

Philippe M Campeau

List of Publications by Year in descending order

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174
papers

7,956
citations

53751

45
h-index

69214

77
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193
all docs

193
docs citations

193
times ranked

13785
citing authors

#	ARTICLE	IF	CITATIONS
1	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
2	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.	0.4	326
3	<i>WNT1</i> Mutations in Early-Onset Osteoporosis and Osteogenesis Imperfecta. New England Journal of Medicine, 2013, 368, 1809-1816.	13.9	308
4	Hereditary breast cancer: new genetic developments, new therapeutic avenues. Human Genetics, 2008, 124, 31-42.	1.8	276
5	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. Human Molecular Genetics, 2014, 23, R1-R8.	1.4	234
6	miRNA-34c regulates Notch signaling during bone development. Human Molecular Genetics, 2012, 21, 2991-3000.	1.4	210
7	Requirement of argininosuccinate lyase for systemic nitric oxide production. Nature Medicine, 2011, 17, 1619-1626.	15.2	189
8	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	9.4	177
9	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. American Journal of Human Genetics, 2020, 106, 356-370.	2.6	171
10	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27.	1.2	165
11	A longitudinal study of urea cycle disorders. Molecular Genetics and Metabolism, 2014, 113, 127-130.	0.5	153
12	A Recurrent PDGFRB Mutation Causes Familial Infantile Myofibromatosis. American Journal of Human Genetics, 2013, 92, 996-1000.	2.6	135
13	Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. American Journal of Human Genetics, 2018, 102, 156-174.	2.6	135
14	Yunis-Varãn Syndrome Is Caused by Mutations in FIG4, Encoding a Phosphoinositide Phosphatase. American Journal of Human Genetics, 2013, 92, 781-791.	2.6	124
15	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	3.8	121
16	Mutations in KAT6B, Encoding a Histone Acetyltransferase, Cause Genitopatellar Syndrome. American Journal of Human Genetics, 2012, 90, 282-289.	2.6	112
17	De Novo Mutations in CHD4 , an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dismorphisms. American Journal of Human Genetics, 2016, 99, 934-941.	2.6	111
18	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	4.9	108

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19	Recessive mutations in <i>VPS13D</i> cause childhood onset movement disorders. <i>Annals of Neurology</i> , 2018, 83, 1089-1095.	2.8	104
20	Mutations Impairing GSK3-Mediated MAF Phosphorylation Cause Cataract, Deafness, Intellectual Disability, Seizures, and a Down Syndrome-like Facies. <i>American Journal of Human Genetics</i> , 2015, 96, 816-825.	2.6	102
21	<i>TBC1D24</i> genotype-phenotype correlation. <i>Neurology</i> , 2016, 87, 77-85.	1.5	97
22	MicroRNA miR-23a cluster promotes osteocyte differentiation by regulating TGF- β 2 signalling in osteoblasts. <i>Nature Communications</i> , 2017, 8, 15000.	5.8	91
23	Genotype-Phenotype Correlation Promiscuity in the Era of Next-Generation Sequencing. <i>New England Journal of Medicine</i> , 2014, 371, 593-596.	13.9	86
24	Characterization of Gaucher disease bone marrow mesenchymal stromal cells reveals an altered inflammatory secretome. <i>Blood</i> , 2009, 114, 3181-3190.	0.6	85
25	BAFopathies™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffin-Siris and Nicolaides-Baraitser syndromes. <i>Nature Communications</i> , 2018, 9, 4885.	5.8	83
26	Transfection of large plasmids in primary human myoblasts. <i>Gene Therapy</i> , 2001, 8, 1387-1394.	2.3	81
27	Whole-exome sequencing identifies mutations in the nucleoside transporter gene <i>SLC29A3</i> in dysosteosclerosis, a form of osteopetrosis. <i>Human Molecular Genetics</i> , 2012, 21, 4904-4909.	1.4	81
28	<i>BCL11B</i> mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. <i>Brain</i> , 2018, 141, 2299-2311.	3.7	81
29	The <i>ARID1B</i> spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. <i>Genetics in Medicine</i> , 2019, 21, 1295-1307.	1.1	80
30	Long-term outcome in methylmalonic aciduria: A series of 30 French patients. <i>Molecular Genetics and Metabolism</i> , 2009, 97, 172-178.	0.5	79
31	Mutation of <i>KCNJ8</i> in a patient with Cantu syndrome with unique vascular abnormalities Support for the role of K(ATP) channels in this condition. <i>European Journal of Medical Genetics</i> , 2013, 56, 678-682.	0.7	79
32	Clinical variability in inherited glycosylphosphatidylinositol deficiency disorders. <i>Clinical Genetics</i> , 2019, 95, 112-121.	1.0	76
33	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. <i>American Journal of Human Genetics</i> , 2012, 90, 836-846.	2.6	73
34	Mutations in the Chromatin Regulator Gene <i>BRPF1</i> Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. <i>American Journal of Human Genetics</i> , 2017, 100, 91-104.	2.6	72
35	<i>CHD3</i> helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. <i>Nature Communications</i> , 2018, 9, 4619.	5.8	70
36	The <i>KAT6B</i> -related disorders genitopatellar syndrome and Ohdo/SBBYS syndrome have distinct clinical features reflecting distinct molecular mechanisms. <i>Human Mutation</i> , 2012, 33, 1520-1525.	1.1	68

