settings. , 2018, , 25-35.		0

#	Article	IF	Citations
1851	SÃndrome de Phelan-McDermid: reporte de un caso y revisiÃ³n de la literatura. Acta Pediatrica De Mexico, 2018, 1, 42.	0.2	0
1854	The possible implications of advances in genetic testing technologies for Genetic Counsellors working with families of children with developmental disorders. F1000Research, 2018, 7, 420.	0.8	1
1856	Comparative genomic hybridisation as a first option in genetic diagnosis: 1000 cases and a costâ€“benefit analysis. Anales De PediatrÃa (English Edition), 2018, 89, 3-11.	0.1	0
1857	Genetic profile and clinical application of chromosomal microarray in children with intellectual disability in Hong Kong. Hong Kong Medical Journal, 2018, 24, 451-459.	0.1	3
1858	Attention Ã lâ€™attention. Lâ€™attention est-elle toujours bÃ©nÃ©fiqueÂ? Effets secondaires du dÃ©pistage prÃ©natal. , 2018, , 125-147.		1
1859	A Rare Case of 15q11.2 Microdeletion Syndrome with Atypical Features: Diagnostic Dilemma. Cureus, 2018, 10, e3543.	0.2	1
1861	Diagnostic distal 16p11.2 deletion in a preterm infant with facial dysmorphism. Journal of Genetic Medicine, 2018, 15, 115-119.	0.1	1
1863	Molecular Diagnostics in Cancer: A Fundamental Component of Precision Oncology. , 2019, , 5-31.		0
1864	Chromosomal Microarray in the New High-Throughput Technological and Bioinformatic Era. , 2019, , 61-84.		0
1865	Complex Genetics and the Etiology of Human Congenital Heart Disease. Cardiac and Vascular Biology, 2019, , 169-184.	0.2	0
1866	Genomic Architecture of ASD. , 2019, , 23-34.		0
1867	Associations between congenital heart defects and genetic and morphological anomalies. The importance of prenatal screening. Biomedical Papers of the Medical Faculty of the University Palacký, Olomouc, Czechoslovakia, 2019, 163, 67-74.	0.2	5
1869	High-performance DNA sequencing to identify genetically determined diseases in pediatric practice. Rossiyskiy Vestnik Perinatologii I Pediatrii, 2019, 64, 103-109.	0.1	4
1873	Znaczenie techniki aCGH w nowotworach mieloproliferacyjnych â€“ przeglÄ…d literatury. Acta Haematologica Polonica, 2019, 50, 135-141.	0.1	0
1877	Towards New Approaches to Evaluate Dynamic Mosaicism in Ring Chromosome 13 Syndrome. Case Reports in Genetics, 2019, 2019, 1-10.	0.1	3
1878	Genetic testing in adults. , 2020, , 43-57.		0
1879	Autism Spectrum Disorders: Prenatal Genetic Testing and Abortion Decision-Making among Taiwanese Mothers of Affected Children. International Journal of Environmental Research and Public Health, 2020, 17, 476.	1.2	3
1880	Clinical application of chromosomal microarray for pathogenic genomic imbalance in fetuses with increased nuchal translucency but normal karyotype. Journal of Genetic Medicine, 2020, 17, 21-26.	0.1	1

#	Article	IF	Citations
1882	Deciphering congenital anomalies for the next generation. Journal of Physical Education and Sports Management, 2020, 6, a005504.	0.5	5
1883	Tetrasomy 9p Syndrome in a Filipino Infant. Acta Medica Philippina, 2020, 54, .	0.0	0
1884	Correlating Neuroimaging and CNVs Data: 7 Years of Cytogenomic Microarray Analysis on Patients Affected by Neurodevelopmental Disorders. Journal of Pediatric Genetics, 2021, 10, 292-299.	0.3	0
1885	Voltage-Gated Sodium Channel Î²₁ Gene: An Overview. Human Heredity, 2020, 85, 101-109.	0.4	3
1886	Neurologic Features with Pathogenic Copy Number Variants. Pediatric Neurology Briefs, 2020, 34, 20.	0.2	2
1887	Detection of Copy Number Variants by Short Multiply Aggregated Sequence Homologies. Journal of Molecular Diagnostics, 2020, 22, 1476-1481.	1.2	0
1888	Role of Copy Number Variations inÂADHD. , 0, , .		1
1889	Microarray and Next Generation Sequencing. Comprehensive Gynecology and Obstetrics, 2021, , 335-343.	0.0	0
1890	Whole-exome sequencing: A changing landscape of prenatal counseling. , 2022, , 39-67.		0
1891	Preoperative Counseling. , 2020, , 69-86.		0
1892	Distal interstitial 1p36 deletion syndrome in a case of global developmental delay with multiple congenital malformations. MGM Journal of Medical Sciences, 2020, 7, 101.	0.1	0
1893	Intellectual and developmental disabilities. , 2020, , 249-259.		1
1894	Identification of MECP2 Duplication Using Low-Depth Whole-Genome Sequencing-Based Copy Number Variation Analysis. Laboratory Medicine Online, 2020, 10, 165.	0.0	0
1895	Routine Chromosomal Microarray Analysis Is Necessary in Turkish Patients with Unexplained Developmental Delay/Intellectual Disability Disorder. Noropsikiyatri Arsivi, 2020, 57, 177-191.	0.2	3
1897	A Diagnostic Approach for Neurogenetic Disorders in the Genome Era. , 2020, , 319-326.		0
1899	Copy number variations in a Brazilian cohort with autism spectrum disorders highlight the contribution of cell adhesion genes. Clinical Genetics, 2022, 101, 134-141.	1.0	13
1900	Molecular genetic analysis and growth hormone response in patients with syndromic short stature. BMC Medical Genomics, 2021, 14, 261.	0.7	1
1901	A recurrent SHANK3 frameshift variant in Autism Spectrum Disorder. Npj Genomic Medicine, 2021, 6, 91.	1.7	9

