Zeynep Tumer

List of Publications by Year in descending order

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224 papers	9,187 citations	50273 46 h-index	54911 84 g-index
234 all docs	234 docs citations	234 times ranked	13034 citing authors

#	Article	IF	Citations
1	A human phenome-interactome network of protein complexes implicated in genetic disorders. Nature Biotechnology, 2007, 25, 309-316.	17.5	871
2	Isolation of a candidate gene for Menkes disease that encodes a potential heavy metal binding protein. Nature Genetics, 1993, 3, 14-19.	21.4	708
3	Clinical and molecular diagnosis, screening and management of Beckwithâ€“Wiedemann syndrome: an international consensus statement. Nature Reviews Endocrinology, 2018, 14, 229-249.	9.6	388
4	Diagnosis and management of Silverâ€“Russell syndrome: first international consensus statement. Nature Reviews Endocrinology, 2017, 13, 105-124.	9.6	336
5	Menkes disease. European Journal of Human Genetics, 2010, 18, 511-518.	2.8	283
6	Axenfeldâ€“Rieger syndrome and spectrum of PITX2 and FOXC1 mutations. European Journal of Human Genetics, 2009, 17, 1527-1539.	2.8	243
7	An Overview and Update ofATP7AMutations Leading to Menkes Disease and Occipital Horn Syndrome. Human Mutation, 2013, 34, 417-429.	2.5	237
8	Diagnosis and management of Cornelia de Lange syndrome: first international consensus statement. Nature Reviews Genetics, 2018, 19, 649-666.	16.3	223
9	Imprinting disorders: a group of congenital disorders with overlapping patterns of molecular changes affecting imprinted loci. Clinical Epigenetics, 2015, 7, 123.	4.1	174
10	Cornelia de Lange syndrome. Clinical Genetics, 2015, 88, 1-12.	2.0	157
11	Truncation of the Down Syndrome Candidate Gene DYRK1A in Two Unrelated Patients with Microcephaly. American Journal of Human Genetics, 2008, 82, 1165-1170.	6.2	145
12	Similar Splice-Site Mutations of the ATP7A Gene Lead to Different Phenotypes: Classical Menkes Disease or Occipital Horn Syndrome. American Journal of Human Genetics, 2000, 66, 1211-1220.	6.2	122
13	Transient p53 Suppression Increases Reprogramming of Human Fibroblasts without Affecting Apoptosis and DNA Damage. Stem Cell Reports, 2014, 3, 404-413.	4.8	114
14	High frequency of submicroscopic genomic aberrations detected by tiling path array comparative genome hybridisation in patients with isolated congenital heart disease. Journal of Medical Genetics, 2008, 45, 704-709.	3.2	110
15	Early treatment of Menkes disease with parenteral Cooper-Histidine: Long-term follow-up of four treated patients. , 1998, 76, 154-164.		109
16	Recent Advances in Imprinting Disorders. Clinical Genetics, 2017, 91, 3-13.	2.0	101
17	Haploinsufficiency of TAB2 Causes Congenital Heart Defects in Humans. American Journal of Human Genetics, 2010, 86, 839-849.	6.2	97
18	Early copper-histidine treatment for Menkes disease. Nature Genetics, 1996, 12, 11-13.	21.4	94

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19	Characterization of the exon structure of the Menkes disease gene using vectorette PCR. Genomics, 1995, 26, 437-442.	2.9	91
20	Pierre Robin sequence may be caused by dysregulation of SOX9 and KCNJ2. Journal of Medical Genetics, 2007, 44, 381-386.	3.2	91
21	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	6.2	90
22	Identification of point mutations in 41 unrelated patients affected with Menkes disease. American Journal of Human Genetics, 1997, 60, 63-71.	6.2	83
23	Menkes disease: recent advances and new aspects.. Journal of Medical Genetics, 1997, 34, 265-274.	3.2	82
24	Genetic anticipation in Behcet's syndrome. Annals of the Rheumatic Diseases, 1998, 57, 45-48.	0.9	81
25	Hedgehog signaling in small-cell lung cancer: Frequent in vivo but a rare event in vitro. Lung Cancer, 2006, 52, 281-290.	2.0	80
26	Molecular genetic analysis using targeted NGS analysis of 677 individuals with retinal dystrophy. Scientific Reports, 2019, 9, 1219.	3.3	76
