Bertrand Isidor

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/708155/publications.pdf

Version: 2024-02-01

209 papers	9,744 citations	38742 50 h-index	56724 83 g-index
222 all docs	222 docs citations	222 times ranked	17695 citing authors

#	Article	IF	Citations
1	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. Genetics in Medicine, 2022, 24, 179-191.	2.4	9
2	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 361-372.	6.2	6
3	Expanding the phenotype of <scp>HNRNPU</scp>â€•related neurodevelopmental disorder with emphasis on seizure phenotype and review of literature. American Journal of Medical Genetics, Part A, 2022, 188, 1497-1514.	1.2	6
4	First evidence of <scp>SOX2</scp> mutations in Peters' anomaly: Lessons from molecular screening of 95 patients. Clinical Genetics, 2022, 101, 494-506.	2.0	9
5	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 601-617.	6.2	16
6	Possible association of 16p11.2 copy number variation with altered lymphocyte and neutrophil counts. Npj Genomic Medicine, 2022, 7, .	3.8	3
7	SETD1B-associated neurodevelopmental disorder. Journal of Medical Genetics, 2021, 58, 196-204.	3.2	22
8	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
9	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
10	Neuropsychological study in 19 French patients with <scp>Whiteâ€Sutton</scp> syndrome and <scp>POGZ</scp> mutations. Clinical Genetics, 2021, 99, 407-417.	2.0	10
11	Pathogenic variants in THSD4, encoding the ADAMTS-like 6 protein, predispose to inherited thoracic aortic aneurysm. Genetics in Medicine, 2021, 23, 111-122.	2.4	25
12	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	2.8	17
13	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
14	Touch and olfaction/taste differentiate children carrying a 16p11.2 deletion from children with ASD. Molecular Autism, 2021, 12, 8.	4.9	6
15	Letter regarding the article â€œtwo girls with short stature, short neck, vertebral anomalies, Sprengel deformity and intellectual disabilityâ€•(Isidor et al., 2015). European Journal of Medical Genetics, 2021, 64, 104179.	1.3	5
16	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	6.2	15
17	Neurodevelopmental phenotypes in individuals with pathogenic variants in CHAMP1. Journal of Physical Education and Sports Management, 2021, 7, a006092.	1.2	9
18	Effects of eight neuropsychiatric copy number variants on human brain structure. Translational Psychiatry, 2021, 11, 399.	4.8	18

#	Article	IF	Citations
19	High rate of hypomorphic variants as the cause of inherited ataxia and related diseases: study of a cohort of 366 families. Genetics in Medicine, 2021, 23, 2160-2170.	2.4	13
20	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159.	2.4	21
21	Imatinib, a New Adjuvant Medical Treatment for Multifocal Villonodular Synovitis Associated to Noonan Syndrome: A Case Report and Literature Review. Frontiers in Medicine, 2021, 8, 817873.	2.6	1
22	Increasing knowledge in IGF1R defects: lessons from 35 new patients. Journal of Medical Genetics, 2020, 57, 160-168.	3.2	20
23	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. Genetics in Medicine, 2020, 22, 181-188.	2.4	30
24	iPSC line derived from a Bloom syndrome patient retains an increased disease-specific sister-chromatid exchange activity.. Stem Cell Research, 2020, 43, 101696.	0.7	4
25	Growth charts in Kabuki syndrome 1. American Journal of Medical Genetics, Part A, 2020, 182, 446-453.	1.2	7
26	Gain-of-Function MN1 Truncation Variants Cause a Recognizable Syndrome with Craniofacial and Brain Abnormalities. American Journal of Human Genetics, 2020, 106, 13-25.	6.2	25
27	Recurrent arginine substitutions in the ACTG2 gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654.	2.5	27
28	Phenotypic spectrum of TGFB3 diseaseâ€causing variants in a Dutchâ€French cohort and first report of a homozygous patient. Clinical Genetics, 2020, 97, 723-730.	2.0	15
29	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	2.4	19
30	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	5.1	17
31	Ribosomopathies: New Therapeutic Perspectives. Cells, 2020, 9, 2080.	4.1	21
32	RLIM Is a Candidate Dosage-Sensitive Gene for Individuals with Varying Duplications of Xq13, Intellectual Disability, and Distinct Facial Features. American Journal of Human Genetics, 2020, 107, 1157-1169.	6.2	6
33	Nextâ€generation sequencing in a series of 80 fetuses with complex cardiac malformations and/or heterotaxy. Human Mutation, 2020, 41, 2167-2178.	2.5	21
34	Psychosocial Impact of Predictive Genetic Testing in Hereditary Heart Diseases: The PREDICT Study. Journal of Clinical Medicine, 2020, 9, 1365.	2.4	9
35	De novo mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. Journal of Medical Genetics, 2020, 57, 808-819.	3.2	11
36	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. American Journal of Human Genetics, 2020, 106, 893-904.	6.2	29