#	Article	IF	Citations
1902	17p13.3 Microduplication Syndrome: Further Delineating the Clinical Spectrum. Journal of Pediatric Genetics, 2021, 10, 239-244.	0.3	2
1904	Array-CGH and SNP-Arrays, the New Karyotype. , 2012, , 39-52.		0
1905	Clinical Genetic Testing Options. , 2021, , 205-213.		0
1908	A novel 1p33p32.2 deletion involving SCP2, ORC1, and DAB1 genes in a patient with craniofacial dysplasia, short stature, developmental delay, and leukoencephalopathy. Medicine (United States), 2020, 99, e23033.	0.4	2
1909	14q12q13.3 Deletion Diagnosed Using Chromosomal Microarray Analysis in an Infant Showing Seizures, Hypoplasia of the Corpus Callosum, and Developmental Delay. Neonatal Medicine, 2020, 27, 207-213.	0.1	0
1910	Genetics in autism diagnosis: adding molecular subtypes to neurobehavioral diagnoses. Medicine and Health, Rhode Island, 2011, 94, 124-6.	0.1	7
1911	Commentary: Anxiety Disorders and Perceptual Disturbances in Adolescents with 22q11.2 Deletion Syndrome Treated with SSRI: A Case Series. Journal of the Canadian Academy of Child and Adolescent Psychiatry, 2011, 20, 311.	0.7	0
1912	Fluorescence in situ hybridization (FISH) using non-commercial probes in the diagnosis of clinically suspected microdeletion syndromes. Indian Journal of Medical Research, 2013, 138, 135-42.	0.4	2
1913	Social Communication is an Emerging Target for Pharmacotherapy in Autism Spectrum Disorder - A Review of the Literature on Potential Agents. Journal of the Canadian Academy of Child and Adolescent Psychiatry, 2014, 23, 20-30.	0.7	12
1914	Evaluation of the young children with neurodevelopmental disability: a prospective study at hamadan university of medical sciences clinics. Iranian Journal of Child Neurology, 2013, 7, 29-34.	0.2	0
1915	Approach to the diagnosis of developmental delay - the changing scenario. Indian Journal of Medical Research, 2014, 139, 4-6.	0.4	19
1916	Whole Genome Sequencing as a Genetic Test for Autism Spectrum Disorder: From Bench to Bedside and then Back Again. Journal of the Canadian Academy of Child and Adolescent Psychiatry, 2016, 25, 116-21.	0.7	9
1917	Array CGH - A Powerful Tool in Molecular Diagnostic of Pathogenic Microdeletions - Williams-Beuren Syndrome - A Case Report. Current Health Sciences Journal, 2016, 42, 207-212.	0.2	0
1918	Genome-Wide Sequencing for Unexplained Developmental Disabilities or Multiple Congenital Anomalies: A Health Technology Assessment. Ontario Health Technology Assessment Series, 2020, 20, 1-178.	3.0	11
1921	Phenotype Heterogeneity in 3q29 Microduplication Syndrome. Current Health Sciences Journal, 2020, 46, 193-197.	0.2	1
1922	Documentation of units of inheritance and their contribution to phenotype. , 2022, , 3-16.		0
1923	Induced pluripotent stem cells for modeling Smithâ€“Magenis syndrome. , 2022, , 217-246.		1
1924	Genetic Testing in Patients with Neurodevelopmental Disorders: Experience of 511 Patients at Cincinnati Children's Hospital Medical Center. Journal of Autism and Developmental Disorders, 2022, 52, 4828-4842.	1.7	1

#	Article	IF	Citations
1925	The Efficacy of Whole Genome Sequencing and RNA-Seq in the Diagnosis of Whole Exome Sequencing Negative Patients with Complex Neurological Phenotypes. Journal of Pediatric Genetics, 0, , .	0.3	0
1926	The diagnostic trajectory of infants and children with clinical features of genetic disease. Npj Genomic Medicine, 2021, 6, 98.	1.7	6
1927	Medical manifestations and healthcare utilization among adult MyCode participants with neurodevelopmental psychiatric copy number variants. Genetics in Medicine, 2021, , .	1.1	2
1928	Germline 3p22.1 microdeletion encompassing RPSA gene is an ultra-rare cause of isolated asplenia. Molecular Cytogenetics, 2021, 14, 51.	0.4	0
1929	The opinions and experiences of people with intellectual disability regarding genetic testing and genetic medicine: A systematic review. Genetics in Medicine, 2022, 24, 535-548.	1.1	9
1931	Novel de novo pathogenic variant in the GNAI1 gene as a cause of severe disorders of intellectual development. Journal of Human Genetics, 2022, 67, 209-214.	1.1	2
1932	Genetic Analysis of Children With Unexplained Developmental Delay and/or Intellectual Disability by Whole-Exome Sequencing. Frontiers in Genetics, 2021, 12, 738561.	1.1	12
1933	Outcome of 185 pregnancies studied by prenatal genomic array technique: Bioethical reflections. Clinical Ethics, 0, , 147775092110618.	0.5	0
1934	Comprehensive genome sequencing analyses identify novel gene mutations and copy number variations associated with infant developmental delay or intellectual disability (DD/ID). Genes and Diseases, 2022, 9, 1166-1169.	1.5	0
1935	Retardo mental: detecÃ§Ã£o precoce. Revista Debates Em Psiquiatria, 2014, 4, 6-11.	0.3	0
1936	Genotypic and phenotypic variability of 22q11.2 microdeletions â€“ an institutional experience. AIMS Molecular Science, 2021, 8, 257-274.	0.3	3
1937	Cognitive Development and Disorders. , 2018, , .		0
1939	Update on the genetic architecture of schizophrenia. Medizinische Genetik, 2020, 32, 19-24.	0.1	4
1941	Diagnosis of Genetic and Metabolic Conditions. , 2021, , .		0
1942	Considerations for the identification of autism spectrum disorder in children with vision or hearing impairment: A critical review of the literature and recommendations for practice. Clinical Neuropsychologist, 2022, 36, 1049-1068.	1.5	9
1943	Variant interpretation: UCSC Genome Browser Recommended Track Sets. Human Mutation, 2022, , .	1.1	2
1944	Application of array comparative genomic hybridization (aCGH) for identification of chromosomal aberrations in the recurrent pregnancy loss. Journal of Assisted Reproduction and Genetics, 2022, 39, 357-367.	1.2	4
1945	Delayed diagnosis and racial bias in children with genetic conditions. American Journal of Medical Genetics, Part A, 2022, 188, 1118-1123.	0.7	10

