Bertrand Isidor

List of Publications by Year in Descending Order

205 papers	6,460 citations	45 h-index	71 g-index
222 ext. papers	8,306 ext. citations	6.6 avg, IF	4.65 L-index

#	Paper	IF	Citations
205	Germline variants in tumor suppressor FBXW7 lead to impaired ubiquitination and a neurodevelopmental syndrome.. *American Journal of Human Genetics*, **2022**, 109, 601-617	11	0
204	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	4.9	0
203	Letter regarding the article "two girls with short stature, short neck, vertebral anomalies, Sprengel deformity and intellectual disability" (Isidor et al., 2015). *European Journal of Medical Genetics*, **2021**, 64, 104179	2.6	0
202	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	11	0
201	Neurodevelopmental phenotypes in individuals with pathogenic variants in. *Journal of Physical Education and Sports Management*, **2021**, 7,	2.8	1
200	Effects of eight neuropsychiatric copy number variants on human brain structure. *Translational Psychiatry*, **2021**, 11, 399	8.6	3
199	-associated neurodevelopmental disorder. *Journal of Medical Genetics*, **2021**, 58, 196-204	5.8	4
198	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. *Genetics in Medicine*, **2021**, 23, 363-373	8.1	4
197	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. *Genetics in Medicine*, **2021**, 23, 374-383	8.1	0
196	Neuropsychological study in 19 French patients with White-Sutton syndrome and POGZ mutations. *Clinical Genetics*, **2021**, 99, 407-417	4	5
195	Pathogenic variants in THSD4, encoding the ADAMTS-like 6 protein, predispose to inherited thoracic aortic aneurysm. *Genetics in Medicine*, **2021**, 23, 111-122	8.1	7
194	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	5.3	4
193	DLG4-related synaptopathy: a new rare brain disorder. *Genetics in Medicine*, **2021**, 23, 888-899	8.1	0
192	Touch and olfaction/taste differentiate children carrying a 16p11.2 deletion from children with ASD. *Molecular Autism*, **2021**, 12, 8	6.5	0
191	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	8.1	2
190	Integrative approach to interpret DYRK1A variants, leading to a frequent neurodevelopmental disorder. *Genetics in Medicine*, **2021**, 23, 2150-2159	8.1	4
189	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	11	1

188	Next-generation sequencing in a series of 80 fetuses with complex cardiac malformations and/or heterotaxy. *Human Mutation*, **2020**, 41, 2167-2178	4.7	8
187	Psychosocial Impact of Predictive Genetic Testing in Hereditary Heart Diseases: The PREDICT Study. *Journal of Clinical Medicine*, **2020**, 9,	5.1	3
186	mutations in the X-linked gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features. *Journal of Medical Genetics*, **2020**, 57, 808-819	5.8	5
185	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. *American Journal of Human Genetics*, **2020**, 106, 893-904	11	14
184	Clinical and Molecular Spectrum of Nonsyndromic Early-Onset Osteoarthritis. *Arthritis and Rheumatology*, **2020**, 72, 1689-1693	9.5	2
183	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	11	6
182	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. *Neuron*, **2020**, 106, 404-420.e8	13.9	49
181	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	5.3	2
180	A dominant vimentin variant causes a rare syndrome with premature aging. *European Journal of Human Genetics*, **2020**, 28, 1218-1230	5.3	12
179	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. *Epilepsia*, **2020**, 61, 387-399	6.4	35
178	De Novo SOX6 Variants Cause a Neurodevelopmental Syndrome Associated with ADHD, Craniosynostosis, and Osteochondromas. *American Journal of Human Genetics*, **2020**, 106, 830-845	11	6
177	iPSC line derived from a Bloom syndrome patient retains an increased disease-specific sister-chromatid exchange activity. *Stem Cell Research*, **2020**, 43, 101696	1.6	2
176	Growth charts in Kabuki syndrome 1. *American Journal of Medical Genetics, Part A*, **2020**, 182, 446-453	2.5	4
175	Gain-of-Function MN1 Truncation Variants Cause a Recognizable Syndrome with Craniofacial and Brain Abnormalities. *American Journal of Human Genetics*, **2020**, 106, 13-25	11	11
174	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	4.7	14
173	Phenotypic spectrum of TGFB3 disease-causing variants in a Dutch-French cohort and first report of a homozygous patient. *Clinical Genetics*, **2020**, 97, 723-730	4	5
172	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. *Genetics in Medicine*, **2020**, 22, 1863-1873	8.1	9
171	Developmental and epilepsy spectrum of KCNB1 encephalopathy with long-term outcome. *Epilepsia*, **2020**, 61, 2461-2473	6.4	4

