Philippe M Campeau

List of Publications by Year
in descending order

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174	7,956	53794	69250
papers	citations	45 h-index	77 g-index

193	193	193	13785
all docs	docs citations	times ranked	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.	6.2	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.	0.8	326
3	WNT1 Mutations in Early-Onset Osteoporosis and Osteogenesis Imperfecta. New England Journal of Medicine, 2013, 368, 1809-1816.	27.0	308
4	Hereditary breast cancer: new genetic developments, new therapeutic avenues. Human Genetics, 2008, 124, 31-42.	3.8	276
5	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. Human Molecular Genetics, 2014, 23, R1-R8.	2.9	234
6	miRNA-34c regulates Notch signaling during bone development. Human Molecular Genetics, 2012, 21, 2991-3000.	2.9	210
7	Requirement of argininosuccinate lyase for systemic nitric oxide production. Nature Medicine, 2011, 17, 1619-1626.	30.7	189
8	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	21.4	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.	6.2	171
10	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27.	2.9	165
11	A longitudinal study of urea cycle disorders. Molecular Genetics and Metabolism, 2014, 113, 127-130.	1.1	153
12	A Recurrent PDGFRB Mutation Causes Familial Infantile Myofibromatosis. American Journal of Human Genetics, 2013, 92, 996-1000.	6.2	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.	6.2	135
14	Yunis-VarÃ³n Syndrome Is Caused by Mutations in FIG4, Encoding a Phosphoinositide Phosphatase. American Journal of Human Genetics, 2013, 92, 781-791.	6.2	124
15	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	8.1	121
16	Mutations in KAT6B, Encoding a Histone Acetyltransferase, Cause Genitopatellar Syndrome. American Journal of Human Genetics, 2012, 90, 282-289.	6.2	112
17	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.	6.2	111
18	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	10.2	108

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19	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	5.3	104
20	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, 816-825.	6.2	102
21	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.1	97
22	MicroRNA miR-23a cluster promotes osteocyte differentiation by regulating TGF-Î² signalling in osteoblasts. Nature Communications, 2017, 8, 15000.	12.8	91
23	Genotypeâ€“Phenotype Correlation â€” Promiscuity in the Era of Next-Generation Sequencing. New England Journal of Medicine, 2014, 371, 593-596.	27.0	86
24	Characterization of Gaucher disease bone marrow mesenchymal stromal cells reveals an altered inflammatory secretome. Blood, 2009, 114, 3181-3190.	1.4	85
25	BAFopathiesâ€™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Nature Communications, 2018, 9, 4885.	12.8	83
26	Transfection of large plasmids in primary human myoblasts. Gene Therapy, 2001, 8, 1387-1394.	4.5	81
27	Whole-exome sequencing identifies mutations in the nucleoside transporter gene SLC29A3 in dysosteosclerosis, a form of osteopetrosis. Human Molecular Genetics, 2012, 21, 4904-4909.	2.9	81
28	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	7.6	81
29	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
30	Long-term outcome in methylmalonic aciduria: A series of 30 French patients. Molecular Genetics and Metabolism, 2009, 97, 172-178.	1.1	79
31	Mutation of KCNJ8 in a patient with CantÃº syndrome with unique vascular abnormalities â€“ Support for the role of K(ATP) channels in this condition. European Journal of Medical Genetics, 2013, 56, 678-682.	1.3	79
32	Clinical variability in inherited glycosylphosphatidylinositol deficiency disorders. Clinical Genetics, 2019, 95, 112-121.	2.0	76
33	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	6.2	73
34	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. American Journal of Human Genetics, 2017, 100, 91-104.	6.2	72
35	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	12.8	70
36	TheKAT6B-related disorders genitopatellar syndrome and Ohdo/SBBYS syndrome have distinct clinical features reflecting distinct molecular mechanisms. Human Mutation, 2012, 33, 1520-1525.	2.5	68