27	Autism and developmental disability caused by KCNQ3 gainâ€ofâ€function variants. Annals of Neurology, 2019, 86, 181-192.	5.3	73
28	Aberrant expression of miRâ€218 and miRâ€204 in human mesial temporal lobe epilepsy and hippocampal sclerosisâ€"Convergence on axonal guidance. Epilepsia, 2014, 55, 2017-2027.	5.1	71
29	Recommendations for a nomenclature system for reporting methylation aberrations in imprinted domains. Epigenetics, 2018, 13, 117-121.	2.7	70
30	Intragenic deletions affecting two alternative transcripts of the IMMP2L gene in patients with Tourette syndrome. European Journal of Human Genetics, 2014, 22, 1283-1289.	2.8	69
31	Phenotypes and genotypes in individuals with SMC1A variants. American Journal of Medical Genetics, Part A, 2017, 173, 2108-2125.	1.2	69
32	Menkes Disease: An Xâ€Linked Neurological Disorder of the Copper Metabolism. Brain Pathology, 1992, 2, 351-362.	4.1	68
33	Disruption of the CNTNAP2 gene in a t(7;15) translocation family without symptoms of Gilles de la Tourette syndrome. European Journal of Human Genetics, 2007, 15, 711-713.	2.8	68
34	EMQN best practice guidelines for the molecular genetic testing and reporting of chromosome 11p15 imprinting disorders: Silverâ€"Russell and Beckwithâ€"Wiedemann syndrome. European Journal of Human Genetics, 2016, 24, 1377-1387.	2.8	68
35	Clinical and biochemical consequences of copper-histidine therapy in Menkes disease. European Journal of Pediatrics, 1993, 152, 828-832.	2.7	67
36	Mutation Spectrum of ATP7A, the Gene Defective in Menkes Disease. Advances in Experimental Medicine and Biology, 1999, 448, 83-95.	1.6	67

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37	Mapping of the Menkes locus to Xq13.3 distal to the X-inactivation center by an intrachromosomal insertion of the segment Xq13.3-q21.2. Human Genetics, 1992, 88, 668-672.	3.8	66
38	An excess of chromosome 1 breakpoints in male infertility. European Journal of Human Genetics, 2004, 12, 993-1000.	2.8	56
39	Molecular cytogenetic characterization of ring chromosome 15 in three unrelated patients. , 2004, 130A, 340-344.		56
40	Transient Neonatal Diabetes, ZFP57, and Hypomethylation of Multiple Imprinted Loci. Diabetes Care, 2013, 36, 505-512.	8.6	56
41	Screening of 383 unrelated patients affected with Menkes disease and finding of 57 gross deletions inATP7A. Human Mutation, 2003, 22, 457-464.	2.5	55
42	Additional chromosomal abnormalities in patients with a previously detected abnormal karyotype, mental retardation, and dysmorphic features. American Journal of Medical Genetics, Part A, 2006, 140A, 2180-2187.	1.2	54
43	Single gene microdeletions and microduplication of 3p26.3 in three unrelated families: CNTN6 as a new candidate gene for intellectual disability. Molecular Cytogenetics, 2014, 7, 97.	0.9	51
44	Three New Loci for Determining X Chromosome Inactivation Patterns. Journal of Molecular Diagnostics, 2011, 13, 537-540.	2.8	50
45	Germline Chromothripsis Driven by L1-Mediated Retrotransposition and Alu/Alu Homologous Recombination. Human Mutation, 2016, 37, 385-395.	2.5	50
46	Identification and analysis of 21 novel disease-causing amino acid substitutions in the conserved part of ATP7A. Human Mutation, 2005, 26, 84-93.	2.5	49
47	A germline chromothripsis event stably segregating in 11 individuals through three generations. Genetics in Medicine, 2016, 18, 494-500.	2.4	48
48	A novel heterozygous nonsense mutation of the OPTN gene segregating in a Danish family with ALS. Neurobiology of Aging, 2012, 33, 208.e1-208.e5.	3.1	47
49	High resolution comparative genomic hybridisation analysis reveals imbalances in dyschromosomal patients with normal or apparently balanced conventional karyotypes. European Journal of Human Genetics, 2000, 8, 661-668.	2.8	45
50	A homozygous nonsense mutation (c.214C->A) in the biliverdin reductase alpha gene (BLVRA) results in accumulation of biliverdin during episodes of cholestasis. Journal of Medical Genetics, 2011, 48, 219-225.	3.2	45