#	Article	IF	Citations
37	Clinical and Molecular Spectrum of Nonsyndromic Earlyâ€Onset Osteoarthritis. Arthritis and Rheumatology, 2020, 72, 1689-1693.	5.6	10
38	De Novo Frameshift Variants in the Neuronal Splicing Factor NOVA2 Result in a Common C-Terminal Extension and Cause a Severe Form of Neurodevelopmental Disorder. American Journal of Human Genetics, 2020, 106, 438-452.	6.2	17
39	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	8.1	121
40	Primrose syndrome: a phenotypic comparison of patients with a ZBTB20 missense variant versus a 3q13.31 microdeletion including ZBTB20. European Journal of Human Genetics, 2020, 28, 1044-1055.	2.8	4
41	A dominant vimentin variant causes a rare syndrome with premature aging. European Journal of Human Genetics, 2020, 28, 1218-1230.	2.8	23
42	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	5.1	65
43	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. American Journal of Human Genetics, 2020, 106, 830-845.	6.2	17
44	Abstract 3595: Riboprotein variant and their role in chondrogenic differentiation and osteosarcoma development. , 2020, , .		0
45	A step towards precision medicine in management of severe transient polyhydramnios: MAGED2 variant. Journal of Obstetrics and Gynaecology, 2019, 39, 395-397.	0.9	6
46	Oro-dental phenotype in patients with RUNX2 duplication. European Journal of Medical Genetics, 2019, 62, 85-89.	1.3	9
47	Expanding the phenotype of the X-linked BCOR microphthalmia syndromes. Human Genetics, 2019, 138, 1051-1069.	3.8	35
48	Encephalopathies with KCNC1 variants: genotypeâ€phenotypeâ€functional correlations. Annals of Clinical and Translational Neurology, 2019, 6, 1263-1272.	3.7	33
49	Genotype/phenotype correlations of childhoodâ€onset congenital sideroblastic anaemia in a European cohort. British Journal of Haematology, 2019, 187, 530-542.	2.5	18
50	SETD2 related overgrowth syndrome: Presentation of four new patients and review of the literature. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 509-518.	1.6	24
51	Developmental trajectories of neuroanatomical alterations associated with the 16p11.2 Copy Number Variations. NeuroImage, 2019, 203, 116155.	4.2	9
52	RPL13 Variants Cause Spondyloepimetaphyseal Dysplasia with Severe Short Stature. American Journal of Human Genetics, 2019, 105, 1040-1047.	6.2	17
53	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	2.3	36
54	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	2.4	41