#	Article	IF	Citations
1946	Exome and Whole Genome Sequencing in the Neonatal Intensive Care Unit. Clinics in Perinatology, 2022, 49, 167-179.	0.8	6
1947	Implication of Genetic Testing and Pregnancy Outcome in a Woman with Unbalanced Translocation t(1;6). American Journal of Case Reports, 2022, 23, e935370.	0.3	0
1948	Novel deletion of exon 3 in TYRÂgene causing Oculocutaneous albinism 1B in an Indian family along with intellectual disability associated with chromosomal copy number variations. BMC Medical Genomics, 2022, 15, 2.	0.7	2
1949	JAX-CNV: A Whole-genome Sequencing-based Algorithm for Copy Number Detection at Clinical Grade Level. Genomics, Proteomics and Bioinformatics, 2022, 20, 1197-1206.	3.0	3
1950	Chromosomal microarray analysis of 410 Han Chinese patients with autism spectrum disorder or unexplained intellectual disability and developmental delay. Npj Genomic Medicine, 2022, 7, 1.	1.7	11
1951	Clinical Utility of a Unique Genome-Wide DNA Methylation Signature for KMT2A-Related Syndrome. International Journal of Molecular Sciences, 2022, 23, 1815.	1.8	8
1952	A rare etiology of tetralogy of Fallot with pulmonary atresia: Renpenning syndrome. , 2022, 26, 149-150.		1
1953	Prenatal Silver-Russell Syndrome in a Chinese Family Identified by Non-Invasive Prenatal Testing. Molecular Syndromology, 0, , 1-5.	0.3	2
1954	Machine learning models for accurate prioritization of variants of uncertain significance. Human Mutation, 2022, 43, 449-460.	1.1	6
1955	Genome sequencing demonstrates high diagnostic yield in children with undiagnosed global developmental delay/intellectual disability: A prospective study. Human Mutation, 2022, 43, 568-581.	1.1	12
1956	Characterization of Associated Nonclassical Phenotypes in Patients with Deletion in the WAGR Region Identified by Chromosomal Microarray: New Insights and Literature Review. Molecular Syndromology, 0, , 1-15.	0.3	0
1957	Rare variants in the outcome of social skills group training for autism. Autism Research, 2022, 15, 434-446.	2.1	7
1958	High rate of abnormal findings in Prenatal Exome Trio in low risk pregnancies and apparently normal fetuses. Prenatal Diagnosis, 2022, 42, 725-735.	1.1	10
1959	A novel de novo 20q13.11q13.12 microdeletion in a boy with neurodevelopmental disorders - case report. Medycyna Wieku Rozwojowego, 2017, 21, 91-94.	0.2	1
1960	Increased runs of homozygosity in the autosomal genome of Brazilian individuals with neurodevelopmental delay/intellectual disability and/or multiple congenital anomalies investigated by chromosomal microarray analysis. Genetics and Molecular Biology, 2022, 45, e20200480.	0.6	4
1961	Artificial Intelligence in Blood Transcriptomics. , 2022, , 1109-1123.		0
1962	Study on the application value of BACs-on-Beads technology combined with chromosome karyotype analysis in prenatal diagnosis. Translational Pediatrics, 2022, 11, 212-218.	0.5	1
1963	Identification of a novel variant of FOXP3 resulting in severe immune dysregulation, polyendocrinopathy, enteropathy, Xâ€linked syndrome highlights potential pitfalls of molecular testing. Pediatric Dermatology, 2022, 39, 483-485.	0.5	1

#	Article	IF	Citations
1964	Diagnostic yield of next-generation sequencing in 87 families with neurodevelopmental disorders. Orphanet Journal of Rare Diseases, 2022, 17, 60.	1.2	21
1965	Progress in Methods for Copy Number Variation Profiling. International Journal of Molecular Sciences, 2022, 23, 2143.	1.8	9
1966	Duration of untreated autism in rural America: emerging public health crisis. CNS Spectrums, 2022, , 1-4.	0.7	5
1967	Clinical Manifestations of Various Molecular Cytogenetic Variants of Eight Cases of â€œ8p Inverted Duplication/Deletion Syndromeâ€• Biomedicines, 2022, 10, 567.	1.4	3
1968	Genes To Mental Health (G2MH): A Framework to Map the Combined Effects of Rare and Common Variants on Dimensions of Cognition and Psychopathology. American Journal of Psychiatry, 2022, 179, 189-203.	4.0	29
1969	Reciprocal Xp11.4p11.3 microdeletion/microduplication spanning USP9X, DDX3X, and CASK genes in two patients with syndromic intellectual disability. American Journal of Medical Genetics, Part A, 2022, 188, 1836-1847.	0.7	4
1970	Reactive Oxygen Species and Their Consequences on the Structure and Function of Mammalian Spermatozoa. Antioxidants and Redox Signaling, 2022, 37, 481-500.	2.5	12
1971	Genetic care in geographically isolated small island communities: 8Âyears of experience in the Dutch Caribbean. American Journal of Medical Genetics, Part A, 2022, 188, 1777-1791.	0.7	2
1972	The Role of Genetic Testing Among Autistic Individuals. Pediatrics, 2022, 149, .	1.0	1
1973	Lessons learned: next-generation sequencing applied to undiagnosed genetic diseases. Journal of Clinical Investigation, 2022, 132, .	3.9	11
1974	Chromosomal Microarray Reinterpretation: Applications to Pediatric Practice. Journal of Pediatrics, 2022, 243, 219-223.	0.9	0
1975	16p13.11p11.2 triplication syndrome: a new recognizable genomic disorder characterized by optical genome mapping and whole genome sequencing. European Journal of Human Genetics, 2022, , .	1.4	5
1976	Diagnostic yield of patients with undiagnosed intellectual disability, global developmental delay and multiples congenital anomalies using karyotype, microarray analysis, whole exome sequencing from Central Brazil. PLoS ONE, 2022, 17, e0266493.	1.1	9
1977	A scalable artificial intelligence platform that automatically finds copy number variations (CNVs) in journal articles and transforms them into a database: CNV extraction, transformation, and loading AI (CNV-ETLAI). Computers in Biology and Medicine, 2022, 144, 105332.	3.9	1
1978	Global RNAseq of ocular cells reveals gene dysregulation in both asymptomatic and with Congenital Zika Syndrome infants exposed prenatally to Zika virus. Experimental Cell Research, 2022, 414, 113086.	1.2	1
1979	Translational Study of Copy Number Variations in Schizophrenia. International Journal of Molecular Sciences, 2022, 23, 457.	1.8	4
1980	Access, utilization, and awareness for clinical genetic testing in autism spectrum disorder in Sweden: A survey study. Autism, 2022, 26, 1795-1804.	2.4	5
1981	Diagnostic Yield and Economic Implications of Whole-Exome Sequencing for ASD Diagnosis in Israel. Genes, 2022, 13, 36.	1.0	5

