Philippe M Campeau

List of Publications by Citations

165 papers	5,313 citations	39 h-index	67 g-index
193 ext. papers	6,736 ext. citations	7.4 avg, IF	5.19 L-index

#	Paper	IF	Citations
165	Mesenchymal stromal cells ameliorate experimental autoimmune encephalomyelitis by inhibiting CD4 Th17 T cells in a CC chemokine ligand 2-dependent manner. *Journal of Immunology*, **2009**, 182, 5994-6002	5.3	297
164	WNT1 mutations in early-onset osteoporosis and osteogenesis imperfecta. *New England Journal of Medicine*, **2013**, 368, 1809-16	59.2	253
163	Hereditary breast cancer: new genetic developments, new therapeutic avenues. *Human Genetics*, **2008**, 124, 31-42	6.3	233
162	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. *American Journal of Human Genetics*, **2017**, 101, 664-685	11	214
161	miRNA-34c regulates Notch signaling during bone development. *Human Molecular Genetics*, **2012**, 21, 2991-3000	5.6	182
160	Requirement of argininosuccinate lyase for systemic nitric oxide production. *Nature Medicine*, **2011**, 17, 1619-26	50.5	161
159	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. *Nature Genetics*, **2015**, 47, 661-7	36.3	128
158	A recurrent PDGFRB mutation causes familial infantile myofibromatosis. *American Journal of Human Genetics*, **2013**, 92, 996-1000	11	108
157	A longitudinal study of urea cycle disorders. *Molecular Genetics and Metabolism*, **2014**, 113, 127-30	3.7	106
156	Yunis-Varón syndrome is caused by mutations in FIG4, encoding a phosphoinositide phosphatase. *American Journal of Human Genetics*, **2013**, 92, 781-91	11	101
155	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. *Genetics*, **2017**, 207, 9-27	4	99
154	Mutations in KAT6B, encoding a histone acetyltransferase, cause Genitopatellar syndrome. *American Journal of Human Genetics*, **2012**, 90, 282-9	11	99
153	The genetic basis of DOORS syndrome: an exome-sequencing study. *Lancet Neurology, The*, **2014**, 13, 44-58	24.1	96
152	Characterization of Gaucher disease bone marrow mesenchymal stromal cells reveals an altered inflammatory secretome. *Blood*, **2009**, 114, 3181-90	2.2	76
151	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. *American Journal of Human Genetics*, **2015**, 96, 816-25	11	75
150	Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. *American Journal of Human Genetics*, **2018**, 102, 156-174	11	75
149	TBC1D24 genotype-phenotype correlation: Epilepsies and other neurologic features. *Neurology*, **2016**, 87, 77-85	6.5	75

148	Transfection of large plasmids in primary human myoblasts. *Gene Therapy*, **2001**, 8, 1387-94	4	75
147	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. *Human Molecular Genetics*, **2014**, 23, R1-8	5.6	73
146	De Novo Mutations in CHD4, an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. *American Journal of Human Genetics*, **2016**, 99, 934-941	11	68
145	Genotype-phenotype correlation--promiscuity in the era of next-generation sequencing. *New England Journal of Medicine*, **2014**, 371, 593-6	59.2	68
144	Mutation of KCNJ8 in a patient with Cant syndrome with unique vascular abnormalities - support for the role of K(ATP) channels in this condition. *European Journal of Medical Genetics*, **2013**, 56, 678-82	2.6	65
143	MicroRNA miR-23a cluster promotes osteocyte differentiation by regulating TGF- signalling in osteoblasts. *Nature Communications*, **2017**, 8, 15000	17.4	63
142	Whole-exome sequencing identifies mutations in the nucleoside transporter gene SLC29A3 in dysosteosclerosis, a form of osteopetrosis. *Human Molecular Genetics*, **2012**, 21, 4904-9	5.6	63
141	Long-term outcome in methylmalonic aciduria: a series of 30 French patients. *Molecular Genetics and Metabolism*, **2009**, 97, 172-8	3.7	63
140	Recessive mutations in VPS13D cause childhood onset movement disorders. *Annals of Neurology*, **2018**, 83, 1089-1095	9.4	61
139	The KAT6B-related disorders genitopatellar syndrome and Ohdo/SBBYS syndrome have distinct clinical features reflecting distinct molecular mechanisms. *Human Mutation*, **2012**, 33, 1520-5	4.7	56
138	Nitric-oxide supplementation for treatment of long-term complications in argininosuccinic aciduria. *American Journal of Human Genetics*, **2012**, 90, 836-46	11	56
137	Phenotypic variability of osteogenesis imperfecta type V caused by an IFITM5 mutation. *Journal of Bone and Mineral Research*, **2013**, 28, 1523-30	6.3	55
136	Clinical variability in inherited glycosylphosphatidylinositol deficiency disorders. *Clinical Genetics*, **2019**, 95, 112-121	4	52
135	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. *American Journal of Human Genetics*, **2020**, 106, 356-370	11	51
134	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. *Neuron*, **2020**, 106, 404-420.e8	13.9	49
133	BAFopathiesRDNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffin-Siris and Nicolaides-Baraitser syndromes. *Nature Communications*, **2018**, 9, 4885	17.4	48
132	Identification of novel mutations confirms PDE4D as a major gene causing acrodysostosis. *Human Mutation*, **2013**, 34, 97-102	4.7	46
131	A cross-sectional multicenter study of osteogenesis imperfecta in North America - results from the linked clinical research centers. *Clinical Genetics*, **2015**, 87, 133-40	4	45