170	Ribosomopathies: New Therapeutic Perspectives. *Cells*, **2020**, 9,	7.9	7
169	Increasing knowledge in defects: lessons from 35 new patients. *Journal of Medical Genetics*, **2020**, 57, 160-168	5.8	11
168	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. *Genetics in Medicine*, **2020**, 22, 181-188	8.1	12
167	RPL13 Variants Cause Spondyloepimetaphyseal Dysplasia with Severe Short Stature. *American Journal of Human Genetics*, **2019**, 105, 1040-1047	11	9
166	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. *Journal of Human Genetics* , **2019**, 64, 1173-1186	4.3	20
165	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. *Genetics in Medicine*, **2019**, 21, 1797-1807	8.1	17
164	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	8.1	16
163	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. *Human Mutation*, **2019**, 40, 1993-2000	4.7	4
162	Pycnodysostosis: Natural history and management guidelines from 27 French cases and a literature review. *Clinical Genetics*, **2019**, 96, 309-316	4	15
161	Novel mutations in NLGN3 causing autism spectrum disorder and cognitive impairment. *Human Mutation*, **2019**, 40, 2021-2032	4.7	23
160	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	5.6	36
159	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	8.1	14
158	Clinical, Histopathological, and Molecular Diagnostics in Lethal Lung Developmental Disorders. *American Journal of Respiratory and Critical Care Medicine*, **2019**, 200, 1093-1101	10.2	20
157	Searching for secondary findings: considering actionability and preserving the right not to know. *European Journal of Human Genetics*, **2019**, 27, 1481-1484	5.3	9
156	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
155	Exome sequencing identifies a novel missense variant in CTSC causing nonsyndromic aggressive periodontitis. *Journal of Human Genetics*, **2019**, 64, 689-694	4.3	7
154	Population genetic screening: current issues in a European country. *European Journal of Human Genetics*, **2019**, 27, 1321-1323	5.3	1
153	Biallelic MYORG mutation carriers exhibit primary brain calcification with a distinct phenotype. *Brain*, **2019**, 142, 1573-1586	11.2	36

152	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	5.3	5
151	Bilateral retinoblastoma due to a germline mutation of RB1 in a child with down syndrome. *Ophthalmic Genetics*, **2019**, 40, 86	1.2	0
150	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	2.5	5
149	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	5.8	29
148	A step towards precision medicine in management of severe transient polyhydramnios: MAGED2 variant. *Journal of Obstetrics and Gynaecology*, **2019**, 39, 395-397	1.3	4
147	Oro-dental phenotype in patients with RUNX2 duplication. *European Journal of Medical Genetics*, **2019**, 62, 85-89	2.6	6
146	Expanding the phenotype of the X-linked BCOR microphthalmia syndromes. *Human Genetics*, **2019**, 138, 1051-1069	6.3	18
145	Encephalopathies with KCNC1 variants: genotype-phenotype-functional correlations. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1263-1272	5.3	12
144	Genotype/phenotype correlations of childhood-onset congenital sideroblastic anaemia in a European cohort. *British Journal of Haematology*, **2019**, 187, 530-542	4.5	8
143	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	3.1	13
142	Developmental trajectories of neuroanatomical alterations associated with the 16p11.2 Copy Number Variations. *NeuroImage*, **2019**, 203, 116155	7.9	6
141	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	11	17
140	Acanthosis nigricans, hypochondroplasia, and FGFR3 mutations: Findings with five new patients, and a review of the literature. *Pediatric Dermatology*, **2019**, 36, 242-246	1.9	4
139	Estimating the effect size of the 15Q11.2 BP1-BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. *Journal of Medical Genetics*, **2019**, 56, 701-710	5.8	22
138	Rare variants in the genetic background modulate cognitive and developmental phenotypes in individuals carrying disease-associated variants. *Genetics in Medicine*, **2019**, 21, 816-825	8.1	71
137	Clinical and functional characterization of recurrent missense variants implicated in THOC6-related intellectual disability. *Human Molecular Genetics*, **2019**, 28, 952-960	5.6	9
136	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	2.6	4
135	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2019**, 104, 319-330	11	19