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37	Phenotypic Variability of Osteogenesis Imperfecta Type V Caused by an <i>IFITM5</i> Mutation. <i>Journal of Bone and Mineral Research</i> , 2013, 28, 1523-1530.	3.1	67
38	Genomic study of severe fetal anomalies and discovery of GREB1L mutations in renal agenesis. <i>Genetics in Medicine</i> , 2018, 20, 745-753.	1.1	60
39	A cross-sectional multicenter study of osteogenesis imperfecta in North America—Results from the linked clinical research centers. <i>Clinical Genetics</i> , 2015, 87, 133-140.	1.0	59
40	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. <i>American Journal of Human Genetics</i> , 2019, 104, 815-834.	2.6	59
41	Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay. <i>American Journal of Human Genetics</i> , 2019, 104, 164-178.	2.6	59
42	Structure-based design and mechanisms of allosteric inhibitors for mitochondrial branched-chain α -ketoacid dehydrogenase kinase. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2013, 110, 9728-9733.	3.3	58
43	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. <i>Science Advances</i> , 2020, 6, eaax0021.	4.7	56
44	Early orthotopic liver transplantation in urea cycle defects: Follow up of a developmental outcome study. <i>Molecular Genetics and Metabolism</i> , 2010, 100, S84-S87.	0.5	53
45	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotype-phenotype correlations, and molecular basis. <i>Genetics in Medicine</i> , 2020, 22, 389-397.	1.1	53
46	Identification of Novel Mutations Confirms <i>Pde4d</i> as a Major Gene Causing Acrodysostosis. <i>Human Mutation</i> , 2013, 34, 97-102.	1.1	49
47	Mutations in GPAA1, Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. <i>American Journal of Human Genetics</i> , 2017, 101, 856-865.	2.6	49
48	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. <i>Genetics in Medicine</i> , 2019, 21, 837-849.	1.1	47
49	Next-generation sequencing for disorders of low and high bone mineral density. <i>Osteoporosis International</i> , 2013, 24, 2253-2259.	1.3	46
50	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. <i>American Journal of Human Genetics</i> , 2019, 105, 534-548.	2.6	46
51	Loss of DDRGK1 modulates SOX9 ubiquitination in spondyloepimetaphyseal dysplasia. <i>Journal of Clinical Investigation</i> , 2017, 127, 1475-1484.	3.9	46
52	Biosynthesis of glycosaminoglycans: associated disorders and biochemical tests. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 173-188.	1.7	45
53	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca ²⁺ -Activated K ⁺ Channel SK3 Cause Zimmermann-Laband Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 1139-1157.	2.6	45
54	Mutations in the phosphatidylinositol glycan C (<i>PIGC</i>) gene are associated with epilepsy and intellectual disability. <i>Journal of Medical Genetics</i> , 2017, 54, 196-201.	1.5	44