130	Early orthotopic liver transplantation in urea cycle defects: follow up of a developmental outcome study. *Molecular Genetics and Metabolism*, **2010**, 100 Suppl 1, S84-7	3.7	45
129	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. *American Journal of Human Genetics*, **2017**, 100, 91-104	11	43
128	Next-generation sequencing for disorders of low and high bone mineral density. *Osteoporosis International*, **2013**, 24, 2253-9	5.3	42
127	Structure-based design and mechanisms of allosteric inhibitors for mitochondrial branched-chain ketoacid dehydrogenase kinase. *Proceedings of the National Academy of Sciences of the United States of America*, **2013**, 110, 9728-33	11.5	40
126	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. *Nature Communications*, **2018**, 9, 4619	17.4	39
125	Genomic study of severe fetal anomalies and discovery of GREB1L mutations in renal agenesis. *Genetics in Medicine*, **2018**, 20, 745-753	8.1	38
124	Mutations in the phosphatidylinositol glycan C () gene are associated with epilepsy and intellectual disability. *Journal of Medical Genetics*, **2017**, 54, 196-201	5.8	36
123	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. *Brain*, **2018**, 141, 2299-2311	11.2	36
122	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. *Genetics in Medicine*, **2019**, 21, 1295-1307	8.1	36
121	Mutations in GPAA1, Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. *American Journal of Human Genetics*, **2017**, 101, 856-865	11	35
120	Clinical heterogeneity in ethylmalonic encephalopathy. *Journal of Child Neurology*, **2009**, 24, 991-6	2.5	34
119	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. *Human Molecular Genetics*, **2017**, 26, 1706-1715	5.6	33
118	DOORS syndrome: phenotype, genotype and comparison with Coffin-Siris syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2014**, 166C, 327-32	3.1	33
117	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. *European Journal of Human Genetics*, **2014**, 22, 1272-7	5.3	32
116	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. *Genetics in Medicine*, **2019**, 21, 837-849	8.1	32
115	Mutations in PIGS, Encoding a GPI Transamidase, Cause a Neurological Syndrome Ranging from Fetal Akinesia to Epileptic Encephalopathy. *American Journal of Human Genetics*, **2018**, 103, 602-611	11	32
114	Mesenchymal stromal cells engineered to express erythropoietin induce anti-erythropoietin antibodies and anemia in allorecipients. *Molecular Therapy*, **2009**, 17, 369-72	11.7	31
113	Prenatal diagnosis of monosomy 1p36: a focus on brain abnormalities and a review of the literature. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 3062-9	2.5	31