51	Dysfunction of the Heteromeric KV7.3/KV7.5 Potassium Channel is Associated with Autism Spectrum Disorders. Frontiers in Genetics, 2013, 4, 54.	2.3	45
52	Refinement of the critical 2p25.3 deletion region: the role of MYT1L in intellectual disability and obesity. Genetics in Medicine, 2015, 17, 460-466.	2.4	45
53	Breakpoints around the HOXD cluster result in various limb malformations. Journal of Medical Genetics, 2005, 43, 111-118.	3.2	44
54	Prenatal molecular testing for Beckwithâ€“Wiedemann and Silverâ€“Russell syndromes: a challenge for molecular analysis and genetic counseling. European Journal of Human Genetics, 2016, 24, 784-793.	2.8	44

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55	Evidence of oxidative stress and mitochondrial dysfunction in spinocerebellar ataxia type 2 (SCA2) patient fibroblasts: Effect of coenzyme Q10 supplementation on these parameters. Mitochondrion, 2017, 34, 103-114.	3.4	42
56	Microdeletions ofELP4Are Associated with Language Impairment, Autism Spectrum Disorder, and Mental Retardation. Human Mutation, 2015, 36, 842-850.	2.5	41
57	Association of AADAC Deletion and Gilles de la Tourette Syndrome in a Large European Cohort. Biological Psychiatry, 2016, 79, 383-391.	1.3	41
58	Interstitial deletion 9q22.32â€q33.2 associated with additional familial translocation t(9;17)(q34.11;p11.2) in a patient with Gorlinâ€“Goltz syndrome and features of Nailâ€Patella syndrome. American Journal of Medical Genetics, Part A, 2004, 124A, 179-191.	1.2	38
59	Deletion of 7q34â€“q36.2 in two siblings with mental retardation, language delay, primary amenorrhea, and dysmorphic features. American Journal of Medical Genetics, Part A, 2010, 152A, 3115-3119.	1.2	37
60	Clinical expression of Menkes disease in females with normal karyotype. Orphanet Journal of Rare Diseases, 2012, 7, 6.	2.7	37
61	Deletions and rearrangements of the H19/IGF2 enhancer region in patients with Silver-Russell syndrome and growth retardation. Journal of Medical Genetics, 2011, 48, 308-311.	3.2	35
62	Characterization of a 1.0 Mb YAC contig spannning two chromosome breakpoints related to Menkes disease. Human Molecular Genetics, 1992, 1, 483-489.	2.9	34
63	Menkes Disease: Recent Advances and New Insights into Copper Metabolism. Annals of Medicine, 1996, 28, 121-129.	3.8	34
64	First trimester prenatal diagnosis of Menkes disease by DNA analysis.. Journal of Medical Genetics, 1994, 31, 615-617.	3.2	33
65	Menkes disease: Underlying genetic defect and new diagnostic possibilities. Journal of Inherited Metabolic Disease, 1998, 21, 604-612.	3.6	33
66	<scp>SLC26A4</scp> mutation frequency and spectrum inÂ109 Danish Pendred syndrome/<scp>DFNB4</scp> probands and a report of nine novel mutations. Clinical Genetics, 2013, 84, 388-391.	2.0	33
67	Delineation of an interstitial 9q22 deletion in basal cell nevus syndrome. , 2005, 132A, 324-328.		32
68	Early treatment of Menkes disease with parenteral copper-histidine: long-term follow-up of four treated patients. American Journal of Medical Genetics Part A, 1998, 76, 154-64.	2.4	32
69	X;1 translocation in a female Menkes patient: characterization by fluorescence in situ hybridization. Clinical Genetics, 1994, 46, 295-298.	2.0	31
70	Metaphase FISH on a Chip: Miniaturized Microfluidic Device for Fluorescence in situ Hybridization. Sensors, 2010, 10, 9831-9846.	3.8	30
71	Heterozygous loss-of-function variants of MEIS2 cause a triad of palatal defects, congenital heart defects, and intellectual disability. European Journal of Human Genetics, 2019, 27, 278-290.	2.8	30
72	Delineation of a 2.2 Mb microdeletion at 5q35 associated with microcephaly and congenital heart disease. American Journal of Medical Genetics, Part A, 2006, 140A, 427-433.	1.2	29