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55	Mutations in PIGS, Encoding a GPI Transamidase, Cause a Neurological Syndrome Ranging from Fetal Akinesia to Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2018, 103, 602-611.	2.6	44
56	International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia. <i>Nature Reviews Endocrinology</i> , 2022, 18, 173-189.	4.3	44
57	Estimating the effect size of the 15Q11.2 BP1-BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. <i>Journal of Medical Genetics</i> , 2019, 56, 701-710.	1.5	43
58	Mutations in ANAPC1, Encoding a Scaffold Subunit of the Anaphase-Promoting Complex, Cause Rothmund-Thomson Syndrome Type 1. <i>American Journal of Human Genetics</i> , 2019, 105, 625-630.	2.6	42
59	Novel diagnostic DNA methylation epesignatures expand and refine the epigenetic landscapes of Mendelian disorders. <i>Human Genetics and Genomics Advances</i> , 2022, 3, 100075.	1.0	42
60	Clinical Heterogeneity in Ethylmalonic Encephalopathy. <i>Journal of Child Neurology</i> , 2009, 24, 991-996.	0.7	40
61	MCTP2 is a dosage-sensitive gene required for cardiac outflow tract development. <i>Human Molecular Genetics</i> , 2013, 22, 4339-4348.	1.4	40
62	DOORS syndrome: Phenotype, genotype and comparison with Coffin-Siris syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2014, 166, 327-332.	0.7	40
63	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. <i>Journal of Clinical Investigation</i> , 2020, 130, 1431-1445.	3.9	40
64	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. <i>Human Molecular Genetics</i> , 2017, 26, 1706-1715.	1.4	39
65	Prenatal diagnosis of monosomy 1p36: A focus on brain abnormalities and a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 3062-3069.	0.7	38
66	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. <i>European Journal of Human Genetics</i> , 2014, 22, 1272-1277.	1.4	38
67	The Undernourished Neonatal Mouse Metabolome Reveals Evidence of Liver and Biliary Dysfunction, Inflammation, and Oxidative Stress. <i>Journal of Nutrition</i> , 2014, 144, 273-281.	1.3	38
68	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with "Corner Fractures". <i>American Journal of Human Genetics</i> , 2017, 101, 815-823.	2.6	37
69	Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. <i>American Journal of Human Genetics</i> , 2019, 105, 384-394.	2.6	37
70	Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. <i>American Journal of Human Genetics</i> , 2016, 98, 363-372.	2.6	36
71	The epilepsy-associated protein TBC1D24 is required for normal development, survival and vesicle trafficking in mammalian neurons. <i>Human Molecular Genetics</i> , 2019, 28, 584-597.	1.4	35
72	Mesenchymal Stromal Cells Engineered to Express Erythropoietin Induce Anti-erythropoietin Antibodies and Anemia in Allogeneic Recipients. <i>Molecular Therapy</i> , 2009, 17, 369-372.	3.7	34

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73	Exome Sequencing Identifies a Novel Homozygous Mutation in the Phosphate Transporter SLC34A1 in Hypophosphatemia and Nephrocalcinosis. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2014, 99, E2451-E2456.	1.8	34
74	Epilepsy in <i>KCNH1</i> -related syndromes. <i>Epileptic Disorders</i> , 2016, 18, 123-136.	0.7	34
75	Loss of Oxidation Resistance 1, OXR1, Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. <i>American Journal of Human Genetics</i> , 2019, 105, 1237-1253.	2.6	34
76	A 25-year longitudinal analysis of treatment efficacy in inborn errors of metabolism. <i>Molecular Genetics and Metabolism</i> , 2008, 95, 11-16.	0.5	33
77	FHF1 (FGF12) epileptic encephalopathy. <i>Neurology: Genetics</i> , 2016, 2, e115.	0.9	32
78	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. <i>American Journal of Human Genetics</i> , 2019, 104, 596-610.	2.6	32
79	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , 2019, 104, 530-541.	2.6	30
80	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. <i>American Journal of Human Genetics</i> , 2020, 106, 143-152.	2.6	30
81	A Novel <i>PGM3</i> Mutation Is Associated With a Severe Phenotype of Bone Marrow Failure, Severe Combined Immunodeficiency, Skeletal Dysplasia, and Congenital Malformations. <i>Journal of Bone and Mineral Research</i> , 2017, 32, 1853-1859.	3.1	28
82	Dysregulation of cotranscriptional alternative splicing underlies CHARGE syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, E620-E629.	3.3	28
83	Functional EGFP-dystrophin fusion proteins for gene therapy vector development. <i>Protein Engineering, Design and Selection</i> , 2000, 13, 611-615.	1.0	27
84	Canadian Open Genetics Repository (COGR): a unified clinical genomics database as a community resource for standardising and sharing genetic interpretations. <i>Journal of Medical Genetics</i> , 2015, 52, 438-445.	1.5	27
85	Heterozygous variants in <i>ACTL6A</i> , encoding a component of the BAF complex, are associated with intellectual disability. <i>Human Mutation</i> , 2017, 38, 1365-1371.	1.1	27
86	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La Réunion Island, in patients with Frys syndrome. <i>European Journal of Human Genetics</i> , 2018, 26, 340-349.	1.4	27
87	Data sharing as a national quality improvement program: reporting on BRCA1 and BRCA2 variant-interpretation comparisons through the Canadian Open Genetics Repository (COGR). <i>Genetics in Medicine</i> , 2018, 20, 294-302.	1.1	27
88	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. <i>American Journal of Human Genetics</i> , 2019, 104, 422-438.	2.6	27
89	Osteogenesis imperfecta without features of type V caused by a mutation in the <i>IFITM5</i> gene. <i>Journal of Bone and Mineral Research</i> , 2013, 28, 2333-2337.	3.1	26
90	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. <i>Genetics in Medicine</i> , 2019, 21, 2521-2531.	1.1	25