112	Biosynthesis of glycosaminoglycans: associated disorders and biochemical tests. *Journal of Inherited Metabolic Disease*, **2016**, 39, 173-88	5.4	30
111	MCTP2 is a dosage-sensitive gene required for cardiac outflow tract development. *Human Molecular Genetics*, **2013**, 22, 4339-48	5.6	30
110	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. *American Journal of Human Genetics*, **2019**, 105, 384-394	11	29
109	Exome sequencing identifies a novel homozygous mutation in the phosphate transporter SLC34A1 in hypophosphatemia and nephrocalcinosis. *Journal of Clinical Endocrinology and Metabolism*, **2014**, 99, E2451-6	5.6	29
108	A 25-year longitudinal analysis of treatment efficacy in inborn errors of metabolism. *Molecular Genetics and Metabolism*, **2008**, 95, 11-6	3.7	28
107	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. *American Journal of Human Genetics*, **2019**, 104, 164-178	11	27
106	Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. *American Journal of Human Genetics*, **2016**, 98, 363-72	11	26
105	The undernourished neonatal mouse metabolome reveals evidence of liver and biliary dysfunction, inflammation, and oxidative stress. *Journal of Nutrition*, **2014**, 144, 273-81	4.1	26
104	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. *American Journal of Human Genetics*, **2019**, 105, 534-548	11	25
103	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. *Science Advances*, **2020**, 6, eaax0021	14.3	24
102	Functional EGFP-dystrophin fusion proteins for gene therapy vector development. *Protein Engineering, Design and Selection*, **2000**, 13, 611-5	1.9	24
101	Canadian Open Genetics Repository (COGR): a unified clinical genomics database as a community resource for standardising and sharing genetic interpretations. *Journal of Medical Genetics*, **2015**, 52, 438-45	5.8	23
100	The epilepsy-associated protein TBC1D24 is required for normal development, survival and vesicle trafficking in mammalian neurons. *Human Molecular Genetics*, **2019**, 28, 584-597	5.6	23
99	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. *American Journal of Human Genetics*, **2019**, 104, 596-610	11	22
98	FHF1 (FGF12) epileptic encephalopathy. *Neurology: Genetics*, **2016**, 2, e115	3.8	22
97	Mutations in ANAPC1, Encoding a Scaffold Subunit of the Anaphase-Promoting Complex, Cause Rothmund-Thomson Syndrome Type 1. *American Journal of Human Genetics*, **2019**, 105, 625-630	11	22
96	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with "Corner Fractures". *American Journal of Human Genetics*, **2017**, 101, 815-823	11	22
95	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

94	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotype-phenotype correlations, and molecular basis. *Genetics in Medicine*, **2020**, 22, 389-397	8.1	22
93	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. *American Journal of Human Genetics*, **2019**, 104, 815-834	11	21
92	Osteogenesis imperfecta without features of type V caused by a mutation in the IFITM5 gene. *Journal of Bone and Mineral Research*, **2013**, 28, 2333-7	6.3	21
91	Epilepsy in KCNH1-related syndromes. *Epileptic Disorders*, **2016**, 18, 123-36	1.9	21
90	A Novel PGM3 Mutation Is Associated With a Severe Phenotype of Bone Marrow Failure, Severe Combined Immunodeficiency, Skeletal Dysplasia, and Congenital Malformations. *Journal of Bone and Mineral Research*, **2017**, 32, 1853-1859	6.3	20
89	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca-Activated K Channel SK3 Cause Zimmermann-Laband Syndrome. *American Journal of Human Genetics*, **2019**, 104, 1139-1157	11	20
88	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). *Genetics in Medicine*, **2018**, 20, 294-302	8.1	20
87	Loss of DDRGK1 modulates SOX9 ubiquitination in spondyloepimetaphyseal dysplasia. *Journal of Clinical Investigation*, **2017**, 127, 1475-1484	15.9	20
86	Case Report: Novel mutations in are associated with autosomal dominant tonic-clonic and myoclonic epilepsy and recessive Parkinsonism, psychosis, and intellectual disability. *F1000Research* , **2017**, 6, 553	3.6	20
85	The spectrum of infantile myofibromatosis includes both non-penetrance and adult recurrence. *European Journal of Medical Genetics*, **2017**, 60, 353-358	2.6	19
84	FBN1 contributing to familial congenital diaphragmatic hernia. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 831-6	2.5	19
83	Dysregulation of cotranscriptional alternative splicing underlies CHARGE syndrome. *Proceedings of the National Academy of Sciences of the United States of America*, **2018**, 115, E620-E629	11.5	19
82	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La Réunion Island, in patients with Fryns syndrome. *European Journal of Human Genetics*, **2018**, 26, 340-349	5.3	18
81	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. *Genetics in Medicine*, **2019**, 21, 2521-2531	8.1	17
80	Argininosuccinate lyase in enterocytes protects from development of necrotizing enterocolitis. *American Journal of Physiology - Renal Physiology*, **2014**, 307, G347-54	5.1	17
79	Heterozygous variants in ACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. *Human Mutation*, **2017**, 38, 1365-1371	4.7	17
78	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. *Journal of Clinical Investigation*, **2020**, 130, 1431-1445	15.9	17
77	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