#	Article	IF	Citations
1982	Increased Diagnostic Yield of Array Comparative Genomic Hybridization for Autism Spectrum Disorder in One Institution in Taiwan. Medicina (Lithuania), 2022, 58, 15.	0.8	5
1984	Screening for FMR1 CGG Repeat Expansion in Thai Patients with Autism Spectrum Disorder. BioMed Research International, 2021, 2021, 1-11.	0.9	1
1985	A Next Generation Sequencing-Based Protocol for Screening of Variants of Concern in Autism Spectrum Disorder. Cells, 2022, 11, 10.	1.8	16
1986	Noninvasive prenatal testing suggesting an abnormality in chromosome 15 confirmed to be a case of Praderâ€“Willi syndrome caused by trisomy rescue in the neonatal period. Journal of Obstetrics and Gynaecology Research, 2022, , .	0.6	1
1987	Investigation of Chromosomal Structural Abnormalities in Patients With Undiagnosed Neurodevelopmental Disorders. Frontiers in Genetics, 2022, 13, 803088.	1.1	1
1988	Chromosomal microarray in postnatal diagnosis of congenital anomalies and neurodevelopmental disorders in Serbian patients. Journal of Clinical Laboratory Analysis, 2022, 36, e24441.	0.9	8
2022	A familial 3q28q29 duplication induced mild intellectual disability: case presentation and literature review.. American Journal of Translational Research (discontinued), 2022, 14, 1663-1671.	0.0	0
2023	Interpretation challenge of small copy number variations in the imprinting regions. Molecular Genetics & Genomic Medicine, 2022, , e1961.	0.6	0
2024	Genome interpretation using in silico predictors of variant impact. Human Genetics, 2022, 141, 1549-1577.	1.8	26
2025	Experience of Low-Pass Whole-Genome Sequencing-Based Copy Number Variant Analysis: A Survey of Chinese Tertiary Hospitals. Diagnostics, 2022, 12, 1098.	1.3	4
2026	Third-Generation Cytogenetic Analysis. Journal of Molecular Diagnostics, 2022, 24, 711-718.	1.2	4
2027	Copy Number Variant Risk Scores Associated With Cognition, Psychopathology, and Brain Structure in Youths in the Philadelphia Neurodevelopmental Cohort. JAMA Psychiatry, 2022, 79, 699.	6.0	8
2028	Prenatal chromosomal microarray analysis in foetuses with isolated absent or hypoplastic nasal bone. Annals of Medicine, 2022, 54, 1297-1302.	1.5	4
2029	De Novo Large Deletion Leading to Fragile X Syndrome. Frontiers in Genetics, 2022, 13, .	1.1	1
2030	Genetic Screening and Prenatal Genetic Diagnosis. , 2017, , 193-218.		3
2031	Case Report: Novel Biallelic Null Variants of SMPD4 Confirm Its Involvement in Neurodevelopmental Disorder With Microcephaly, Arthrogryposis, and Structural Brain Anomalies. Frontiers in Genetics, 2022, 13, .	1.1	3
2032	Variability in Phelan-McDermid Syndrome in a Cohort of 210 Individuals. Frontiers in Genetics, 2022, 13, 652454.	1.1	27
2033	Isolated Coarctation of the Aorta: Current Concepts and Perspectives. Frontiers in Cardiovascular Medicine, 2022, 9, .	1.1	4