#	Article	IF	Citations
55	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. Genetics in Medicine, 2019, 21, 2025-2035.	2.4	40
56	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. Human Mutation, 2019, 40, 1993-2000.	2.5	4
57	Pycnodysostosis: Natural history and management guidelines from 27 French cases and a literature review. Clinical Genetics, 2019, 96, 309-316.	2.0	31
58	Novel mutations in NLGN3 causing autism spectrum disorder and cognitive impairment. Human Mutation, 2019, 40, 2021-2032.	2.5	42
59	De novo loss-of-function KCNMA1 variants are associated with a new multiple malformation syndrome and a broad spectrum of developmental and neurological phenotypes. Human Molecular Genetics, 2019, 28, 2937-2951.	2.9	76
60	Variants in MED12L, encoding a subunit of the mediator kinase module, are responsible for intellectual disability associated with transcriptional defect. Genetics in Medicine, 2019, 21, 2713-2722.	2.4	28
61	Clinical, Histopathological, and Molecular Diagnostics in Lethal Lung Developmental Disorders. American Journal of Respiratory and Critical Care Medicine, 2019, 200, 1093-1101.	5.6	47
62	Searching for secondary findings: considering actionability and preserving the right not to know. European Journal of Human Genetics, 2019, 27, 1481-1484.	2.8	13
63	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
64	Exome sequencing identifies a novel missense variant in CTSC causing nonsyndromic aggressive periodontitis. Journal of Human Genetics, 2019, 64, 689-694.	2.3	8
65	Population genetic screening: current issues in a European country. European Journal of Human Genetics, 2019, 27, 1321-1323.	2.8	2
66	Biallelic MYORG mutation carriers exhibit primary brain calcification with a distinct phenotype. Brain, 2019, 142, 1573-1586.	7.6	49
67	Variable expressivity of syndromic BMP4-related eye, brain, and digital anomalies: A review of the literature and description of three new cases. European Journal of Human Genetics, 2019, 27, 1379-1388.	2.8	8
68	Bilateral retinoblastoma due to a germline mutation of RB1 in a child with down syndrome. Ophthalmic Genetics, 2019, 40, 86-86.	1.2	1
69	Autosomal recessive Treacher Collins syndrome due to POLR1C mutations: Report of a new family and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 1390-1394.	1.2	19
70	Whole genome paired-end sequencing elucidates functional and phenotypic consequences of balanced chromosomal rearrangement in patients with developmental disorders. Journal of Medical Genetics, 2019, 56, 526-535.	3.2	46
71	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
72	Acanthosis nigricans, hypochondroplasia, and FGFR 3 mutations: Findings with five new patients, and a review of the literature. Pediatric Dermatology, 2019, 36, 242-246.	0.9	6

#	Article	IF	Citations
73	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.	3.2	43
74	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. Genetics in Medicine, 2019, 21, 816-825.	2.4	127
75	Clinical and functional characterization of recurrent missense variants implicated inTHOC6-related intellectual disability. Human Molecular Genetics, 2019, 28, 952-960.	2.9	23
76	A de novo 2q37.2 deletion encompassing AGAP1 and SH3BP4 in a patient with autism and intellectual disability. European Journal of Medical Genetics, 2019, 62, 103586.	1.3	12
77	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	6.2	30
78	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
79	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
80	KAT6A Syndrome: genotypeâ€“phenotype correlation in 76 patients with pathogenic KAT6A variants. Genetics in Medicine, 2019, 21, 850-860.	2.4	68
81	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
82	Duplications at 19q13.33 in patients with neurodevelopmental disorders. Neurology: Genetics, 2018, 4, e210.	1.9	4
83	Diagnostic strategy in segmentation defect of the vertebrae: a retrospective study of 73 patients. Journal of Medical Genetics, 2018, 55, 422.2-429.	3.2	14
84	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. Biological Psychiatry, 2018, 84, 253-264.	1.3	56
85	Further delineation of theMECP2duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features. Journal of Medical Genetics, 2018, 55, 359-371.	3.2	45
86	A novel mutation in the transmembrane 6 domain of GABBR2 leads to a Rettâ€like phenotype. Annals of Neurology, 2018, 83, 437-439.	5.3	19
87	Rare Coding Variants in ANGPTL6 Are Associated with Familial Forms of Intracranial Aneurysm. American Journal of Human Genetics, 2018, 102, 133-141.	6.2	37
88	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
89	Familial autosomal dominant severe ankyloglossia with tooth abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 1614-1617.	1.2	10
90	Familial deep endometriosis: A rare monogenic disease?. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2018, 221, 190-193.	1.1	5