134	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	11	58
133	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	5.3	15
132	KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants. *Genetics in Medicine*, **2019**, 21, 850-860	8.1	33
131	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. *Biological Psychiatry*, **2019**, 85, 287-297	7.9	55
130	Duplications at 19q13.33 in patients with neurodevelopmental disorders. *Neurology: Genetics*, **2018**, 4, e210	3.8	1
129	Diagnostic strategy in segmentation defect of the vertebrae: a retrospective study of 73 patients. *Journal of Medical Genetics*, **2018**, 55, 422-429	5.8	8
128	Quantifying the Effects of 16p11.2 Copy Number Variants on Brain Structure: A Multisite Genetic-First Study. *Biological Psychiatry*, **2018**, 84, 253-264	7.9	33
127	Further delineation of the duplication syndrome phenotype in 59 French male patients, with a particular focus on morphological and neurological features. *Journal of Medical Genetics*, **2018**, 55, 359-371	5.8	25
126	A novel mutation in the transmembrane 6 domain of GABBR2 leads to a Rett-like phenotype. *Annals of Neurology*, **2018**, 83, 437-439	9.4	9
125	Rare Coding Variants in ANGPTL6 Are Associated with Familial Forms of Intracranial Aneurysm. *American Journal of Human Genetics*, **2018**, 102, 133-141	11	25
124	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	11	30
123	Familial autosomal dominant severe ankyloglossia with tooth abnormalities. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1614-1617	2.5	4
122	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	11	21
121	Expanding the phenotypic spectrum of variants in PDE4D/PRKAR1A: from acrodysostosis to acroscyphodysplasia. *European Journal of Human Genetics*, **2018**, 26, 1611-1622	5.3	11
120	Novel SUZ12 mutations in Weaver-like syndrome. *Clinical Genetics*, **2018**, 94, 461-466	4	22
119	Natural history of GATA2 deficiency in a survey of 79 French and Belgian patients. *Haematologica*, **2018**, 103, 1278-1287	6.6	74
118	New splicing pathogenic variant in EBP causing extreme familial variability of Conradi-H□ermann-Happle Syndrome. *European Journal of Human Genetics*, **2018**, 26, 1784-1790	5.3	3
117	Microduplication in the 2p16.1p15 chromosomal region linked to developmental delay and intellectual disability. *Molecular Cytogenetics*, **2018**, 11, 39	2	2

116	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	11	24
115	Familial deep endometriosis: A rare monogenic disease?. *European Journal of Obstetrics, Gynecology and Reproductive Biology*, **2018**, 221, 190-193	2.4	2
114	Delineating syndrome: From congenital microcephaly to hyperkinetic encephalopathy. *Neurology: Genetics*, **2018**, 4, e281	3.8	29
113	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. *Annals of Neurology*, **2018**, 84, 788-795	9.4	18
112	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	5.6	32
111	Molecular diagnosis of PIK3CA-related overgrowth spectrum (PROS) in 162 patients and recommendations for genetic testing. *Genetics in Medicine*, **2017**, 19, 989-997	8.1	62
110	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2017**, 100, 352-363	11	49
109	Barber-Say Syndrome and Ablepharon-Macrostomia Syndrome: A Patient's View. *Molecular Syndromology*, **2017**, 8, 172-178	1.5	4
108	Understanding the Pathophysiology of Intracranial Aneurysm: The ICAN Project. *Neurosurgery*, **2017**, 80, 621-626	3.2	14
107	Sex chromosome aneuploidies and copy-number variants: a further explanation for neurodevelopmental prognosis variability?. *European Journal of Human Genetics*, **2017**, 25, 930-934	5.3	13
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	11	36
105	Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. *Human Genetics*, **2017**, 136, 463-479	6.3	41
104	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. *American Journal of Human Genetics*, **2017**, 100, 117-127	11	39
103	Novel KCNB1 mutation associated with non-syndromic intellectual disability. *Journal of Human Genetics*, **2017**, 62, 569-573	4.3	22
102	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. *American Journal of Human Genetics*, **2017**, 101, 768-788	11	81
101	Mutations in GREB1L Cause Bilateral Kidney Agenesis in Humans and Mice. *American Journal of Human Genetics*, **2017**, 101, 803-814	11	41
100	Novel promoters and coding first exons in DLG2 linked to developmental disorders and intellectual disability. *Genome Medicine*, **2017**, 9, 67	14.4	17
99	Two novel variants in CNTNAP1 in two siblings presenting with congenital hypotonia and hypomyelinating neuropathy. *European Journal of Human Genetics*, **2017**, 25, 150-152	5.3	9