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37	Phenotypic Variability of Osteogenesis Imperfecta Type V Caused by an IFITM 5 Mutation. Journal of Bone and Mineral Research, 2013, 28, 1523-1530.	2.8	67
38	Genomic study of severe fetal anomalies and discovery of GREB1L mutations in renal agenesis. Genetics in Medicine, 2018, 20, 745-753.	2.4	60
39	A crossâ€sectional multicenter study ofÂosteogenesis imperfecta in North AmericaÂâ€“Âresults from the linked clinical research centers. Clinical Genetics, 2015, 87, 133-140.	2.0	59
40	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.	6.2	59
41	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.	6.2	59
42	Structure-based design and mechanisms of allosteric inhibitors for mitochondrial branched-chain Î±-ketoacid dehydrogenase kinase. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 9728-9733.	7.1	58
43	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. Science Advances, 2020, 6, eaax0021.	10.3	56
44	Early orthotopic liver transplantation in urea cycle defects: Follow up of a developmental outcome study. Molecular Genetics and Metabolism, 2010, 100, S84-S87.	1.1	53
45	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	2.4	53
46	Identification of Novel Mutations ConfirmsPde4das a Major Gene Causing Acrodysostosis. Human Mutation, 2013, 34, 97-102.	2.5	49
47	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.	6.2	49
48	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	2.4	47
49	Next-generation sequencing for disorders of low and high bone mineral density. Osteoporosis International, 2013, 24, 2253-2259.	3.1	46
50	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	6.2	46
51	Loss of DDRGK1 modulates SOX9 ubiquitination in spondyloepimetaphyseal dysplasia. Journal of Clinical Investigation, 2017, 127, 1475-1484.	8.2	46
52	Biosynthesis of glycosaminoglycans: associated disorders and biochemical tests. Journal of Inherited Metabolic Disease, 2016, 39, 173-188.	3.6	45
53	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca2+-Activated K+ Channel SK3 Cause Zimmermann-Laband Syndrome. American Journal of Human Genetics, 2019, 104, 1139-1157.	6.2	45
54	Mutations in the phosphatidylinositol glycan C (PIGC) gene are associated with epilepsy and intellectual disability. Journal of Medical Genetics, 2017, 54, 196-201.	3.2	44

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55	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.	6.2	44
56	International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia. Nature Reviews Endocrinology, 2022, 18, 173-189.	9.6	44
57	Estimating the effect size of the 15Q11.2 BP1â€“BP2 deletion and its contribution to neurodevelopmental symptoms: recommendations for practice. Journal of Medical Genetics, 2019, 56, 701-710.	3.2	43
58	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.	6.2	42
59	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.7	42
60	Clinical Heterogeneity in Ethylmalonic Encephalopathy. Journal of Child Neurology, 2009, 24, 991-996.	1.4	40
61	MCTP2 is a dosage-sensitive gene required for cardiac outflow tract development. Human Molecular Genetics, 2013, 22, 4339-4348.	2.9	40
62	DOORS syndrome: Phenotype, genotype and comparison with Coffinâ€Siris syndrome. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2014, 166, 327-332.	1.6	40
63	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. Journal of Clinical Investigation, 2020, 130, 1431-1445.	8.2	40
64	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	2.9	39
65	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, 3062-3069.	1.2	38
66	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. European Journal of Human Genetics, 2014, 22, 1272-1277.	2.8	38
67	The Undernourished Neonatal Mouse Metabolome Reveals Evidence of Liver and Biliary Dysfunction, Inflammation, and Oxidative Stress. Journal of Nutrition, 2014, 144, 273-281.	2.9	38
68	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€• American Journal of Human Genetics, 2017, 101, 815-823.	6.2	37
69	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.	6.2	37
70	Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. American Journal of Human Genetics, 2016, 98, 363-372.	6.2	36
71	The epilepsy-associated protein TBC1D24 is required for normal development, survival and vesicle trafficking in mammalian neurons. Human Molecular Genetics, 2019, 28, 584-597.	2.9	35
72	Mesenchymal Stromal Cells Engineered to Express Erythropoietin Induce Anti-erythropoietin Antibodies and Anemia in Allorecipients. Molecular Therapy, 2009, 17, 369-372.	8.2	34

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73	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-E2456.	3.6	34
74	Epilepsy in KCNH1â€related syndromes. Epileptic Disorders, 2016, 18, 123-136.	1.3	34
75	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.	6.2	34
76	A 25-year longitudinal analysis of treatment efficacy in inborn errors of metabolism. Molecular Genetics and Metabolism, 2008, 95, 11-16.	1.1	33
77	FHF1 (FGF12) epileptic encephalopathy. Neurology: Genetics, 2016, 2, e115.	1.9	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. American Journal of Human Genetics, 2019, 104, 596-610.	6.2	32
79	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	6.2	30
80	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. American Journal of Human Genetics, 2020, 106, 143-152.	6.2	30
81	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.	2.8	28
82	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.	7.1	28
83	Functional EGFPâ€“dystrophin fusion proteins for gene therapy vector development. Protein Engineering, Design and Selection, 2000, 13, 611-615.	2.1	27
84	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, 438-445.	3.2	27
85	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	2.5	27
86	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La RÃ©union Island, in patients with Fryns syndrome. European Journal of Human Genetics, 2018, 26, 340-349.	2.8	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). Genetics in Medicine, 2018, 20, 294-302.	2.4	27
88	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
89	Osteogenesis imperfecta without features of type V caused by a mutation in the IFITM 5 gene. Journal of Bone and Mineral Research, 2013, 28, 2333-2337.	2.8	26
90	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	2.4	25