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91	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. <i>Genetics in Medicine</i> , 2020, 22, 1338-1347.	1.1	25
92	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. <i>European Journal of Human Genetics</i> , 2020, 28, 1422-1431.	1.4	25
93	<i>FBN1</i> contributing to familial congenital diaphragmatic hernia. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 831-836.	0.7	24
94	Case Report: Novel mutations in TBC1D24 are associated with autosomal dominant tonic-clonic and myoclonic epilepsy and recessive Parkinsonism, psychosis, and intellectual disability. <i>F1000Research</i> , 2017, 6, 553.	0.8	24
95	The spectrum of infantile myofibromatosis includes both non-penetrance and adult recurrence. <i>European Journal of Medical Genetics</i> , 2017, 60, 353-358.	0.7	23
96	Hot water epilepsy and <i>SYN1</i> variants. <i>Epilepsia</i> , 2018, 59, 2162-2163.	2.6	23
97	Arginase overexpression in neurons and its effect on traumatic brain injury. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 112-117.	0.5	22
98	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. <i>Genetics in Medicine</i> , 2019, 21, 1058-1064.	1.1	22
99	Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. <i>American Journal of Human Genetics</i> , 2020, 106, 484-495.	2.6	22
100	Selective Inhibition of CCR2 Expressing Lymphomyeloid Cells in Experimental Autoimmune Encephalomyelitis by a GM-CSF-MCP1 Fusokine. <i>Journal of Immunology</i> , 2009, 182, 2620-2627.	0.4	21
101	Argininosuccinate lyase in enterocytes protects from development of necrotizing enterocolitis. <i>American Journal of Physiology - Renal Physiology</i> , 2014, 307, G347-G354.	1.6	21
102	Yunis-VarÃ³n syndrome caused by biallelic VAC14 mutations. <i>European Journal of Human Genetics</i> , 2017, 25, 1049-1054.	1.4	21
103	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. <i>Human Molecular Genetics</i> , 2018, 27, 589-600.	1.4	20
104	Clinical and molecular characterization of a severe form of partial lipodystrophy expanding the phenotype of PPAR δ deficiency. <i>Journal of Lipid Research</i> , 2012, 53, 1968-1978.	2.0	18
105	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a <i>TUBB3</i> mutation. <i>Journal of Physical Education and Sports Management</i> , 2017, 3, a000984.	0.5	18
106	Disruption of exon-bridging interactions between the minor and major spliceosomes results in alternative splicing around minor introns. <i>Nucleic Acids Research</i> , 2021, 49, 3524-3545.	6.5	18
107	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. <i>Human Genetics</i> , 2021, 140, 1109-1120.	1.8	18
108	Diagnosis of ALG12-CDG by exome sequencing in a case of severe skeletal dysplasia. <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 213-219.	0.4	16

