Bertrand Isidor

List of Publications by Year
in descending order

209	9,744	38742 50	56724 83
papers	citations	h-index	g-index

222	222	222	17695
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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
2	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
3	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. Nature, 2011, 478, 97-102.	27.8	394
4	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
5	NF1 microdeletions in neurofibromatosis type 1: from genotype to phenotype. Human Mutation, 2010, 31, E1506-E1518.	2.5	208
6	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. JAMA Psychiatry, 2016, 73, 20.	11.0	195
7	Truncating mutations in the last exon of NOTCH2 cause a rare skeletal disorder with osteoporosis. Nature Genetics, 2011, 43, 306-308.	21.4	181
8	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
9	CantÃº Syndrome Is Caused by Mutations in ABCC9. American Journal of Human Genetics, 2012, 90, 1094-1101.	6.2	141
10	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
11	Mutation Update for Kabuki Syndrome GenesKMT2DandKDM6Aand Further Delineation of X-Linked Kabuki Syndrome Subtype 2. Human Mutation, 2016, 37, 847-864.	2.5	134
12	A de novoADCY5 mutation causes earlyâ€onset autosomal dominant chorea and dystonia. Movement Disorders, 2015, 30, 423-427.	3.9	131
13	Natural history of GATA2 deficiency in a survey of 79 French and Belgian patients. Haematologica, 2018, 103, 1278-1287.	3.5	129
14	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
15	Mutational, functional, and expression studies of theTCF4gene in Pitt-Hopkins syndrome. Human Mutation, 2009, 30, 669-676.	2.5	126
16	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
17	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
18	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	8.1	121

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19	Phenotypic spectrum associated with CASK loss-of-function mutations. Journal of Medical Genetics, 2011, 48, 741-751.	3.2	114
20	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
21	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	1.3	108
22	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
23	CYP7B1 mutations in pure and complex forms of hereditary spastic paraplegia type 5. Brain, 2009, 132, 1589-1600.	7.6	102
24	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
25	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
26	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
27	New insights into genotypeâ€“phenotype correlation for GLI3 mutations. European Journal of Human Genetics, 2015, 23, 92-102.	2.8	97
28	Mutations in STAMBP, encoding a deubiquitinating enzyme, cause microcephalyâ€“capillary malformation syndrome. Nature Genetics, 2013, 45, 556-562.	21.4	94
29	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
30	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
31	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
32	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
33	Nephrocalcinosis (Enamel Renal Syndrome) Caused by Autosomal Recessive FAM20A Mutations. Nephron Physiology, 2013, 122, 1-6.	1.2	84
34	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
35	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
36	Mutations in GREB1L Cause Bilateral Kidney Agenesis in Humans and Mice. American Journal of Human Genetics, 2017, 101, 803-814.	6.2	76

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37	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
38	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
39	Expanding the Phenotype Associated with NAA10â€Related Nâ€Terminal Acetylation Deficiency. Human Mutation, 2016, 37, 755-764.	2.5	70
40	KAT6A Syndrome: genotypeâ€“phenotype correlation in 76 patients with pathogenic KAT6A variants. Genetics in Medicine, 2019, 21, 850-860.	2.4	68
41	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
42	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. Epilepsia, 2020, 61, 387-399.	5.1	65
43	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
44	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
45	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
46	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
47	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
48	Thromboxane synthase mutations in an increased bone density disorder (Ghosal syndrome). Nature Genetics, 2008, 40, 284-286.	21.4	61
49	Refining the phenotype associated with MEF2C point mutations. Neurogenetics, 2013, 14, 71-75.	1.4	60
50	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
51	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
52	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. Npj Genomic Medicine, 2017, 2, 32.	3.8	58
53	Non-USH2A mutations in USH2 patients. Human Mutation, 2012, 33, 504-510.	2.5	57
54	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

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55	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. American Journal of Human Genetics, 2019, 104, 1210-1222.	6.2	56
56	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
57	Redefining the MED13L syndrome. European Journal of Human Genetics, 2015, 23, 1308-1317.	2.8	53
58	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
59	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
60	Delineating FOXG1 syndrome. Neurology: Genetics, 2018, 4, e281.	1.9	51
61	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
62	Biallelic MYORG mutation carriers exhibit primary brain calcification with a distinct phenotype. Brain, 2019, 142, 1573-1586.	7.6	49
63	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
64	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
65	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
66	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
67	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
68	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
69	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
70	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
71	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
72	A new microdeletion syndrome of 5q31.3 characterized by severe developmental delays, distinctive facial features, and delayed myelination. , 2011, 155, 732-736.		43