#	Article	IF	Citations
2037	Expanding the Molecular Spectrum of ANKRD11 Gene Defects in 33 Patients with a Clinical Presentation of KBG Syndrome. International Journal of Molecular Sciences, 2022, 23, 5912.	1.8	6
2038	Clinical, demographic and genetic features of patients with congenital heart disease : A single center experience. Marmara Medical Journal, 0, , .	0.2	0
2039	Mxc, a Drosophila homolog of mental retardation-associated gene NPAT, maintains neural stem cell fate. Cell and Bioscience, 2022, 12, .	2.1	4
2040	Prenatal diagnosis and genetic counseling of a 10p11.23q11.21 duplication associated with normal phenotype. Molecular Cytogenetics, 2022, 15, .	0.4	2
2041	Evidence-Based Genetic Testing for Individuals with Congenital Diaphragmatic Hernia. Journal of Pediatrics, 2022, 248, 13-14.	0.9	1
2043	Array-Based Comparative Genomic Hybridization Analysis in Children with Developmental Delay/Intellectual Disability. Balkan Journal of Medical Genetics, 2021, 24, 15-24.	0.5	3
2044	Current Situation in Planning and Evaluation of Etiological Genetic Tests in Children with Developmental Delay/Intellectual Disability: Single Center Experience. Celal Bayar Ãœniversitesi SaÄŸlÄ±k Bilimleri EnstitÃ¼sÃ¼ Dergisi, 0, , .	0.1	0
2045	Mechanisms of structural chromosomal rearrangement formation. Molecular Cytogenetics, 2022, 15, .	0.4	28
2046	Deleterious Variation in BR Serine/Threonine Kinase 2 Classified a Subtype of Autism. Frontiers in Molecular Neuroscience, 0, 15, .	1.4	8
2047	Prenatal Diagnosis Using Chromosomal Microarray Analysis in High-Risk Pregnancies. Journal of Clinical Medicine, 2022, 11, 3624.	1.0	3
2048	Impaired Neurodevelopmental Genes in Slovenian Autistic Children Elucidate the Comorbidity of Autism With Other Developmental Disorders. Frontiers in Molecular Neuroscience, 0, 15, .	1.4	1
2049	A Decade's Experience in Pediatric Chromosomal Microarray Reveals Distinct Characteristics Across Ordering Specialties. Journal of Molecular Diagnostics, 2022, 24, 1031-1040.	1.2	3
2050	Two Genetic Mechanisms in Two Siblings with Intellectual Disability, Autism Spectrum Disorder, and Psychosis. Journal of Personalized Medicine, 2022, 12, 1013.	1.1	7
2051	Better and faster is cheaper. Human Mutation, 2022, 43, 1495-1506.	1.1	2
2052	Diagnostic Value of Microarray Method in Autism Spectrum Disorder, Intellectual Disability, and Multiple Congenital Anomalies and Some Candidate Genes for Autism: Experience of Two Centers. Medeniyet Medical Journal, 2022, 37, 180-193.	0.4	0
2053	Yield of arrayâ€‹<scp>CGH</scp> analysis in Tunisian children with autism spectrum disorder. Molecular Genetics & Genomic Medicine, 2022, 10, .	0.6	8
2054	Evaluation of rotavirus vaccine administration among a 22q11.2DS patient population. Allergy, Asthma and Clinical Immunology, 2022, 18, .	0.9	0
2055	Genetic Analysis Algorithm for the Study of Patients with Multiple Congenital Anomalies and Isolated Congenital Heart Disease. Genes, 2022, 13, 1172.	1.0	2

#	Article	IF	Citations
2056	DNA Methylation Episignatures in Neurodevelopmental Disorders Associated with Large Structural Copy Number Variants: Clinical Implications. International Journal of Molecular Sciences, 2022, 23, 7862.	1.8	8
2057	A bibliometrics analysis and visualization of autism spectrum disorder. Frontiers in Psychiatry, 0, 13, .	1.3	6
2058	Clinical and genetic study of three families with 15q11q13 duplications. Taiwanese Journal of Obstetrics and Gynecology, 2022, 61, 717-721.	0.5	0
2059	COL4A4 variant recently identified: lessons learned in variant interpretationâ€"a case report. BMC Nephrology, 2022, 23, .	0.8	0
2060	Genetic causes of rare and common epilepsies: What should the epileptologist know?. European Journal of Medical Genetics, 2022, 65, 104570.	0.7	4
2061	Utility and Outcomes of the 2019 American College of Medical Genetics and Genomicsâ€"Clinical Genome Resource Guidelines for Interpretation of Copy Number Variants with Borderline Classifications at an Academic Clinical Diagnostic Laboratory. Journal of Molecular Diagnostics, 2022, 24, 1100-1111.	1.2	2
2062	The second-tier status of fragile X syndrome testing for unexplained intellectual disability/global developmental delay in the era of next-generation sequencing. Frontiers in Pediatrics, 0, 10, .	0.9	1
2063	Potentials and challenges of chromosomal microarray analysis in prenatal diagnosis. Frontiers in Genetics, 0, 13, .	1.1	6
2064	Similar Rates of Deleterious Copy Number Variants in Early-Onset Psychosis and Autism Spectrum Disorder. American Journal of Psychiatry, 2022, 179, 853-861.	4.0	10
2066	Deletion of 2q22.2q22.3 in Mowatâ€"Wilson Syndrome: A Case Report and Review of the Literature. Journal of Pediatric Neurology, 2022, 20, 440-444.	0.0	1
2067	Prenatal diagnosis and genetic counseling of an inherited unbalanced chromosome abnormalities in a Chinese family. Molecular Cytogenetics, 2022, 15, .	0.4	0
2069	Genetic investigations pathway for people with intellectual disability, autism and/or epilepsy. BJ Psych Advances, 0, , 1-10.	0.5	0
2070	How do genetic tests answer questions about neurodevelopmental differences? A sociological take. Developmental Medicine and Child Neurology, 2022, 64, 1462-1469.	1.1	1
2071	Longâ€read sequencing for molecular diagnostics in constitutional genetic disorders. Human Mutation, 2022, 43, 1531-1544.	1.1	9
2072	Fragile X Syndrome Caused by Maternal Somatic Mosaicism of FMR1 Gene: Case Report and Literature Review. Genes, 2022, 13, 1609.	1.0	1
2073	Copy Number Variation Analysis Facilitates Identification of Genetic Causation in Patients with Congenital Anomalies of the Kidney and Urinary Tract. European Urology Open Science, 2022, 44, 106-112.	0.2	4
2074	Molecular evaluation of gene mutation profiles and copy number variations in pediatric acute myeloid leukemia. Leukemia Research, 2022, 122, 106954.	0.4	2
2075	Dopaminergic Gene Dosage Reveals Distinct Biological Partitions between Autism and Developmental Delay as Revealed by Complex Network Analysis and Machine Learning Approaches. Journal of Personalized Medicine, 2022, 12, 1579.	1.1	2