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73	Microduplication of 15q13.3 and Xq21.31 in a family with tourette syndrome and comorbidities. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2013, 162, 825-831.	1.7	28
74	Replicative and non-replicative mechanisms in the formation of clustered CNVs are indicated by whole genome characterization. PLoS Genetics, 2018, 14, e1007780.	3.5	28
75	Assignment of the human gene for pregnancy-associated plasma protein A (PAPPA) to 9q33.1 by fluorescence in situ hybridization to mitotic and meiotic chromosomes. Cytogenetic and Genome Research, 1993, 62, 214-216.	1.1	27
76	Analysis of Mnk, the Murine Homologue of the Locus for Menkes Disease, in Normal and Mottled (Mo) Mice. Genomics, 1994, 22, 27-35.	2.9	27
77	Genome-wide DNA methylation analysis of transient neonatal diabetes type 1 patients with mutations in ZFP57. BMC Medical Genetics, 2016, 17, 29.	2.1	27
78	Induced pluripotent stem cell - derived neurons for the study of spinocerebellar ataxia type 3. Stem Cell Research, 2016, 17, 306-317.	0.7	27
79	Congenital Microphthalmia, Anophthalmia and Coloboma among Live Births in Denmark. Ophthalmic Epidemiology, 2016, 23, 324-330.	1.7	27
80	Lethal neonatal Menkes' disease with severe vasculopathy and fractures. Acta Paediatrica, International Journal of Paediatrics, 1998, 87, 1297-1300.	1.5	26
81	A homozygous mutation in a consanguineous family consolidates the role of <scp>ALDH1A3</scp> in autosomal recessive microphthalmia. Clinical Genetics, 2014, 86, 276-281.	2.0	26
82	Clinicianâ€™s guide to genes associated with Rettâ€like phenotypesâ€”Investigation of a Danish cohort and review of the literature. Clinical Genetics, 2019, 95, 221-230.	2.0	26
83	Subtelomeric study of 132 patients with mental retardation reveals 9 chromosomal anomalies and contributes to the delineation of submicroscopic deletions of 1pter, 2qter, 4pter, 5qter and 9qter. BMC Medical Genetics, 2005, 6, 21.	2.1	25
84	Refinement of genotypeâ€phenotype correlation in 18 patients carrying a 1q24q25 deletion. American Journal of Medical Genetics, Part A, 2015, 167, 1008-1017.	1.2	25
85	Screening of 99 Danish Patients with Congenital Heart Disease for GATA4 Mutations. Genetic Testing and Molecular Biomarkers, 2006, 10, 277-280.	1.7	24
86	High frequency of rare copy number variants affecting functionally related genes in patients with structural brain malformations. Human Mutation, 2011, 32, 1427-1435.	2.5	24
87	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
88	X-linked recessive Menkes disease: identification of partial gene deletions in affected males. Clinical Genetics, 2002, 62, 449-457.	2.0	23
89	Breakpoint Cloning and Haplotype Analysis Indicate a Single Origin of the Common Inv(10)(p11.2q21.2) Mutation among Northern Europeans. American Journal of Human Genetics, 2006, 78, 878-883.	6.2	23
90	Congenital imprinting disorders: EUCID.net - a network to decipher their aetiology and to improve the diagnostic and clinical care. Clinical Epigenetics, 2015, 7, 23.	4.1	23

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91	Association Study of CHRNA7 Promoter Variants with Sensory and Sensorimotor Gating in Schizophrenia Patients and Healthy Controls: A Danish Caseâ€“Control Study. NeuroMolecular Medicine, 2015, 17, 423-430.	3.4	23
92	Structural and sequence variants in patients with Silver-Russell syndrome or similar features-Curation of a disease database. Human Mutation, 2018, 39, 345-364.	2.5	23
93	Clinical and molecular delineation of <scp>PUS3</scp>â€associated neurodevelopmental disorders. Clinical Genetics, 2021, 100, 628-633.	2.0	23
94	Neurodevelopmental Disorders Associated with PSD-95 and Its Interaction Partners. International Journal of Molecular Sciences, 2022, 23, 4390.	4.1	23
95	Splice Site Mutations in the ATP7A Gene. PLoS ONE, 2011, 6, e18599.	2.5	22
96	TS-EUROTRAIN: A European-Wide Investigation and Training Network on the Etiology and Pathophysiology of Gilles de la Tourette Syndrome. Frontiers in Neuroscience, 2016, 10, 384.	2.8	21
