Bertrand Isidor

List of Publications by Citations

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	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
204	Mirror extreme BMI phenotypes associated with gene dosage at the chromosome 16p11.2 locus. *Nature*, **2011**, 478, 97-102	50.4	322
203	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
202	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
201	NF1 microdeletions in neurofibromatosis type 1: from genotype to phenotype. *Human Mutation*, **2010**, 31, E1506-18	4.7	176
200	Truncating mutations in the last exon of NOTCH2 cause a rare skeletal disorder with osteoporosis. *Nature Genetics*, **2011**, 43, 306-8	36.3	156
199	Defining the Effect of the 16p11.2 Duplication on Cognition, Behavior, and Medical Comorbidities. *JAMA Psychiatry*, **2016**, 73, 20-30	14.5	120
198	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
197	Cant□syndrome is caused by mutations in ABCC9. *American Journal of Human Genetics*, **2012**, 90, 1094-101	11	112
196	Mutational, functional, and expression studies of the TCF4 gene in Pitt-Hopkins syndrome. *Human Mutation*, **2009**, 30, 669-76	4.7	112
195	CYP7B1 mutations in pure and complex forms of hereditary spastic paraplegia type 5. *Brain*, **2009**, 132, 1589-600	11.2	93
194	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
193	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
192	Phenotypic spectrum associated with CASK loss-of-function mutations. *Journal of Medical Genetics*, **2011**, 48, 741-51	5.8	88
191	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. *American Journal of Human Genetics*, **2017**, 101, 768-788	11	81
190	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
189	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

188	New insights into genotype-phenotype correlation for GLI3 mutations. *European Journal of Human Genetics*, **2015**, 23, 92-102	5.3	75
187	Natural history of GATA2 deficiency in a survey of 79 French and Belgian patients. *Haematologica*, **2018**, 103, 1278-1287	6.6	74
186	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
185	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
184	Nephrocalcinosis (enamel renal syndrome) caused by autosomal recessive FAM20A mutations. *Nephron Physiology*, **2012**, 122, 1-6		70
183	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. *American Journal of Human Genetics*, **2016**, 98, 1001-1010	11	70
182	Mutations in STAMBP, encoding a deubiquitinating enzyme, cause microcephaly-capillary malformation syndrome. *Nature Genetics*, **2013**, 45, 556-62	36.3	69
181	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
180	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
179	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**, 140, 1285-96	2.5	64
178	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
177	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
176	Haploinsufficiency of SOX5 at 12p12.1 is associated with developmental delays with prominent language delay, behavior problems, and mild dysmorphic features. *Human Mutation*, **2012**, 33, 728-40	4.7	62
175	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
174	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
173	Thromboxane synthase mutations in an increased bone density disorder (Ghosal syndrome). *Nature Genetics*, **2008**, 40, 284-6	36.3	55
172	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. *Biological Psychiatry*, **2019**, 85, 287-297	7.9	55
171	A de novo ADCY5 mutation causes early-onset autosomal dominant chorea and dystonia. *Movement Disorders*, **2015**, 30, 423-7	7	51

170	GATAD2B loss-of-function mutations cause a recognisable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth in Drosophila. *Journal of Medical Genetics*, **2013**, 50, 507-14	5.8	51
169	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
168	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. *Neuron*, **2020**, 106, 404-420.e8	13.9	49
167	Expanding the Phenotype Associated with NAA10-Related N-Terminal Acetylation Deficiency. *Human Mutation*, **2016**, 37, 755-64	4.7	49
166	Non-USH2A mutations in USH2 patients. *Human Mutation*, **2012**, 33, 504-10	4.7	49
165	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, 1633-40	2.5	49
164	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
163	Refining the phenotype associated with MEF2C point mutations. *Neurogenetics*, **2013**, 14, 71-5	3	46
162	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
161	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
160	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, 175-80	2.5	42
159	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
158	Mutations in GREB1L Cause Bilateral Kidney Agenesis in Humans and Mice. *American Journal of Human Genetics*, **2017**, 101, 803-814	11	41
157	Redefining the MED13L syndrome. *European Journal of Human Genetics*, **2015**, 23, 1308-17	5.3	41
156	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
155	A new microdeletion syndrome of 5q31.3 characterized by severe developmental delays, distinctive facial features, and delayed myelination. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 732-6	2.5	40
154	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
153	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