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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	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
75	Novel mutations in NLGN3 causing autism spectrum disorder and cognitive impairment. Human Mutation, 2019, 40, 2021-2032.	2.5	42
76	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
77	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	2.5	40
78	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
79	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
80	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
81	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
82	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
83	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
84	Novel SUZ12 mutations in Weaverâ€like syndrome. Clinical Genetics, 2018, 94, 461-466.	2.0	36
85	Genetic abnormalities in a large cohort of Coffinâ€“Siris syndrome patients. Journal of Human Genetics, 2019, 64, 1173-1186.	2.3	36
86	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
87	Expanding the phenotype of the X-linked BCOR microphthalmia syndromes. Human Genetics, 2019, 138, 1051-1069.	3.8	35
88	Encephalopathies with KCNC1 variants: genotypeâ€phenotypeâ€functional correlations. Annals of Clinical and Translational Neurology, 2019, 6, 1263-1272.	3.7	33
89	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
90	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

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91	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. PLoS ONE, 2016, 11, e0150555.	2.5	32
92	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
93	Pycnodysostosis: Natural history and management guidelines from 27 French cases and a literature review. Clinical Genetics, 2019, 96, 309-316.	2.0	31
94	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
95	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	6.2	30
96	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
97	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. Genetics in Medicine, 2020, 22, 181-188.	2.4	30
98	Serpentine fibula-polycystic kidney syndrome caused by truncating mutations in NOTCH2. Human Mutation, 2011, 32, 1239-1242.	2.5	29
99	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
100	Novel promoters and coding first exons in DLG2 linked to developmental disorders and intellectual disability. Genome Medicine, 2017, 9, 67.	8.2	29
101	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. American Journal of Human Genetics, 2020, 106, 893-904.	6.2	29
102	Novel KCNB1 mutation associated with non-syndromic intellectual disability. Journal of Human Genetics, 2017, 62, 569-573.	2.3	28
103	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
104	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	2.4	28
105	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. Journal of Human Genetics, 2016, 61, 835-838.	2.3	27
106	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
107	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
108	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

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109	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
110	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
111	Five children with deletions of 1p34.3 encompassing AGO1 and AGO3. European Journal of Human Genetics, 2015, 23, 761-765.	2.8	25
112	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
113	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
114	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
115	Renal phenotypic variability in HDR syndrome: glomerular nephropathy as a novel finding. European Journal of Pediatrics, 2013, 172, 107-110.	2.7	23
116	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
117	Clinical and functional characterization of recurrent missense variants implicated inTHOC6-related intellectual disability. Human Molecular Genetics, 2019, 28, 952-960.	2.9	23
118	A dominant vimentin variant causes a rare syndrome with premature aging. European Journal of Human Genetics, 2020, 28, 1218-1230.	2.8	23
119	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
120	Understanding the Pathophysiology of Intracranial Aneurysm: The ICAN Project. Neurosurgery, 2017, 80, 621-626.	1.1	22
121	SETD1B-associated neurodevelopmental disorder. Journal of Medical Genetics, 2021, 58, 196-204.	3.2	22
122	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
123	Ribosomopathies: New Therapeutic Perspectives. Cells, 2020, 9, 2080.	4.1	21
124	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
125	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. Genetics in Medicine, 2021, 23, 2150-2159.	2.4	21
126	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

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127	Increasing knowledge in IGF1R defects: lessons from 35 new patients. Journal of Medical Genetics, 2020, 57, 160-168.	3.2	20
128	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
129	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
130	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
131	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
132	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
133	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
134	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
135	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
136	Genotype/phenotype correlations of childhoodâ€onset congenital sideroblastic anaemia in a European cohort. British Journal of Haematology, 2019, 187, 530-542.	2.5	18
137	Effects of eight neuropsychiatric copy number variants on human brain structure. Translational Psychiatry, 2021, 11, 399.	4.8	18
138	RPL13 Variants Cause Spondyloepimetaphyseal Dysplasia with Severe Short Stature. American Journal of Human Genetics, 2019, 105, 1040-1047.	6.2	17
139	Developmental and epilepsy spectrum of KCNB1 encephalopathy with longâ€term outcome. Epilepsia, 2020, 61, 2461-2473.	5.1	17
140	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
141	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
142	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
143	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
144	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

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145	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
146	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
147	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
148	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
149	Expanding the Spectrum of PMM2-CDG Phenotype. JIMD Reports, 2011, 5, 123-125.	1.5	13
150	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
151	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
152	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
153	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
154	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
155	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
156	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
157	Finger creases lend a hand in Kabuki syndrome. European Journal of Medical Genetics, 2013, 56, 556-560.	1.3	11
158	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
159	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
160	Patients with isolated oligo/hypodontia caused by RUNX2 duplication. American Journal of Medical Genetics, Part A, 2015, 167, 1386-1390.	1.2	10
161	Familial autosomal dominant severe ankyloglossia with tooth abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 1614-1617.	1.2	10
162	Clinical and Molecular Spectrum of Nonsyndromic Earlyâ€Onset Osteoarthritis. Arthritis and Rheumatology, 2020, 72, 1689-1693.	5.6	10

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