76	Loss of Oxidation Resistance 1, OXR1, Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. *American Journal of Human Genetics*, **2019**, 105, 1237-1253	11	17
75	Yunis-Varón syndrome caused by biallelic VAC14 mutations. *European Journal of Human Genetics*, **2017**, 25, 1049-1054	5.3	16
74	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. *American Journal of Human Genetics*, **2020**, 106, 143-152	11	16
73	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. *American Journal of Human Genetics*, **2020**, 106, 484-495	11	16
72	Clinical and molecular characterization of a severe form of partial lipodystrophy expanding the phenotype of PPARγ deficiency. *Journal of Lipid Research*, **2012**, 53, 1968-78	6.3	15
71	Diagnosis of ALG12-CDG by exome sequencing in a case of severe skeletal dysplasia. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 213-219	1.8	14
70	Selective inhibition of CCR2 expressing lymphomyeloid cells in experimental autoimmune encephalomyelitis by a GM-CSF-MCP1 fusokine. *Journal of Immunology*, **2009**, 182, 2620-7	5.3	14
69	Arginase overexpression in neurons and its effect on traumatic brain injury. *Molecular Genetics and Metabolism*, **2018**, 125, 112-117	3.7	13
68	Hot water epilepsy and SYN1 variants. *Epilepsia*, **2018**, 59, 2162-2163	6.4	13
67	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a mutation. *Journal of Physical Education and Sports Management*, **2017**, 3, a000984	2.8	12
66	Genomic approaches to diagnose rare bone disorders. *Bone*, **2017**, 102, 5-14	4.7	12
65	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. *Genetics in Medicine*, **2019**, 21, 1058-1064	8.1	12
64	Kaufman oculo-cerebro-facial syndrome in a child with small and absent terminal phalanges and absent nails. *Journal of Human Genetics*, **2017**, 62, 465-471	4.3	11
63	Management of West syndrome in a patient with methylmalonic aciduria. *Journal of Child Neurology*, **2010**, 25, 94-7	2.5	11
62	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. *European Journal of Human Genetics*, **2020**, 28, 1422-1431	5.3	10
61	A PIGH mutation leading to GPI deficiency is associated with developmental delay and autism. *Human Mutation*, **2018**, 39, 827-829	4.7	10
60	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. *American Journal of Human Genetics*, **2019**, 104, 422-438	11	10
59	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. *Genetics in Medicine*, **2020**, 22, 1338-1347	8.1	9