152	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
151	Biallelic MYORG mutation carriers exhibit primary brain calcification with a distinct phenotype. *Brain*, **2019**, 142, 1573-1586	11.2	36
150	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
149	Biological concepts in human sodium channel epilepsies and their relevance in clinical practice. *Epilepsia*, **2020**, 61, 387-399	6.4	35
148	Mesomelia-synostoses syndrome results from deletion of SULF1 and SLCO5A1 genes at 8q13. *American Journal of Human Genetics*, **2010**, 87, 95-100	11	35
147	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
146	KAT6A Syndrome: genotype-phenotype correlation in 76 patients with pathogenic KAT6A variants. *Genetics in Medicine*, **2019**, 21, 850-860	8.1	33
145	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
144	A framework to identify contributing genes in patients with Phelan-McDermid syndrome. *Npj Genomic Medicine*, **2017**, 2, 32	6.2	32
143	ALK germline mutations in patients with neuroblastoma: a rare and weakly penetrant syndrome. *European Journal of Human Genetics*, **2012**, 20, 291-7	5.3	32
142	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
141	Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. *American Journal of Human Genetics*, **2019**, 104, 1210-1222	11	31
140	Epileptic patients with de novo STXBP1 mutations: Key clinical features based on 24 cases. *Epilepsia* , **2015**, 56, 1931-40	6.4	31
139	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
138	Familial frameshift SRY mutation inherited from a mosaic father with testicular dysgenesis syndrome. *Journal of Clinical Endocrinology and Metabolism*, **2009**, 94, 3467-71	5.6	30
137	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
136	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment Gene CHAMP1 Cause Syndromic Intellectual Disability. *Human Mutation*, **2016**, 37, 354-8	4.7	29
135	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

134	Delineating syndrome: From congenital microcephaly to hyperkinetic encephalopathy. *Neurology: Genetics*, **2018**, 4, e281	3.8	29
133	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
132	Serpentine fibula-polycystic kidney syndrome caused by truncating mutations in NOTCH2. *Human Mutation*, **2011**, 32, 1239-42	4.7	28
131	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
130	Rare Coding Variants in ANGPTL6 Are Associated with Familial Forms of Intracranial Aneurysm. *American Journal of Human Genetics*, **2018**, 102, 133-141	11	25
129	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
128	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
127	Mesomelic dysplasia Kantaputra type is associated with duplications of the HOXD locus on chromosome 2q. *European Journal of Human Genetics*, **2010**, 18, 1310-4	5.3	24
126	De novo MEIS2 mutation causes syndromic developmental delay with persistent gastro-esophageal reflux. *Journal of Human Genetics*, **2016**, 61, 835-8	4.3	24
125	Novel mutations in NLGN3 causing autism spectrum disorder and cognitive impairment. *Human Mutation*, **2019**, 40, 2021-2032	4.7	23
124	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
123	Novel KCNB1 mutation associated with non-syndromic intellectual disability. *Journal of Human Genetics*, **2017**, 62, 569-573	4.3	22
122	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
121	Novel SUZ12 mutations in Weaver-like syndrome. *Clinical Genetics*, **2018**, 94, 461-466	4	22
120	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
119	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
118	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, 1388-96	4.7	21
117	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, 2696-9	2.5	21

116	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. *PLoS ONE*, **2016**, 11, e0150555	3.7	21
115	Genetic abnormalities in a large cohort of Coffin-Siris syndrome patients. *Journal of Human Genetics*, **2019**, 64, 1173-1186	4.3	20
114	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
113	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
112	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
111	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
110	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
109	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. *American Journal of Human Genetics*, **2019**, 104, 319-330	11	19
108	Expanding the phenotype of the X-linked BCOR microphthalmia syndromes. *Human Genetics*, **2019**, 138, 1051-1069	6.3	18
107	A 8.26Mb deletion in 6q16 and a 4.95Mb deletion in 20p12 including JAG1 and BMP2 in a patient with Alagille syndrome and Wolff-Parkinson-White syndrome. *European Journal of Medical Genetics*, **2008**, 51, 651-7	2.6	18
106	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. *Annals of Neurology*, **2018**, 84, 788-795	9.4	18
105	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
104	Novel promoters and coding first exons in DLG2 linked to developmental disorders and intellectual disability. *Genome Medicine*, **2017**, 9, 67	14.4	17
103	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
102	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
101	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
100	Renal phenotypic variability in HDR syndrome: glomerular nephropathy as a novel finding. *European Journal of Pediatrics*, **2013**, 172, 107-10	4.1	16
99	Pycnodysostosis: Natural history and management guidelines from 27 French cases and a literature review. *Clinical Genetics*, **2019**, 96, 309-316	4	15