#	Article	IF	Citations
91	Delineating FOXG1 syndrome. Neurology: Genetics, 2018, 4, e281.	1.9	51
92	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
93	Chromosome 14q32.2 Imprinted Region Disruption as an Alternative Molecular Diagnosis of Silver-Russell Syndrome. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2436-2446.	3.6	48
94	De Novo Variants in the F-Box Protein FBXO11 in 20 Individuals with a Variable Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 103, 305-316.	6.2	48
95	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscyphodysplasia. European Journal of Human Genetics, 2018, 26, 1611-1622.	2.8	18
96	Novel SUZ12 mutations in Weaverâ€like syndrome. Clinical Genetics, 2018, 94, 461-466.	2.0	36
97	Natural history of GATA2 deficiency in a survey of 79 French and Belgian patients. Haematologica, 2018, 103, 1278-1287.	3.5	129
98	New splicing pathogenic variant in EBP causing extreme familial variability of Conradiâ€“HÃ¼nermannâ€“Happle Syndrome. European Journal of Human Genetics, 2018, 26, 1784-1790.	2.8	7
99	Microduplication in the 2p16.1p15 chromosomal region linked to developmental delay and intellectual disability. Molecular Cytogenetics, 2018, 11, 39.	0.9	4
100	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	6.2	37
101	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. Genetics in Medicine, 2017, 19, 989-997.	2.4	90
102	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
103	Barber-Say Syndrome and Ablepharon-Macrostomia Syndrome: A Patient's View. Molecular Syndromology, 2017, 8, 172-178.	0.8	6
104	Understanding the Pathophysiology of Intracranial Aneurysm: The ICAN Project. Neurosurgery, 2017, 80, 621-626.	1.1	22
105	Sex chromosome aneuploidies and copy-number variants: a further explanation for neurodevelopmental prognosis variability?. European Journal of Human Genetics, 2017, 25, 930-934.	2.8	19
106	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	6.2	54
107	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Human Genetics, 2017, 136, 463-479.	3.8	66
108	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127.	6.2	62

#	Article	IF	Citations
109	Novel KCNB1 mutation associated with non-syndromic intellectual disability. Journal of Human Genetics, 2017, 62, 569-573.	2.3	28
110	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	6.2	136
111	Mutations in GREB1L Cause Bilateral Kidney Agenesis in Humans and Mice. American Journal of Human Genetics, 2017, 101, 803-814.	6.2	76
112	Two novel variants in CNTNAP1 in two siblings presenting with congenital hypotonia and hypomyelinating neuropathy. European Journal of Human Genetics, 2017, 25, 150-152.	2.8	13
113	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
114	Novel promoters and coding first exons in DLG2 linked to developmental disorders and intellectual disability. Genome Medicine, 2017, 9, 67.	8.2	29
115	Mutations in signal recognition particle SRP54 cause syndromic neutropenia with Shwachman-Diamondâ€“like features. Journal of Clinical Investigation, 2017, 127, 4090-4103.	8.2	126
116	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
117	Protein-altering MYH3 variants are associated with a spectrum of phenotypes extending to spondylocarpotarsal synostosis syndrome. European Journal of Human Genetics, 2016, 24, 1746-1751.	2.8	21
118	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. Journal of Human Genetics, 2016, 61, 835-838.	2.3	27
119	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	6.2	102
120	Mutations in the HECT domain of NEDD4L lead to AKTâ€“mTOR pathway deregulation and cause periventricular nodular heterotopia. Nature Genetics, 2016, 48, 1349-1358.	21.4	101
121	Large national series of patients with Xq28 duplication involving MECP2: Delineation of brain MRI abnormalities in 30 affected patients. American Journal of Medical Genetics, Part A, 2016, 170, 116-129.	1.2	19
122	Oncologic Phenotype of Peripheral Neuroblastic Tumors Associated With PHOX2B Nonâ€Polyalanine Repeat Expansion Mutations. Pediatric Blood and Cancer, 2016, 63, 71-77.	1.5	14
123	Mandibular dysostosis without microphthalmia caused by OTX2 deletion. American Journal of Medical Genetics, Part A, 2016, 170, 2466-2470.	1.2	4
124	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	6.2	45
125	Autosomal-Recessive Mutations in AP3B2, Adaptor-Related Protein Complex 3 Beta 2 Subunit, Cause an Early-Onset Epileptic Encephalopathy with Optic Atrophy. American Journal of Human Genetics, 2016, 99, 1368-1376.	6.2	46
126	Contactin-Associated Protein 1 (CNTNAP1) Mutations Induce Characteristic Lesions of the Paranodal Region. Journal of Neuropathology and Experimental Neurology, 2016, 75, 1155-1159.	1.7	31