#	Article	IF	Citations
2076	Chromosomal microarray analyses from 5778 patients with neurodevelopmental disorders and congenital anomalies in Brazil. Scientific Reports, 2022, 12, .	1.6	8
2077	Exome sequencing as a first-tier test for copy number variant detection: retrospective evaluation and prospective screening in 2418 cases. Journal of Medical Genetics, 2022, 59, 1234-1240.	1.5	13
2079	Clinical experience of noninvasive prenatal testing for rare chromosome abnormalities in singleton pregnancies. Frontiers in Genetics, 0, 13, .	1.1	3
2080	Genome sequencing is a sensitive first-line test to diagnose individuals with intellectual disability. Genetics in Medicine, 2022, 24, 2296-2307.	1.1	15
2081	Patient-facing digital tools for delivering genetic services: a systematic review. Journal of Medical Genetics, 2023, 60, 1-10.	1.5	12
2082	Prenatal diagnosis and genetic counseling of a paternally inherited microduplication 18q11.1 to 18q11.2 in a chinese family. Molecular Cytogenetics, 2022, 15, .	0.4	0
2084	Exploring Genes and Phenotypes Within Chromosomal Regions Using OMIM's GeneScout. Current Protocols, 2022, 2, .	1.3	2
2085	Clinical and genomic delineation of the new proximal 19p13.3 microduplication syndrome. American Journal of Medical Genetics, Part A, 0, , .	0.7	1
2086	Learning to Crawl: Determining the Role of Genetic Abnormalities on Postoperative Outcomes in Congenital Heart Disease. Journal of the American Heart Association, 2022, 11, .	1.6	5
2087	An Approach to the Genetic Evaluation of Children with Autism Spectrum Disorders. Autism and Child Psychopathology Series, 2022, , 263-276.	0.1	0
2088	Laboratory Testing for Prader-Willi Syndrome. , 2022, , 75-91.		0
2089	Genomics technologies and bioinformatics in allergy and immunology. , 2022, , 221-260.		0
2090	Cytogenetics analysis as the central point of genetic testing in acute myeloid leukemia (AML): a laboratory perspective for clinical applications. Clinical and Experimental Medicine, 0, , .	1.9	0
2091	Barriers to genetic testing in clinical psychiatry and ways to overcome them: from cliniciansâ€™ attitudes to sociocultural differences between patients across the globe. Translational Psychiatry, 2022, 12, .	2.4	8
2092	Angelman syndrome with mosaic paternal uniparental disomy suggestive of mitotic nondisjunction. Journal of Human Genetics, 0, , .	1.1	0
2094	Differential newborn <scp>DNA</scp> methylation among individuals with complex congenital heart defects and childhood lymphoma. Birth Defects Research, 0, , .	0.8	0
2096	Maturity-onset diabetes of the young in a large Portuguese cohort. Acta Diabetologica, 2023, 60, 83-91.	1.2	1
2097	Severity Scoring Cutoff for MLPA and Its Diagnostic Yield in 332 North Indian Children with Developmental Delay. Journal of Pediatric Genetics, 0, , .	0.3	0

#	Article	IF	Citations
2098	Copy number variant analysis for syndromic congenital heart disease in the Chinese population. Human Genomics, 2022, 16, .	1.4	1
2099	Molecular Diagnostic Yield of Exome Sequencing and Chromosomal Microarray in Cerebral Palsy. JAMA Neurology, 2022, 79, 1287.	4.5	18
2100	Rates of rare copy number variants in different circumstances among patients with genetic developmental and epileptic encephalopathy. Science Progress, 2022, 105, 003685042211312.	1.0	1
2101	Longâ€read sequence analysis for clustered genomic copy number aberrations revealed architectures of intricately intertwined rearrangements. American Journal of Medical Genetics, Part A, 0, , .	0.7	1
2102	Expanded Phenotypic Spectrum or Multiple Syndromes?. Molecular Syndromology, 0, , 1-2.	0.3	1
2104	Prenatal diagnosis and genetic counseling of an inherited Xq24q25 deletion associated with normal phenotype. Molecular Cytogenetics, 2022, 15, .	0.4	0
2105	<scp>j5p13</scp> microduplication in a malformed fetus and his unaffected father. American Journal of Medical Genetics, Part A, 0, , .	0.7	0
2106	Clinical evaluation of rare copy number variations identified by chromosomal microarray in a Hungarian neurodevelopmental disorder patient cohort. Molecular Cytogenetics, 2022, 15, .	0.4	2
2107	Identifying Genetic Etiology in Patients with Intellectual Disability: An Experience in Public Health Services in Northeastern Brazil. Journal of Pediatric Genetics, 0, , .	0.3	0
2108	Copy Number Variation and Structural Genomic Findings in 116 Cases of Sudden Unexplained Death between 1 and 28 Months of Age. Genetics & Genomics Next, 2023, 4, .	0.8	1
2109	Applications of Noninvasive Prenatal Testing for Subchromosomal Copy Number Variations Using Cell-Free DNA. Clinics in Laboratory Medicine, 2022, 42, 613-625.	0.7	1
2110	Intellectual Developmental Disorders (Developmental Delay). , 2023, , 450-472.e2.		0
2112	New approaches to genetic counseling. , 2024, , 173-195.		0
2113	Navigating the labyrinth of genetic testing. Journal of Marine Medical Society, 2022, .	0.0	0
2114	The clinical diagnostic utility of array CGH in children with syndromic microcephaly. Annals of Indian Academy of Neurology, 2022, 25, 1067.	0.2	0
2115	Genomic strategies to untangle the etiology of autism: A primer. Autism Research, 2023, 16, 31-39.	2.1	2
2116	Deciphering Intellectual Disability. Indian Journal of Pediatrics, 0, , .	0.3	2
2117	Utility of genetic work-up for 46, XY patients with severe hypospadias. Journal of Pediatric Urology, 2022, , .	0.6	1