98	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. *Npj Genomic Medicine*, **2017**, 2, 32	6.2	32
97	Mutations in signal recognition particle SRP54 cause syndromic neutropenia with Shwachman-Diamond-like features. *Journal of Clinical Investigation*, **2017**, 127, 4090-4103	15.9	89
96	Treacher Collins syndrome: a clinical and molecular study based on a large series of patients. *Genetics in Medicine*, **2016**, 18, 49-56	8.1	78
95	Mandibular dysostosis without microphthalmia caused by OTX2 deletion. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2466-70	2.5	4
94	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. *American Journal of Human Genetics*, **2016**, 99, 720-727	11	28
93	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	11	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	3.1	23
91	Expanding the Phenotype Associated with NAA10-Related N-Terminal Acetylation Deficiency. *Human Mutation*, **2016**, 37, 755-64	4.7	49
90	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment Gene CHAMP1 Cause Syndromic Intellectual Disability. *Human Mutation*, **2016**, 37, 354-8	4.7	29
89	The Number of Genomic Copies at the 16p11.2 Locus Modulates Language, Verbal Memory, and Inhibition. *Biological Psychiatry*, **2016**, 80, 129-139	7.9	57
88	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, 1124-31	5.3	20
87	The expanding spectrum of COL2A1 gene variants IN 136 patients with a skeletal dysplasia phenotype. *European Journal of Human Genetics*, **2016**, 24, 992-1000	5.3	29
86	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry*, **2016**, 73, 20-30	14.5	120
85	Abnormal spindle-like microcephaly-associated (ASPM) mutations strongly disrupt neocortical structure but spare the hippocampus and long-term memory. *Cortex*, **2016**, 74, 158-76	3.8	22
84	Neonatal Marfan Syndrome: Report of a Case with an Inherited Splicing Mutation outside the Neonatal Domain. *Molecular Syndromology*, **2016**, 6, 281-6	1.5	3
83	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. *PLoS ONE*, **2016**, 11, e0150555	3.7	21
82	Mutation Update for Kabuki Syndrome Genes KMT2D and KDM6A and Further Delineation of X-Linked Kabuki Syndrome Subtype 2. *Human Mutation*, **2016**, 37, 847-64	4.7	89
81	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	5.3	15

80	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. *Journal of Human Genetics*, **2016**, 61, 835-8	4.3	24
79	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. *American Journal of Human Genetics*, **2016**, 98, 1001-1010	11	70
78	Mutations in the HECT domain of NEDD4L lead to AKT-mTOR pathway deregulation and cause periventricular nodular heterotopia. *Nature Genetics*, **2016**, 48, 1349-1358	36.3	66
77	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**, 170A, 116-29	2.5	15
76	Oncologic Phenotype of Peripheral Neuroblastic Tumors Associated With PHOX2B Non-Polyalanine Repeat Expansion Mutations. *Pediatric Blood and Cancer*, **2016**, 63, 71-7	3	11
75	Redefining the MED13L syndrome. *European Journal of Human Genetics*, **2015**, 23, 1308-17	5.3	41
74	DYRK1A haploinsufficiency causes a new recognizable syndrome with microcephaly, intellectual disability, speech impairment, and distinct facies. *European Journal of Human Genetics*, **2015**, 23, 1473-81	5.3	63
73	Ten new cases further delineate the syndromic intellectual disability phenotype caused by mutations in DYRK1A. *European Journal of Human Genetics*, **2015**, 23, 1482-7	5.3	41
72	A Potential Contributory Role for Ciliary Dysfunction in the 16p11.2 600 kb BP4-BP5 Pathology. *American Journal of Human Genetics*, **2015**, 96, 784-96	11	35
71	Mutations of the Imprinted CDKN1C Gene as a Cause of the Overgrowth Beckwith-Wiedemann Syndrome: Clinical Spectrum and Functional Characterization. *Human Mutation*, **2015**, 36, 894-902	4.7	47
70	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, 956-69	17.6	118
69	Genetics of giant cell tumors of bone **2015**, 341-345		
68	Genomic aberrations of the CACNA2D1 gene in three patients with epilepsy and intellectual disability. *European Journal of Human Genetics*, **2015**, 23, 628-32	5.3	45
67	New insights into genotype-phenotype correlation for GLI3 mutations. *European Journal of Human Genetics*, **2015**, 23, 92-102	5.3	75
66	Patients with isolated oligo/hypodontia caused by RUNX2 duplication. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1386-90	2.5	7
65	A de novo ADCY5 mutation causes early-onset autosomal dominant chorea and dystonia. *Movement Disorders*, **2015**, 30, 423-7	7	51
64	Epileptic patients with de novo STXBP1 mutations: Key clinical features based on 24 cases. *Epilepsia* , **2015**, 56, 1931-40	6.4	31
63	Muscle magnetic resonance imaging abnormalities in X-linked myopathy with excessive autophagy. *Muscle and Nerve*, **2015**, 52, 673-80	3.4	3