#	Article	IF	Citations
127	Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764.	2.5	70
128	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	2.5	40
129	The Number of Genomic Copies at the 16p11.2 Locus Modulates Language, Verbal Memory, and Inhibition. Biological Psychiatry, 2016, 80, 129-139.	1.3	78
130	Mutations in RIT1 cause Noonan syndrome with possible juvenile myelomonocytic leukemia but are not involved in acute lymphoblastic leukemia. European Journal of Human Genetics, 2016, 24, 1124-1131.	2.8	23
131	The expanding spectrum of COL2A1 gene variants IN 136 patients with a skeletal dysplasia phenotype. European Journal of Human Genetics, 2016, 24, 992-1000.	2.8	39
132	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	11.0	195
133	Abnormal spindle-like microcephaly-associated (ASPM) mutations strongly disrupt neocortical structure but spare the hippocampus and long-term memory. Cortex, 2016, 74, 158-176.	2.4	32
134	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. Genetics in Medicine, 2016, 18, 49-56.	2.4	125
135	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. PLoS ONE, 2016, 11, e0150555.	2.5	32
136	Neonatal Marfan Syndrome: Report of a Case with an Inherited Splicing Mutation outside the Neonatal Domain. Molecular Syndromology, 2015, 6, 281-286.	0.8	5
137	Patients with isolated oligo/hypodontia caused by RUNX2 duplication. American Journal of Medical Genetics, Part A, 2015, 167, 1386-1390.	1.2	10
138	A de novoADCY5 mutation causes earlyâ€onset autosomal dominant chorea and dystonia. Movement Disorders, 2015, 30, 423-427.	3.9	131
139	Epileptic patients with de novo <scp>STXBP</scp>1 mutations: Key clinical features based on 24 cases. Epilepsia, 2015, 56, 1931-1940.	5.1	44
140	Muscle magnetic resonance imaging abnormalities in Xâ€linked myopathy with excessive autophagy. Muscle and Nerve, 2015, 52, 673-680.	2.2	6
141	Single amino acid charge switch defines clinically distinct proline-serine-threonine phosphatase-interacting protein 1 (PSTPIP1)â€"associated inflammatory diseases. Journal of Allergy and Clinical Immunology, 2015, 136, 1337-1345.	2.9	103
142	A recurrent KCNQ2 pore mutation causing early onset epileptic encephalopathy has a moderate effect on M current but alters subcellular localization of Kv7 channels. Neurobiology of Disease, 2015, 80, 80-92.	4.4	59
143	Two girls with short stature, short neck, vertebral anomalies, Sprengel deformity and intellectual disability. European Journal of Medical Genetics, 2015, 58, 47-50.	1.3	11
144	Characterization of human disease phenotypes associated with mutations in TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, ADAR, and IFIH1. American Journal of Medical Genetics, Part A, 2015, 167, 296-312.	1.2	447