97	Characterization of a supernumerary small marker X chromosome in two females with similar phenotypes. , 1998, 76, 45-50.		20
98	Characterization of a t(5;8)(q31;q21) translocation in a patient with mental retardation and congenital heart disease: implications for involvement of RUNX1T1 in human brain and heart development. European Journal of Human Genetics, 2009, 17, 1010-1018.	2.8	20
99	Deletion of CUL4B leads to concordant phenotype in a monozygotic twin pair. Clinical Genetics, 2012, 82, 292-294.	2.0	20
100	De novo microdeletions of chromosome 6q14.1-q14.3 and 6q12.1-q14.1 in two patients with intellectual disability - further delineation of the 6q14 microdeletion syndrome and review of the literature. European Journal of Medical Genetics, 2012, 55, 490-497.	1.3	20
101	Fine mapping of a de novo interstitial 10q22â€“q23 duplication in a patient with congenital heart disease and microcephaly. European Journal of Medical Genetics, 2008, 51, 81-86.	1.3	19
102	Epigenetic remodelling and dysregulation of DLGAP4 is linked with early-onset cerebellar ataxia. Human Molecular Genetics, 2014, 23, 6163-6176.	2.9	19
103	Analysis of t(9;17)(q33.2;q25.3) chromosomal breakpoint regions and genetic association reveals novel candidate genes for bipolar disorder. Bipolar Disorders, 2015, 17, 205-211.	1.9	19
104	The D313Y variant in the GLA gene â€“ no evidence of a pathogenic role in Fabry disease. Scandinavian Journal of Clinical and Laboratory Investigation, 2017, 77, 617-621.	1.2	19
105	Prenatal diagnosis of Menkes disease. Prenatal Diagnosis, 1998, 18, 287-289.	2.3	18
106	Expression, purification and copper-binding studies of the first metal-binding domain of Menkes protein. FEBS Journal, 1999, 264, 890-896.	0.2	18
107	Diploid/triploid mosaicism: A rare event or an under-diagnosed syndrome?. European Journal of Medical Genetics, 2011, 54, 374-375.	1.3	18
108	Chromothripsis and DNA Repair Disorders. Journal of Clinical Medicine, 2020, 9, 613.	2.4	18

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109	Detection of genetic defects in Menkes disease by direct mutation analysis and its implications in carrier diagnosis. Journal of Inherited Metabolic Disease, 1994, 17, 267-270.	3.6	17
110	Analysis of a whole arm translocation between chromosomes 18 and 20 using fluorescence in situ hybridization: detection of a break in the centromeric ?-satellite sequences. Human Genetics, 1995, 95, 299-302.	3.8	17
111	Molecular cytogenetic detection of 9q34 breakpoints associated with nail patella syndrome. European Journal of Human Genetics, 1999, 7, 68-76.	2.8	17
112	Menkes Disease and the Occipital Horn Syndrome. , 0, , 651-685.		17
113	Genomic structure, chromosome mapping and expression analysis of the human AVIL gene, and its exclusion as a candidate for locus for inflammatory bowel disease at 12q13â€“14 (IBD2). Gene, 2002, 288, 179-185.	2.2	17
114	Investigation of 4qâ€deletion in two unrelated patients using array CGH. American Journal of Medical Genetics, Part A, 2008, 146A, 2431-2434.	1.2	17
115	X-linked congenital ptosis and associated intellectual disability, short stature, microcephaly, cleft palate, digital and genital abnormalities define novel Xq25q26 duplication syndrome. Human Genetics, 2014, 133, 625-638.	3.8	17
116	Molecular genetics of intracellular copper transport. Journal of Trace Elements in Experimental Medicine, 1999, 12, 297-313.	0.8	16
117	No evidence for pathogenic variants or maternal effect of ZFP57 as the cause of Beckwithâ€“Wiedemann Syndrome. European Journal of Human Genetics, 2012, 20, 119-121.	2.8	16
118	A t(3;9)(q25.1;q34.3) translocation leading to OLFM1 fusion transcripts in Gilles de la Tourette syndrome, OCD and ADHD. Psychiatry Research, 2015, 225, 268-275.	3.3	16