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109	A variant of neonatal progeroid syndrome, or Wiedemann-Rautenstrauch syndrome, is associated with a nonsense variant in POLR3GL. <i>European Journal of Human Genetics</i> , 2020, 28, 461-468.	1.4	16
110	Genomic approaches to diagnose rare bone disorders. <i>Bone</i> , 2017, 102, 5-14.	1.4	15
111	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <i>PIGQ</i> : Report of seven new subjects and review of the literature. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1321-1332.	1.7	15
112	UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. <i>American Journal of Human Genetics</i> , 2021, 108, 134-147.	2.6	15
113	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. <i>American Journal of Human Genetics</i> , 2021, 108, 929-941.	2.6	15
114	Kaufman oculo-cerebro-facial syndrome in a child with small and absent terminal phalanges and absent nails. <i>Journal of Human Genetics</i> , 2017, 62, 465-471.	1.1	14
115	Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases. <i>Clinical Genetics</i> , 2017, 91, 868-880.	1.0	14
116	Bruck syndrome 2 variant lacking congenital contractures and involving a novel compound heterozygous PLOD2 mutation. <i>Bone</i> , 2020, 130, 115047.	1.4	14
117	De Novo KAT5 Variants Cause a Syndrome with Recognizable Facial Dysmorphisms, Cerebellar Atrophy, Sleep Disturbance, and Epilepsy. <i>American Journal of Human Genetics</i> , 2020, 107, 564-574.	2.6	14
118	Retrospective Analysis of Congenital Scoliosis. <i>Spine</i> , 2017, 42, E841-E847.	1.0	13
119	A non-mosaic PORCN mutation in a male with severe congenital anomalies overlapping focal dermal hypoplasia. <i>Molecular Genetics and Metabolism Reports</i> , 2017, 12, 57-61.	0.4	13
120	A <i>PIGH</i> mutation leading to GPI deficiency is associated with developmental delay and autism. <i>Human Mutation</i> , 2018, 39, 827-829.	1.1	13
121	Novel fibronectin mutations and expansion of the phenotype in spondylometaphyseal dysplasia with "corner fractures". <i>Bone</i> , 2019, 121, 163-171.	1.4	13
122	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. <i>Genetics in Medicine</i> , 2021, 23, 374-383.	1.1	13
123	MYSM1 maintains ribosomal protein gene expression in hematopoietic stem cells to prevent hematopoietic dysfunction. <i>JCI Insight</i> , 2020, 5, .	2.3	13
124	Management of West Syndrome in a Patient With Methylmalonic Aciduria. <i>Journal of Child Neurology</i> , 2010, 25, 94-97.	0.7	12
125	Juvenile Paget's Disease From Heterozygous Mutation of SP7 Encoding Osterix (Specificity Protein 7), Tj ETQq1 1 0,784314 rgBT /Overl	1.4	12
126	Genetic burden linked to founder effects in Saguenay-Lac-Saint-Jean illustrates the importance of genetic screening test availability. <i>Journal of Medical Genetics</i> , 2021, 58, 653-665.	1.5	12

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127	Lowry-Wood syndrome: further evidence of association with RNU4ATAC, and correlation between genotype and phenotype. <i>Human Genetics</i> , 2018, 137, 905-909.	1.8	11
128	Inherited glycosylphosphatidylinositol deficiency variant database and analysis of pathogenic variants. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e00743.	0.6	11
129	Retrospective analysis of fetal vertebral defects: Associated anomalies, etiologies, and outcome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 664-672.	0.7	11
130	Disrupted minor intron splicing is prevalent in Mendelian disorders. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1374.	0.6	11
131	DOORS syndrome and a recurrent truncating ATP6V1B2 variant. <i>Genetics in Medicine</i> , 2021, 23, 149-154.	1.1	11
132	Expanding the phenotype of <i>PIGS</i> -associated early onset epileptic developmental encephalopathy. <i>Epilepsia</i> , 2021, 62, e35-e41.	2.6	11
133	Early childhood presentation of Czech dysplasia. <i>Clinical Dysmorphology</i> , 2013, 22, 76-80.	0.1	9
134	Clinicopathological Relationships in an Aged Case of DOORS Syndrome With a p.Arg506X Mutation in the ATP6V1B2 Gene. <i>Frontiers in Neurology</i> , 2020, 11, 767.	1.1	9
135	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. <i>Genetics in Medicine</i> , 2022, 24, 1283-1296.	1.1	9
136	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 733-739.	0.7	8
137	<i>MYOD1</i> involvement in myopathy. <i>European Journal of Neurology</i> , 2018, 25, e123-e124.	1.7	8
138	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. <i>European Journal of Medical Genetics</i> , 2020, 63, 103822.	0.7	8
139	Variant-specific effects define the phenotypic spectrum of HNRNP2-associated neurodevelopmental disorders in males. <i>Human Genetics</i> , 2022, 141, 257-272.	1.8	8
140	Genetic Testing in a Cohort of Complex Esophageal Atresia. <i>Molecular Syndromology</i> , 2017, 8, 236-243.	0.3	7
141	Spondyloepimetaphysial Dysplasia with Joint Laxity in Three Siblings with <i>B3GALT6</i> Mutations. <i>Molecular Syndromology</i> , 2017, 8, 303-307.	0.3	7
142	Genetics of the patella. <i>European Journal of Human Genetics</i> , 2019, 27, 671-680.	1.4	7
143	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. <i>Clinical Genetics</i> , 2021, 99, 313-317.	1.0	7
144	Ethanolamine phosphate on the second mannose is a preferential bridge for some GPI-anchored proteins. <i>EMBO Reports</i> , 2022, 23, .	2.0	7