#	Article	IF	Citations
145	Five children with deletions of 1p34.3 encompassing AGO1 and AGO3. European Journal of Human Genetics, 2015, 23, 761-765.	2.8	25
146	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	2.8	53
147	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. European Journal of Human Genetics, 2015, 23, 1473-1481.	2.8	101
148	Ten new cases further delineate the syndromic intellectual disability phenotype caused by mutations in DYRK1A. European Journal of Human Genetics, 2015, 23, 1482-1487.	2.8	62
149	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. American Journal of Human Genetics, 2015, 96, 784-796.	6.2	53
150	Mutations of the ImprintedCDKN1CGene as a Cause of the Overgrowth Beckwith-Wiedemann Syndrome: Clinical Spectrum and Functional Characterization. Human Mutation, 2015, 36, 894-902.	2.5	62
151	USP7 Acts as a Molecular Rheostat to Promote WASH-Dependent Endosomal Protein Recycling and Is Mutated in a Human Neurodevelopmental Disorder. Molecular Cell, 2015, 59, 956-969.	9.7	175
152	Genetics of giant cell tumors of bone. , 2015, , 341-345.		0
153	Genomic aberrations of the CACNA2D1 gene in three patients with epilepsy and intellectual disability. European Journal of Human Genetics, 2015, 23, 628-632.	2.8	58
154	New insights into genotypeâ€"phenotype correlation for GLI3 mutations. European Journal of Human Genetics, 2015, 23, 92-102.	2.8	97
155	A new mutation in the C-SH2 domain of PTPN11 causes Noonan syndrome with multiple giant cell lesions. Journal of Human Genetics, 2014, 59, 57-59.	2.3	13
156	An emerging phenotype of Xq22 microdeletions in females with severe intellectual disability, hypotonia and behavioral abnormalities. Journal of Human Genetics, 2014, 59, 300-306.	2.3	29
157	Gain-of-function mutations in IFIH1 cause a spectrum of human disease phenotypes associated with upregulated type I interferon signaling. Nature Genetics, 2014, 46, 503-509.	21.4	490
158	Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. Journal of Medical Genetics, 2014, 51, 724-736.	3.2	229
159	Mutations in SLC13A5 Cause Autosomal-Recessive Epileptic Encephalopathy with Seizure Onset in the First Days of Life. American Journal of Human Genetics, 2014, 95, 113-120.	6.2	112
160	Blepharophimosis, short humeri, developmental delay and hirschsprung disease: Expanding the phenotypic spectrum of MED12 mutations. American Journal of Medical Genetics, Part A, 2014, 164, 1821-1825.	1.2	19
161	Clinical spectrum of females with HCCS mutation: from no clinical signs to a neonatal lethal form of the microphthalmia with linear skin defects (MLS) syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 53.	2.7	26
162	Nephrocalcinosis (Enamel Renal Syndrome) Caused by Autosomal Recessive FAM20A Mutations. Nephron Physiology, 2013, 122, 1-6.	1.2	84