#	Article	IF	Citations
2118	Progress in Diagnosis and Management of Intellectual Disability in India: A Journey Over Half-a-Century !. Indian Pediatrics, 2022, 59, 875-878.	0.2	0
2119	Identification of Biallelic dystrophin gene variants during maternal carrier testing for Becker muscular dystrophy and review of the DMD exon 49â€“51 deletion phenotype. Molecular Genetics & Genomic Medicine, 2023, 11, .	0.6	2
2120	Microduplication 3p26.3p24.3 and 4q34.3q35.2 Microdeletion Identified in a Patient with Developmental Delay Associated with Brain Malformation. Diagnostics, 2022, 12, 2887.	1.3	0
2121	Comparing saliva and blood for the detection of mosaic genomic abnormalities that cause syndromic intellectual disability. European Journal of Human Genetics, 2023, 31, 521-525.	1.4	2
2122	Increased Prevalence of Rare Copy Number Variants in Treatment-Resistant Psychosis. Schizophrenia Bulletin, 2023, 49, 881-892.	2.3	6
2123	Evaluating Genetic Disorders in the Neonate: The Role of Exome Sequencing in the NICU. NeoReviews, 2022, 23, e829-e840.	0.4	1
2125	Response to: Letter to Editor - Utility of Genetic Work-Up for 46, XY Patients with Severe Hypospadias. Journal of Pediatric Urology, 2022, , .	0.6	0
2126	Access to clinically indicated genetic tests for pediatric patients with Medicaid: Evidence from outpatient genetics clinics in Texas. Genetics in Medicine, 2023, 25, 100350.	1.1	1
2127	Further characterization of the 9q31 microdeletion phenotype; delineation of a common region of overlap containing ZNF462. Molecular Genetics & Genomic Medicine, 2023, 11, .	0.6	2
2128	DNA methylation episignatures: insight into copy number variation. Epigenomics, 2022, 14, 1373-1388.	1.0	7
2129	Consequences of chromosome gain: A new view on trisomy syndromes. American Journal of Human Genetics, 2022, 109, 2126-2140.	2.6	10
2130	Burden of Rare Copy Number Variants in Microcephaly: A Brazilian Cohort of 185 Microcephalic Patients and Review of the Literature. Journal of Autism and Developmental Disorders, 2024, 54, 1181-1212.	1.7	3
2131	Pathogenic Copy Number Variations Involved in the Genetic Etiology of Syndromic and Non-Syndromic Intellectual Disabilityâ€”Data from a Romanian Cohort. Diagnostics, 2022, 12, 3137.	1.3	0
2133	Child with Global Developmental Delay. , 2022, , 25-45.		0
2134	Development of a clinical microarray system for genetic analysis screening. Practical Laboratory Medicine, 2023, 33, e00306.	0.6	0
2135	A de novo PAK1 likely pathogenic variant and a de novo terminal 1q microdeletion in a Chinese girl with global developmental delay, severe intellectual disability, and seizures. BMC Medical Genomics, 2023, 16, .	0.7	0
2136	Clinical Practice Guidelines for the Immunological Management of Chromosome 22q11.2 Deletion Syndrome and Other Defects in Thymic Development. Journal of Clinical Immunology, 2023, 43, 247-270.	2.0	5
2137	A Prospective Study of Genetic Variants in Infants with Congenital Unilateral Sensorineural Hearing Loss. Journal of Clinical Medicine, 2023, 12, 495.	1.0	1

#	Article	IF	Citations
2138	Genetic Influences on Fetal Alcohol Spectrum Disorder. Genes, 2023, 14, 195.	1.0	7
2139	Molecular Mechanisms Involved in Craniosynostosis. In Vivo, 2023, 37, 36-46.	0.6	1
2140	Clinical application of prenatal chromosomal microarray. Journal of Genetic Medicine, 2022, 19, 43-48.	0.1	0
2142	The first Korean case of 2p15p16.1 microdeletion syndrome, characterized by facial dysmorphism, developmental delay, and congenital hypothyroidism. Journal of Genetic Medicine, 2022, 19, 105-110.	0.1	0
2143	Copy number variation analysis in 189 Romanian patients with global developmental delay/intellectual disability. Italian Journal of Pediatrics, 2022, 48, .	1.0	1
2144	Extended application of BACs-on-Beads technique in prenatal diagnosis. Archives of Medical Science, 2023, 19, 250-257.	0.4	0
2145	HibridaciÃ³n genÃ³mica comparativa: su interpretaciÃ³n y uso como herramienta diagnÃ³stica en retardo mental inespecÃfico y sÃndromes de microdeleciÃ³n/microduplicaciÃ³n. Medicas UIS, 2016, 29, .	0.0	0
2146	Rare Disease Genomics and Clinical Diagnostics. , 2019, , 212-225.		0
2147	Inherited deletion of 9p22.3â€p24.3 and duplication of 18p11.31â€p11.32 associated with neurodevelopmental delay: Phenotypic matching of involved genes. Journal of Cellular and Molecular Medicine, 2023, 27, 496-505.	1.6	3
2148	KIF1A-Associated Neurological Disorder: An Overview of a Rare Mutational Disease. Pharmaceuticals, 2023, 16, 147.	1.7	4
2149	Chromosomal Microarray Analysis Identifies a Novel SALL1 Deletion, Supporting the Association of Haploinsufficiency with a Mild Phenotype of Townesâ€“Brocks Syndrome. Genes, 2023, 14, 258.	1.0	1
2150	Behavioral and Sensory Deficits Associated with Dysfunction of GABAergic System in a Novel shank2-Deficient Zebrafish Model. International Journal of Molecular Sciences, 2023, 24, 2208.	1.8	2
2151	The role of structural variations in Alzheimerâ€™s disease and other neurodegenerative diseases. Frontiers in Aging Neuroscience, 0, 14, .	1.7	5
2152	Evaluation of Individuals with Non-Syndromic Global Developmental Delay and Intellectual Disability. Children, 2023, 10, 414.	0.6	0
2154	Reappraisal of evolving methods in non-invasive prenatal screening: Discovery, biology and clinical utility. Heliyon, 2023, 9, e13923.	1.4	0
2155	Evaluation and Analysis of Absence of Homozygosity (AOH) Using Chromosome Analysis by Medium Coverage Whole Genome Sequencing (CMA-seq) in Prenatal Diagnosis. Diagnostics, 2023, 13, 560.	1.3	1
2157	Detection rates of abnormalities in over 10,000 amniotic fluid samples at a single laboratory. BMC Pregnancy and Childbirth, 2023, 23, .	0.9	0
2158	Multisite Assessment of Optical Genome Mapping for Analysis of Structural Variants in Constitutional Postnatal Cases. Journal of Molecular Diagnostics, 2023, 25, 175-188.	1.2	14