58	Inherited glycophosphatidylinositol deficiency variant database and analysis of pathogenic variants. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00743	2.3	9
57	Bruck syndrome 2 variant lacking congenital contractures and involving a novel compound heterozygous PLOD2 mutation. *Bone*, **2020**, 130, 115047	4.7	9
56	Retrospective Analysis of Congenital Scoliosis: Associated Anomalies and Genetic Diagnoses. *Spine*, **2017**, 42, E841-E847	3.3	8
55	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. *Human Molecular Genetics*, **2018**, 27, 589-600	5.6	8
54	Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases. *Clinical Genetics*, **2017**, 91, 868-880	4	8
53	De Novo KAT5 Variants Cause a Syndrome with Recognizable Facial Dysmorphisms, Cerebellar Atrophy, Sleep Disturbance, and Epilepsy. *American Journal of Human Genetics*, **2020**, 107, 564-574	11	8
52	A non-mosaic mutation in a male with severe congenital anomalies overlapping focal dermal hypoplasia. *Molecular Genetics and Metabolism Reports*, **2017**, 12, 57-61	1.8	7
51	Novel fibronectin mutations and expansion of the phenotype in spondylometaphyseal dysplasia with "corner fractures". *Bone*, **2019**, 121, 163-171	4.7	7
50	Juvenile Paget's Disease From Heterozygous Mutation of SP7 Encoding Osterix (Specificity Protein 7, Transcription Factor SP7). *Bone*, **2020**, 137, 115364	4.7	7
49	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in PIGQ: Report of seven new subjects and review of the literature. *Journal of Inherited Metabolic Disease*, **2020**, 43, 1321-1332	5.4	6
48	A variant of neonatal progeroid syndrome, or Wiedemann-Rautenstrauch syndrome, is associated with a nonsense variant in POLR3GL. *European Journal of Human Genetics*, **2020**, 28, 461-468	5.3	6
47	DOORS syndrome and a recurrent truncating ATP6V1B2 variant. *Genetics in Medicine*, **2021**, 23, 149-154	8.1	6
46	MYOD1 involvement in myopathy. *European Journal of Neurology*, **2018**, 25, e123-e124	6	6
45	Disrupted minor intron splicing is prevalent in Mendelian disorders. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1374	2.3	5
44	Genetic Testing in a Cohort of Complex Esophageal Atresia. *Molecular Syndromology*, **2017**, 8, 236-243	1.5	5
43	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. *American Journal of Medical Genetics, Part A*, **2017**, 173, 733-739	2.5	5
42	Spondyloepimetaphysial Dysplasia with Joint Laxity in Three Siblings with Mutations. *Molecular Syndromology*, **2017**, 8, 303-307	1.5	5
41	Adult presentation of X-linked Conradi-Hünermann-Happle syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1309-14	2.5	5

40	Early childhood presentation of Czech dysplasia. *Clinical Dysmorphology*, **2013**, 22, 76-80	0.9	5
39	A post glycosylphosphatidylinositol (GPI) attachment to proteins, type 2 (PGAP2) variant identified in Mabry syndrome index cases: Molecular genetics of the prototypical inherited GPI disorder. *European Journal of Medical Genetics*, **2020**, 63, 103822	2.6	5
38	Lowry-Wood syndrome: further evidence of association with RNU4ATAC, and correlation between genotype and phenotype. *Human Genetics*, **2018**, 137, 905-909	6.3	5
37	Genetics of the patella. *European Journal of Human Genetics*, **2019**, 27, 671-680	5.3	4
36	Clinical characteristics of patients from Quebec, Canada, with Morquio A syndrome: a longitudinal observational study. *Orphanet Journal of Rare Diseases*, **2020**, 15, 270	4.2	4
35	International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia. *Nature Reviews Endocrinology*, **2021**,	15.2	4
34	Clinicopathological Relationships in an Aged Case of DOORS Syndrome With a p.Arg506X Mutation in the Gene. *Frontiers in Neurology*, **2020**, 11, 767	4.1	4
33	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. *Human Genetics*, **2021**, 140, 1109-1120	6.3	4
32	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. *Clinical Genetics*, **2021**, 99, 313-317	4	4
31	UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. *American Journal of Human Genetics*, **2021**, 108, 134-147	11	4
30	Retrospective analysis of fetal vertebral defects: Associated anomalies, etiologies, and outcome. *American Journal of Medical Genetics, Part A*, **2020**, 182, 664-672	2.5	3
29	Expanding the phenotype of PIGS-associated early onset epileptic developmental encephalopathy. *Epilepsia*, **2021**, 62, e35-e41	6.4	3
28	MYSM1 maintains ribosomal protein gene expression in hematopoietic stem cells to prevent hematopoietic dysfunction. *JCI Insight*, **2020**, 5,	9.9	2
27	Genetic burden linked to founder effects in Saguenay-Lac-Saint-Jean illustrates the importance of genetic screening test availability. *Journal of Medical Genetics*, **2021**, 58, 653-665	5.8	2
26	A de novo frameshift FGFR1 mutation extending the protein in an individual with multiple epiphyseal dysplasia and hypogonadotropic hypogonadism without anosmia. *European Journal of Medical Genetics*, **2020**, 63, 103784	2.6	2
25	Missense substitutions at a conserved 14-3-3 binding site in HDAC4 cause a novel intellectual disability syndrome. *Human Genetics and Genomics Advances*, **2021**, 2, 100015	0.8	2
24	Variable expressivity in a family with an aggrecanopathy. *Molecular Genetics & Genomic Medicine*, **2021**, e1773	2.3	2
23	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders.. *Human Genetics and Genomics Advances*, **2022**, 3, 100075	0.8	1