#	Article	IF	Citations
163	Similar early characteristics but variable neurological outcome of patients with a de novo mutation of KCNQ2. Orphanet Journal of Rare Diseases, 2013, 8, 80.	2.7	82
164	Refining the phenotype associated with MEF2C point mutations. Neurogenetics, 2013, 14, 71-75.	1.4	60
165	Palmoâ€Plantar hyperkeratosis, intellectual disability, and spastic paraplegia in two maternal half brothers: Further evidence for an Xâ€linked inheritance. American Journal of Medical Genetics, Part A, 2013, 161, 1390-1393.	1.2	1
166	Exudative retinopathy, cerebral calcifications, duodenal atresia, preaxial polydactyly, micropenis, microcephaly and short stature: A new syndrome?. American Journal of Medical Genetics, Part A, 2013, 161, 1829-1832.	1.2	2
167	Renal phenotypic variability in HDR syndrome: glomerular nephropathy as a novel finding. European Journal of Pediatrics, 2013, 172, 107-110.	2.7	23
168	Treatment responses in five patients with ribbing disease including two with 466C>T missense mutations in TGFÎ²1. Joint Bone Spine, 2013, 80, 638-644.	1.6	11
169	A novel microdeletion syndrome at 9q21.13 characterised by mental retardation, speech delay, epilepsy and characteristic facial features. European Journal of Medical Genetics, 2013, 56, 163-170.	1.3	35
170	Wilmsâ€™ tumor in patients with 9q22.3 microdeletion syndrome suggests a role for PTCH1 in nephroblastomas. European Journal of Human Genetics, 2013, 21, 784-787.	2.8	26
171	Finger creases lend a hand in Kabuki syndrome. European Journal of Medical Genetics, 2013, 56, 556-560.	1.3	11
172	A new form of severe spondyloepimetaphyseal dysplasia: Clinical and radiological characterization. , 2013, 161A, n/a-n/a.		4
173	GATAD2Bloss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth inDrosophila. Journal of Medical Genetics, 2013, 50, 507-514.	3.2	63
174	Mutations in STAMBP, encoding a deubiquitinating enzyme, cause microcephalyâ€"capillary malformation syndrome. Nature Genetics, 2013, 45, 556-562.	21.4	94
175	ALK germline mutations in patients with neuroblastoma: a rare and weakly penetrant syndrome. European Journal of Human Genetics, 2012, 20, 291-297.	2.8	38
176	Inactive matriptase-2 mutants found in IRIDA patients still repress hepcidin in a transfection assay despite having lost their serine protease activity. Human Mutation, 2012, 33, 1388-1396.	2.5	25
177	De novo duplication and deletions at 7q in a threeâ€generation family. American Journal of Medical Genetics, Part A, 2012, 158A, 1493-1497.	1.2	1
178	Molecular characterization of 1q44 microdeletion in 11 patients reveals three candidate genes for intellectual disability and seizures. American Journal of Medical Genetics, Part A, 2012, 158A, 1633-1640.	1.2	63
179	Progressive polyepiphyseal dysplasia with arthropathy: A distinct disorder from idiopathic juvenile arthritis and pseudorheumatoid dysplasia?. American Journal of Medical Genetics, Part A, 2012, 158A, 1754-1758.	1.2	3
180	Nablus maskâ€like facial syndrome: Deletion of chromosome 8q22.1 is necessary but not sufficient to cause the phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2091-2099.	1.2	9