119	Deletion of 11q12.3â€“11q13.1 in a patient with intellectual disability and childhood facial features resembling Cornelia de Lange syndrome. Gene, 2015, 572, 130-134.	2.2	16
120	A novel RAD21 variant associated with intrafamilial phenotypic variation in Cornelia de Lange syndrome â€“ review of the literature. Clinical Genetics, 2017, 91, 647-649.	2.0	16
121	Delineating the psychiatric and behavioral phenotype of recurrent 2q13 deletions and duplications. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2018, 177, 397-405.	1.7	16
122	Expansion of the phenotypic spectrum of de novo missense variants in kinesin family member 1A () Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	2.5	16
123	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
124	Cytogenetically invisible microdeletions involving PITX2 in Rieger syndrome. Clinical Genetics, 2007, 72, 464-470.	2.0	15
125	A 3.2â€‰Mb deletion on 18q12 in a patient with childhood autism and high-grade myopia. European Journal of Human Genetics, 2008, 16, 312-319.	2.8	15
126	Mowatâ€“Wilson syndrome: an underdiagnosed syndrome?. Clinical Genetics, 2008, 73, 579-584.	2.0	15

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127	A duplication encompassing the SHOX gene and the downstream evolutionarily conserved sequences. American Journal of Medical Genetics, Part A, 2009, 149A, 2900-2901.	1.2	15
128	Sequence analysis of SLITRK1 for var321 in Danish patients with Tourette syndrome and review of the literature. Psychiatric Genetics, 2013, 23, 130-133.	1.1	15
129	The <scp>MECP2</scp> variant c.<scp>925C</scp>>T (p.<scp>Arg309Trp</scp>) causes intellectual disability in both males and females without classic features of Rett syndrome. Clinical Genetics, 2016, 89, 733-738.	2.0	15
130	Deciphering the premature mortality in PIGA-CDG â€“ An untold story. Epilepsy Research, 2021, 170, 106530.	1.6	15
131	BBS Proteins Affect Ciliogenesis and Are Essential for Hedgehog Signaling, but Not for Formation of iPSC-Derived RPE-65 Expressing RPE-Like Cells. International Journal of Molecular Sciences, 2021, 22, 1345.	4.1	14
132	Trans-acting genetic variants causing multilocus imprinting disturbance (MLID): common mechanisms and consequences. Clinical Epigenetics, 2022, 14, 41.	4.1	14
133	Human CCS gene: genomic organization and exclusion as a candidate for amyotrophic lateral sclerosis (ALS). BMC Genetics, 2002, 3, 5.	2.7	13
134	X-Linked Menkes Disease: First Documented Report of Germ-Line Mosaicism. Genetic Testing and Molecular Biomarkers, 2004, 8, 286-291.	1.7	13
135	The Variant inv(2)(p11.2q13) Is a Genuinely Recurrent Rearrangement but Displays Some Breakpoint Heterogeneity. American Journal of Human Genetics, 2007, 81, 847-856.	6.2	13
136	FISHprep: A Novel Integrated Device for Metaphase FISH Sample Preparation. Micromachines, 2011, 2, 116-128.	2.9	13
137	Two new Rett syndrome families and review of the literature: expanding the knowledge of MECP2 frameshift mutations. Orphanet Journal of Rare Diseases, 2011, 6, 58.	2.7	13
138	Advanced microtechnologies for detection of chromosome abnormalities by fluorescent in situ hybridization. Biomedical Microdevices, 2012, 14, 453-460.	2.8	13
139	Screening individuals with intellectual disability, autism and Tourette's syndrome for KCNK9 mutations and aberrant DNA methylation within the 8q24 imprinted cluster.. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2014, 165, 472-478.	1.7	13
140	Biochemical analysis of novel NAA10 variants suggests distinct pathogenic mechanisms involving impaired protein N-terminal acetylation. Human Genetics, 2022, 141, 1355-1369.	3.8	13
141	3q27.3 microdeletional syndrome: a recognisable clinical entity associating dysmorphic features, marfanoid habitus, intellectual disability and psychosis with mood disorder. Journal of Medical Genetics, 2014, 51, 21-27.	3.2	12
142	Elevated Expression of SLC6A4 Encoding the Serotonin Transporter (SERT) in Gilles de la Tourette Syndrome. Genes, 2021, 12, 86.	2.4	12