#	Article	IF	Citations
2159	Genetic and metabolic investigations for neurodevelopmental disorders: position statement of the Canadian College of Medical Geneticists (CCMG). Journal of Medical Genetics, 2023, 60, 523-532.	1.5	8
2160	Genetic evaluation of living kidney donor candidates: A review and recommendations for best practices. American Journal of Transplantation, 2023, 23, 597-607.	2.6	3
2161	Genome-Wide Sequencing Modalities for Children with Unexplained Global Developmental Delay and Intellectual Disabilitiesâ€"A Narrative Review. Children, 2023, 10, 501.	0.6	1
2162	Chromosome Microarray Analysis for the Investigation of Deletions in Pediatric Movement Disorders: A Systematic Review of the Literature. Movement Disorders Clinical Practice, 2023, 10, 547-557.	0.8	0
2163	The evolution of comprehensive genetic analysis in neurology: Implications for precision medicine. Journal of the Neurological Sciences, 2023, 447, 120609.	0.3	3
2164	Construction of copy number variation landscape and characterization of associated genes in a Bangladeshi cohort of neurodevelopmental disorders. Frontiers in Genetics, 0, 14, .	1.1	4
2165	A crowdsourcing database for the copy-number variation of the Spanish population. Human Genomics, 2023, 17, .	1.4	2
2166	Clinical and Genetic Characteristics of Patients with Unexplained Intellectual Disability/Developmental Delay without Epilepsy. Molecular Syndromology, 2023, 14, 208-218.	0.3	3
2167	Targeted sequencing and clinical strategies in children with autism spectrum disorder: A cohort study. Frontiers in Genetics, 0, 14, .	1.1	1
2168	Genetic Counselling for Global Developmental Delay/Intellectual Disability (GDD/ID) â€" Changing Landscapes and Persisting Challenges. Indian Pediatrics, 2023, 60, 142-145.	0.2	0
2169	Novel mutation causing Zellweger syndrome. BMJ Case Reports, 2023, 16, e252014.	0.2	0
2171	Gain of CCND1 May Occur Too Infrequently in Cutaneous Melanoma, and Too Late in Melanomagenesis, to Be Diagnostically Useful: Genomic Analysis of 88 Cases. American Journal of Dermatopathology, 2023, 45, 311-319.	0.3	0
2173	Implementation of Exome Sequencing in Clinical Practice for Neurological Disorders. Genes, 2023, 14, 813.	1.0	2
2174	Chromosomal Microarray in Patients with Non-Syndromic Autism Spectrum Disorders in the Clinical Routine of a Tertiary Hospital. Genes, 2023, 14, 820.	1.0	2
2176	Identification of Extremely Rare Pathogenic CNVs by Array CGH in Saudi Children with Developmental Delay, Congenital Malformations, and Intellectual Disability. Children, 2023, 10, 662.	0.6	1
2177	Opportunities and achievements of using massive parallel sequencing in the diagnosis of neurodevelopmental diseases. Epilepsy and Paroxysmal Conditions, 2023, 15, 44-52.	0.2	1
2178	Validation and depth evaluation of low-pass genome sequencing in prenatal diagnosis using 387 amniotic fluid samples. Journal of Medical Genetics, 2023, 60, 933-938.	1.5	1
2179	Leveraging Unique Chromosomal Microarray Probes to Accurately Detect Copy Number at the Highly Homologous 15q15.3 Deafness-Infertility Syndrome Locus. Clinical Chemistry, 0, , .	1.5	1

#	Article	IF	Citations
2180	Retrospective file review shows limited genetic services fail most patients â€“ an argument for the implementation of exome sequencing as a first-tier test in resource-constrained settings. Orphanet Journal of Rare Diseases, 2023, 18, .	1.2	3
2181	From genetic laboratory to the genetic clinic â€“ Opportunities and challenges: An illustrated review. Apollo Medicine, 2023, 20, 123.	0.0	0
2182	Chromosomal microarrays and next-generation sequencing for diagnosis of fetal abnormalities. , 2023, , 767-787.		0
2183	Next-generation sequencing for gene panels, clinical exome, and whole-genome analysis. , 2023, , 743-766.		0
2193	The Human Genome and Neonatal Care. , 2024, , 309-321.e4.		0
2194	Chromosome Disorders. , 2024, , 347-361.e2.		0
2195	Prenatal Diagnosis and Counseling. , 2024, , 322-334.e2.		0
2228	Whole-genome sequencing as a method of prenatal genetic diagnosis: Ethical issues. , 2023, , 275-304.		0
2240	Development and Developmental Disorders of the Cerebral Cortex. , 2023, , 725-891.		0
2250	Causes of Congenital Malformations. , 2023, , 171-248.		0
2251	Neurobiology of Autism Spectrum Disorder. , 2023, , 1-38.		0
2268	Molecular pathology and testing in melanocytic tumors. , 2024, , 359-384.		0
2269	Epigenomic mechanisms and episignature biomarkers in rare diseases. , 2024, , 1031-1076.		0
2271	Molecular testing in autism spectrum disorder. , 2024, , 291-301.		0
2293	Review Paper: Autism Spectrum Disorderâ€"Molecular Mechanisms and Diagnosis. IFMBE Proceedings, 2024, , 227-235.	0.2	0
2294	An Overview of GeneticTesting. , 2024, , 671-676.		0
2295	The expanding diagnostic toolbox for rare genetic diseases. Nature Reviews Genetics, 0, , .	7.7	0
2296	Clinical Genetics and Referrals. , 2024, , 45-72.		0

#	Article	IF	Citations
2301	Genetic and Genomic Testing. , 2024, , 73-91.		0
2307	Updates in Clinical Genetics. , 2023, , 285-295.		0
2315	Neurodevelopmental Disorders: Intellectual Disability. , 2023, , 1-39.		0
2317	Prenatal Genetic Testing. , 2023, , 197-210.		0
2324	Ring Chromosome 22. , 2024, , 301-335.		0