22	Ethanolamine phosphate on the second mannose as bridge in GPI anchored proteins: Towards understanding inherited PIGG deficiency		1
21	Genetic Burden Contributing to Extremely Low or High Bone Mineral Density in a Senior Male Population From the Osteoporotic Fractures in Men Study (MrOS). *JBMR Plus*, **2020**, 4, e10335	3.9	1
20	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. *Genetics in Medicine*, **2021**, 23, 1873-1881	8.1	1
19	Epileptic encephalopathy caused by ARV1 deficiency: Refinement of the genotype-phenotype spectrum and functional impact on GPI-anchored proteins. *Clinical Genetics*, **2021**, 100, 607-614	4	1
18	Nonsyndromic erythrodermic ichthyosis resulting from a homozygous mutation in PIGL. *Clinical and Experimental Dermatology*, **2020**, 45, 391-394	1.8	1
17	PIGF deficiency causes a phenotype overlapping with DOORS syndrome. *Human Genetics*, **2021**, 140, 879-884	6.3	1
16	Disruption of exon-bridging interactions between the minor and major spliceosomes results in alternative splicing around minor introns. *Nucleic Acids Research*, **2021**, 49, 3524-3545	20.1	1
15	Neurotransmitter diseases and related conditions. *Molecular Genetics and Metabolism*, **2007**, 92, 189-97	3.7	0
14	Establishing a core outcome set for mucopolysaccharidoses (MPS) in children: study protocol for a rapid literature review, candidate outcomes survey, and Delphi surveys. *Trials*, **2021**, 22, 816	2.8	0
13	A homozygous variant in the Lamin B receptor gene LBR results in a non-lethal skeletal dysplasia without Pelger-Hu anomaly. *Bone*, **2020**, 141, 115601	4.7	0
12	Free GPI is the elusive Emm antigen. *Blood*, **2021**, 137, 3588-3589	2.2	0
11	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
10	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. *Genetics in Medicine*, **2021**, 23, 374-383	8.1	0
9	Heterozygous variant in gene in two brothers with early onset osteoporosis. *Bone Reports*, **2021**, 15, 101118	2.6	0
8	Variant-specific effects define the phenotypic spectrum of HNRNPH2-associated neurodevelopmental disorders in males.. *Human Genetics*, **2021**, 141, 257	6.3	0
7	C18orf32 loss-of-function is associated with a neurodevelopmental disorder with hypotonia and contractures.. *Human Genetics*, **2022**, 1	6.3	
6	Expanding the Phenotypic Spectrum of GPI Anchoring Deficiency Due to Biallelic Variants in. *Neurology: Genetics*, **2021**, 7, e631	3.8	
5	Biallelic variants in GLE1 with survival beyond neonatal period. *Clinical Genetics*, **2020**, 98, 622-625	4	

4	Rickets manifestations in a child with metaphyseal anadysplasia, report of a spontaneously resolving case. *BMC Pediatrics*, **2021**, 21, 248	2.6
3	Response to Gao et al. *Genetics in Medicine*, **2021**, 23, 1580-1581	8.1
2	Calvarial doughnut lesions with bone fragility in a French-Canadian family; case report and review of the literature. *Bone Reports*, **2021**, 15, 101121	2.6
1	A Discussion With Dr. Philippe Campeau, Medical Geneticist and Clinician-Scientist.. *Clinical and Investigative Medicine*, **2022**, 45, E5-8	0.9