#	Article	IF	Citations
181	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	6.2	141
182	Non-USH2A mutations in USH2 patients. Human Mutation, 2012, 33, 504-510.	2.5	57
183	Haploinsufficiency ofSOX5at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. Human Mutation, 2012, 33, 728-740.	2.5	85
184	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
185	Phenotypic spectrum associated with CASK loss-of-function mutations. Journal of Medical Genetics, 2011, 48, 741-751.	3.2	114
186	Truncating mutations in the last exon of NOTCH2 cause a rare skeletal disorder with osteoporosis. Nature Genetics, 2011, 43, 306-308.	21.4	181
187	A new microdeletion syndrome of 5q31.3 characterized by severe developmental delays, distinctive facial features, and delayed myelination. , 2011, 155, 732-736.		43
188	Multiple capillary skin malformations, epilepsy, microcephaly, mental retardation, hypoplasia of the distal phalanges: Report of a new case and further delineation of a new syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1458-1460.	1.2	18
189	Homozygous PTEN deletion in neuroblastoma arising in a child with Cowden syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1763-1766.	1.2	3
190	Corrigendum to â€œA New Microdeletion Syndrome of 5q31.3 Characterized by Severe Developmental Delays, Distinctive Facial Features, and Delayed Myelinationâ€•, 2011, 155, 2903-2903.		1
191	Serpentine fibula-polycystic kidney syndrome caused by truncating mutations in NOTCH2. Human Mutation, 2011, 32, 1239-1242.	2.5	29
192	Expanding the Spectrum of PMM2-CDG Phenotype. JIMD Reports, 2011, 5, 123-125.	1.5	13
193	Mesomelia-Synostoses Syndrome Results from Deletion of SULF1 and SLCO5A1 Genes at 8q13. American Journal of Human Genetics, 2010, 87, 95-100.	6.2	42
194	NF1 microdeletions in neurofibromatosis type 1: from genotype to phenotype. Human Mutation, 2010, 31, E1506-E1518.	2.5	208
195	Deletion of the CUL4B gene in a boy with mental retardation, minor facial anomalies, short stature, hypogonadism, and ataxia. American Journal of Medical Genetics, Part A, 2010, 152A, 175-180.	1.2	50
196	Axial spondylometaphyseal dysplasia: Confirmation and further delineation of a new SMD with retinal dystrophy. American Journal of Medical Genetics, Part A, 2010, 152A, 1550-1554.	1.2	8
197	Mesomelic dysplasia Kantaputra type is associated with duplications of the HOXD locus on chromosome 2q. European Journal of Human Genetics, 2010, 18, 1310-1314.	2.8	32
198	CYP7B1 mutations in pure and complex forms of hereditary spastic paraplegia type 5. Brain, 2009, 132, 1589-1600.	7.6	102

#	Article	IF	Citations
199	Familial Frameshift SRY Mutation Inherited from a Mosaic Father with Testicular Dysgenesis Syndrome. Journal of Clinical Endocrinology and Metabolism, 2009, 94, 3467-3471.	3.6	37
200	Coneâ€rod dystrophy, growth hormone deficiency and spondyloepiphyseal dysplasia: Report of a new case without nephronophtisis. American Journal of Medical Genetics, Part A, 2009, 149A, 788-792.	1.2	5
201	Mesomelic dysplasia with acral synostoses Verloesâ€"Davidâ€"Pfeiffer type: Followâ€up study documents progressive clinical course. American Journal of Medical Genetics, Part A, 2009, 149A, 2220-2225.	1.2	3
202	Mutational, functional, and expression studies of theTCF4gene in Pitt-Hopkins syndrome. Human Mutation, 2009, 30, 669-676.	2.5	126
203	Thromboxane synthase mutations in an increased bone density disorder (Ghosal syndrome). Nature Genetics, 2008, 40, 284-286.	21.4	61
204	Inherited 18q23 duplication in a fetus with multiple congenital anomalies. European Journal of Medical Genetics, 2008, 51, 231-238.	1.3	5
205	Complex constitutional subtelomeric 1p36.3 deletion/duplication in a mentally retarded child with neonatal neuroblastoma. European Journal of Medical Genetics, 2008, 51, 679-684.	1.3	20
206	A 8.26Mb deletion in 6q16 and a 4.95Mb deletion in 20p12 including JAG1 and BMP2 in a patient with Alagille syndrome and Wolffâ€"Parkinsonâ€"White syndrome. European Journal of Medical Genetics, 2008, 51, 651-657.	1.3	22
207	Genochondromatosis type II: Report of a new patient and further delineation of the phenotype. American Journal of Medical Genetics, Part A, 2007, 143A, 1919-1921.	1.2	4
208	Third case of paternal isodisomy for chromosome 7 with cystic fibrosis: A new patient presenting with normal growth. American Journal of Medical Genetics, Part A, 2007, 143A, 2696-2699.	1.2	26
209	Blepharophimosis-mental retardation (BMR) syndromes: A proposed clinical classification of the so-called Ohdo syndrome, and delineation of two new BMR syndromes, one X-linked and one autosomal recessive. American Journal of Medical Genetics, Part A, 2006, 140A, 1285-1296.	1.2	73