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145	Fibronectin isoforms in skeletal development and associated disorders. <i>American Journal of Physiology - Cell Physiology</i> , 2022, 323, C536-C549.	2.1	7
146	Adult presentation of X-linked Conradi-Hänermann-Happle syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1309-1314.	0.7	6
147	Clinical characteristics of patients from Quebec, Canada, with Morquio A syndrome: a longitudinal observational study. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 270.	1.2	6
148	Missense substitutions at a conserved 14-3-3 binding site in HDAC4 cause a novel intellectual disability syndrome. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100015.	1.0	6
149	Epileptic encephalopathy caused by <i>ARV1</i> deficiency: Refinement of the genotype-phenotype spectrum and functional impact on GPI-anchored proteins. <i>Clinical Genetics</i> , 2021, 100, 607-614.	1.0	6
150	Hypomorphic GINS3 variants alter DNA replication and cause Meier-Gorlin syndrome. <i>JCI Insight</i> , 2022, 7, .	2.3	6
151	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. <i>Genetics in Medicine</i> , 2021, 23, 1873-1881.	1.1	5
152	Neurotransmitter diseases and related conditions. <i>Molecular Genetics and Metabolism</i> , 2007, 92, 189-197.	0.5	3
153	Establishing a core outcome set for mucopolysaccharidoses (MPS) in children: study protocol for a rapid literature review, candidate outcomes survey, and Delphi surveys. <i>Trials</i> , 2021, 22, 816.	0.7	3
154	Variable expressivity in a family with an aggrecanopathy. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1773.	0.6	3
155	Genotype-Phenotype Correlation. <i>Obstetrical and Gynecological Survey</i> , 2014, 69, 728-730.	0.2	2
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157	A de novo frameshift FGFR1 mutation extending the protein in an individual with multiple epiphyseal dysplasia and hypogonadotropic hypogonadism without anosmia. <i>European Journal of Medical Genetics</i> , 2020, 63, 103784.	0.7	2
158	PIGF deficiency causes a phenotype overlapping with DOORS syndrome. <i>Human Genetics</i> , 2021, 140, 879-884.	1.8	2
159	Heterozygous variant in WNT1 gene in two brothers with early onset osteoporosis. <i>Bone Reports</i> , 2021, 15, 101118.	0.2	2
160	Calvarial doughnut lesions with bone fragility in a French-Canadian family; case report and review of the literature. <i>Bone Reports</i> , 2021, 15, 101121.	0.2	2
161	Expanding the Phenotypic Spectrum of GPI Anchoring Deficiency Due to Biallelic Variants in GPAA1. <i>Neurology: Genetics</i> , 2021, 7, e631.	0.9	2
162	Nonsyndromic erythrodermic ichthyosis resulting from a homozygous mutation in PIGL. <i>Clinical and Experimental Dermatology</i> , 2020, 45, 391-394.	0.6	1

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163	Genetic Burden Contributing to Extremely Low or High Bone Mineral Density in a Senior Male Population From the Osteoporotic Fractures in Men Study (MrOS). <i>JBMR Plus</i> , 2020, 4, e10335.	1.3	1
164	A homozygous variant in the Lamin B receptor gene LBR results in a non-lethal skeletal dysplasia without Pelger-HuÅ«t anomaly. <i>Bone</i> , 2020, 141, 115601.	1.4	1
165	Biallelic variants in <scp><i>GLE1</i></scp> with survival beyond neonatal period. <i>Clinical Genetics</i> , 2020, 98, 622-625.	1.0	1
166	Free GPI is the elusive Emm antigen. <i>Blood</i> , 2021, 137, 3588-3589.	0.6	1
167	Rickets manifestations in a child with metaphyseal anadysplasia, report of a spontaneously resolving case. <i>BMC Pediatrics</i> , 2021, 21, 248.	0.7	0
168	Response to Gao et al.. <i>Genetics in Medicine</i> , 2021, 23, 1580-1581.	1.1	0
169	Urea Cycle. , 2014, , 134-151.		0
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174	A Discussion With Dr. Philippe Campeau, Medical Geneticist and Clinician-Scientist. <i>Clinical and Investigative Medicine</i> , 2022, 45, E5-8.	0.3	0