143	GLI1 Is Involved in Cell Cycle Regulation and Proliferation of NT2 Embryonal Carcinoma Stem Cells. DNA and Cell Biology, 2008, 27, 251-256.	1.9	11
144	A cytogenetic survey in Menkes disease: implications for the detection of chromosomal rearrangements in X linked disorders.. Journal of Medical Genetics, 1993, 30, 314-315.	3.2	10

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145	4q35 deletion and 10p15 duplication associated with immunodeficiency. American Journal of Medical Genetics, Part A, 2006, 140A, 2231-2235.	1.2	10
146	Interstitial deletion of chromosome 4p associated with mild mental retardation, epilepsy and polymicrogyria of the left temporal lobe. Clinical Genetics, 2007, 72, 593-598.	2.0	10
147	Balanced translocation in a patient with severe myoclonic epilepsy of infancy disrupts the sodium channel gene SCN1A. Epilepsia, 2008, 49, 1091-1094.	5.1	10
148	Clinical utility gene card for: Axenfeldâ€“Rieger syndrome. European Journal of Human Genetics, 2011, 19, 1-3.	2.8	10
149	Ring chromosome 9 in a girl with developmental delay and dysmorphic features: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2013, 161, 1447-1452.	1.2	10
150	Mosaic MECP2 variants in males with classical Rett syndrome features, including stereotypical hand movements. Clinical Genetics, 2019, 95, 403-408.	2.0	10
151	Stable Longitudinal Methylation Levels at the CpG Sites Flanking the CTG Repeat of DMPK in Patients with Myotonic Dystrophy Type 1. Genes, 2020, 11, 936.	2.4	10
152	High Resolution Analysis of DMPK Hypermethylation and Repeat Interruptions in Myotonic Dystrophy Type 1. Genes, 2022, 13, 970.	2.4	10
153	A small supernumerary marker chromosome X identified by in situ hybridization. Clinical Genetics, 1995, 47, 270-273.	2.0	9
154	Complex distal 10q rearrangement in a girl with mild intellectual disability: Follow up of the patient and review of the literature of nonâ€acrocentric satellited chromosomes. American Journal of Medical Genetics, Part A, 2011, 155, 2841-2854.	1.2	9
155	Centrifugally driven microfluidic disc for detection of chromosomal translocations. Lab on A Chip, 2012, 12, 4628.	6.0	9
156	Case report: a novel KERA mutation associated with cornea plana and its predicted effect on protein function. BMC Medical Genetics, 2015, 16, 40.	2.1	9
157	Generation of spinocerebellar ataxia type 3 patient-derived induced pluripotent stem cell line SCA3.B11. Stem Cell Research, 2016, 16, 589-592.	0.7	9
158	Mitochondrial Function in Gilles de la Tourette Syndrome Patients With and Without Intragenic IMMP2L Deletions. Frontiers in Neurology, 2020, 11, 163.	2.4	9
159	Candidate Genes and Pathways Associated with Gilles de la Tourette Syndromeâ€”Where Are We?. Genes, 2021, 12, 1321.	2.4	9
160	Classification of MSH6 Variants of Uncertain Significance Using Functional Assays. International Journal of Molecular Sciences, 2021, 22, 8627.	4.1	9
161	Lethal neonatal Menkes' disease with severe vasculopathy and fractures. Acta Paediatrica, International Journal of Paediatrics, 1998, 87, 1297-1300.	1.5	9
162	Two new cases with microdeletion of 17q23.2 suggest presence of a candidate gene for sensorineural hearing loss within this region. American Journal of Medical Genetics, Part A, 2011, 155, 2964-2969.	1.2	8

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163	Segregation of a 4p16.3 duplication with a characteristic appearance, macrocephaly, speech delay and mild intellectual disability in a 3â€generation family. American Journal of Medical Genetics, Part A, 2013, 161, 2358-2362.	1.2	8
164	Proximal 21q deletion as a result of a de novo unbalanced t(12;21) translocation in a patient with dysmorphic features, hepatomegaly, thick myocardium and delayed psychomotor development. Molecular Cytogenetics, 2016, 9, 11.	0.9	8
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