62	Single amino acid charge switch defines clinically distinct proline-serine-threonine phosphatase-interacting protein 1 (PSTPIP1)-associated inflammatory diseases. *Journal of Allergy and Clinical Immunology*, **2015**, 136, 1337-45	11.5	73
61	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	7.5	45
60	Two girls with short stature, short neck, vertebral anomalies, Sprengel deformity and intellectual disability. *European Journal of Medical Genetics*, **2015**, 58, 47-50	2.6	4
59	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**, 167A, 296-312	2.5	321
58	Five children with deletions of 1p34.3 encompassing AGO1 and AGO3. *European Journal of Human Genetics*, **2015**, 23, 761-5	5.3	14
57	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	36.3	376
56	Efficient strategy for the molecular diagnosis of intellectual disability using targeted high-throughput sequencing. *Journal of Medical Genetics*, **2014**, 51, 724-36	5.8	177
55	Mutations in SLC13A5 cause autosomal-recessive epileptic encephalopathy with seizure onset in the first days of life. *American Journal of Human Genetics*, **2014**, 95, 113-20	11	80
54	Blepharophimosis, short humeri, developmental delay and hirschsprung disease: expanding the phenotypic spectrum of MED12 mutations. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1821-5	2.5	17
53	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	4.2	20
52	A new mutation in the C-SH2 domain of PTPN11 causes Noonan syndrome with multiple giant cell lesions. *Journal of Human Genetics*, **2014**, 59, 57-9	4.3	10
51	An emerging phenotype of Xq22 microdeletions in females with severe intellectual disability, hypotonia and behavioral abnormalities. *Journal of Human Genetics*, **2014**, 59, 300-6	4.3	19
50	Similar early characteristics but variable neurological outcome of patients with a de novo mutation of KCNQ2. *Orphanet Journal of Rare Diseases*, **2013**, 8, 80	4.2	64
49	Refining the phenotype associated with MEF2C point mutations. *Neurogenetics*, **2013**, 14, 71-5	3	46
48	Palmo-Plantar hyperkeratosis, intellectual disability, and spastic paraplegia in two maternal half brothers: further evidence for an X-linked inheritance. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1390-3	2.5	1
47	Exudative retinopathy, cerebral calcifications, duodenal atresia, preaxial polydactyly, micropenis, microcephaly and short stature: a new syndrome?. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1829-32	2.5	2
46	Renal phenotypic variability in HDR syndrome: glomerular nephropathy as a novel finding. *European Journal of Pediatrics*, **2013**, 172, 107-10	4.1	16
45	Treatment responses in five patients with Ribbing disease including two with 466C>T missense mutations in TGF1. *Joint Bone Spine*, **2013**, 80, 638-44	2.9	7

44	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, 163-70	2.6	25
43	Wilms' tumor in patients with 9q22.3 microdeletion syndrome suggests a role for PTCH1 in nephroblastomas. *European Journal of Human Genetics*, **2013**, 21, 784-7	5.3	19
42	Finger creases lend a hand in Kabuki syndrome. *European Journal of Medical Genetics*, **2013**, 56, 556-60	2.6	9
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