98	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
97	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
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	5.3	15
95	Understanding the Pathophysiology of Intracranial Aneurysm: The ICAN Project. *Neurosurgery*, **2017**, 80, 621-626	3.2	14
94	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
93	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. *American Journal of Human Genetics*, **2020**, 106, 893-904	11	14
92	Five children with deletions of 1p34.3 encompassing AGO1 and AGO3. *European Journal of Human Genetics*, **2015**, 23, 761-5	5.3	14
91	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**, 155A, 1458-60	2.5	14
90	Complex constitutional subtelomeric 1p36.3 deletion/duplication in a mentally retarded child with neonatal neuroblastoma. *European Journal of Medical Genetics*, **2008**, 51, 679-84	2.6	14
89	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
88	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
87	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
86	A dominant vimentin variant causes a rare syndrome with premature aging. *European Journal of Human Genetics*, **2020**, 28, 1218-1230	5.3	12
85	Encephalopathies with KCNC1 variants: genotype-phenotype-functional correlations. *Annals of Clinical and Translational Neurology*, **2019**, 6, 1263-1272	5.3	12
84	Immunopathological manifestations in Kabuki syndrome: a registry study of 177 individuals. *Genetics in Medicine*, **2020**, 22, 181-188	8.1	12
83	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
82	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
81	Oncologic Phenotype of Peripheral Neuroblastic Tumors Associated With PHOX2B Non-Polyalanine Repeat Expansion Mutations. *Pediatric Blood and Cancer*, **2016**, 63, 71-7	3	11

80	Increasing knowledge in defects: lessons from 35 new patients. *Journal of Medical Genetics*, **2020**, 57, 160-168	5.8	11
79	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
78	Expanding the Spectrum of PMM2-CDG Phenotype. *JIMD Reports*, **2012**, 5, 123-5	1.9	10
77	RPL13 Variants Cause Spondyloepimetaphyseal Dysplasia with Severe Short Stature. *American Journal of Human Genetics*, **2019**, 105, 1040-1047	11	9
76	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
75	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
74	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
73	Finger creases lend a hand in Kabuki syndrome. *European Journal of Medical Genetics*, **2013**, 56, 556-60	2.6	9
72	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
71	Clinical and functional characterization of recurrent missense variants implicated in THOC6-related intellectual disability. *Human Molecular Genetics*, **2019**, 28, 952-960	5.6	9
70	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
69	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
68	Genotype/phenotype correlations of childhood-onset congenital sideroblastic anaemia in a European cohort. *British Journal of Haematology*, **2019**, 187, 530-542	4.5	8
67	Nablus mask-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, 2091-9	2.5	8
66	Exome sequencing identifies a novel missense variant in CTSC causing nonsyndromic aggressive periodontitis. *Journal of Human Genetics*, **2019**, 64, 689-694	4.3	7
65	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
64	Patients with isolated oligo/hypodontia caused by RUNX2 duplication. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1386-90	2.5	7
63	Ribosomopathies: New Therapeutic Perspectives. *Cells*, **2020**, 9,	7.9	7

62	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
61	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
60	Oro-dental phenotype in patients with RUNX2 duplication. *European Journal of Medical Genetics*, **2019**, 62, 85-89	2.6	6
59	Developmental trajectories of neuroanatomical alterations associated with the 16p11.2 Copy Number Variations. *NeuroImage*, **2019**, 203, 116155	7.9	6
58	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
57	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
56	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
55	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
54	Axial spondylometaphyseal dysplasia: Confirmation and further delineation of a new SMD with retinal dystrophy. *American Journal of Medical Genetics, Part A*, **2010**, 152A, 1550-4	2.5	5
53	Inherited 18q23 duplication in a fetus with multiple congenital anomalies. *European Journal of Medical Genetics*, **2008**, 51, 231-8	2.6	5
52	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
51	Neuropsychological study in 19 French patients with White-Sutton syndrome and POGZ mutations. *Clinical Genetics*, **2021**, 99, 407-417	4	5
50	Barber-Say Syndrome and Ablepharon-Macrostomia Syndrome: A Patient's View. *Molecular Syndromology*, **2017**, 8, 172-178	1.5	4
49	Identification of mobile retrocopies during genetic testing: Consequences for routine diagnosis. *Human Mutation*, **2019**, 40, 1993-2000	4.7	4
48	Familial autosomal dominant severe ankyloglossia with tooth abnormalities. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1614-1617	2.5	4
47	Mandibular dysostosis without microphthalmia caused by OTX2 deletion. *American Journal of Medical Genetics, Part A*, **2016**, 170, 2466-70	2.5	4
46	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
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