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91	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	2.4	25
92	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.	2.8	25
93	FBN1 contributing to familial congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2015, 167, 831-836.	1.2	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. F1000Research, 2017, 6, 553.	1.6	24
95	The spectrum of infantile myofibromatosis includes both non-penetrance and adult recurrence. European Journal of Medical Genetics, 2017, 60, 353-358.	1.3	23
96	Hot water epilepsy and <scp>SYN</scp>1 variants. Epilepsia, 2018, 59, 2162-2163.	5.1	23
97	Arginase overexpression in neurons and its effect on traumatic brain injury. Molecular Genetics and Metabolism, 2018, 125, 112-117.	1.1	22
98	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. Genetics in Medicine, 2019, 21, 1058-1064.	2.4	22
99	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.	6.2	22
100	Selective Inhibition of CCR2 Expressing Lymphomyeloid Cells in Experimental Autoimmune Encephalomyelitis by a GM-CSF-MCP1 Fusokine. Journal of Immunology, 2009, 182, 2620-2627.	0.8	21
101	Argininosuccinate lyase in enterocytes protects from development of necrotizing enterocolitis. American Journal of Physiology - Renal Physiology, 2014, 307, G347-G354.	3.4	21
102	Yunis-VarÃ³n syndrome caused by biallelic VAC14 mutations. European Journal of Human Genetics, 2017, 25, 1049-1054.	2.8	21
103	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. Human Molecular Genetics, 2018, 27, 589-600.	2.9	20
104	Clinical and molecular characterization of a severe form of partial lipodystrophy expanding the phenotype of PPARÎ³ deficiency. Journal of Lipid Research, 2012, 53, 1968-1978.	4.2	18
105	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a TUBB3 mutation. Journal of Physical Education and Sports Management, 2017, 3, a000984.	1.2	18
106	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.	14.5	18
107	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	3.8	18
108	Diagnosis of ALG12-CDG by exome sequencing in a case of severe skeletal dysplasia. Molecular Genetics and Metabolism Reports, 2014, 1, 213-219.	1.1	16

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109	A variant of neonatal progeroid syndrome, or Wiedemannâ€“Rautenstrauch syndrome, is associated with a nonsense variant in POLR3GL. European Journal of Human Genetics, 2020, 28, 461-468.	2.8	16
110	Genomic approaches to diagnose rare bone disorders. Bone, 2017, 102, 5-14.	2.9	15
111	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332.	3.6	15
112	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.	6.2	15
113	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	6.2	15
114	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.	2.3	14
115	Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases. Clinical Genetics, 2017, 91, 868-880.	2.0	14
116	Bruck syndrome 2 variant lacking congenital contractures and involving a novel compound heterozygous PLOD2 mutation. Bone, 2020, 130, 115047.	2.9	14
117	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.	6.2	14
118	Retrospective Analysis of Congenital Scoliosis. Spine, 2017, 42, E841-E847.	2.0	13
119	A non-mosaic PORCN mutation in a male with severe congenital anomalies overlapping focal dermal hypoplasia. Molecular Genetics and Metabolism Reports, 2017, 12, 57-61.	1.1	13
120	APIGHmutation leading to GPI deficiency is associated with developmental delay and autism. Human Mutation, 2018, 39, 827-829.	2.5	13
121	Novel fibronectin mutations and expansion of the phenotype in spondylometaphyseal dysplasia with â€œcorner fracturesâ€. Bone, 2019, 121, 163-171.	2.9	13
122	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	2.4	13
123	MYSM1 maintains ribosomal protein gene expression in hematopoietic stem cells to prevent hematopoietic dysfunction. JCI Insight, 2020, 5, .	5.0	13
124	Management of West Syndrome in a Patient With Methylmalonic Aciduria. Journal of Child Neurology, 2010, 25, 94-97.	1.4	12
125	Juvenile Paget's Disease From Heterozygous Mutation of SP7 Encoding Osterix (Specificity Protein 7,) Tj ETQq1 1 0.784314 rgBT /Ove	2.9	12
126	Genetic burden linked to founder effects in Saguenayâ€“Lac-Saint-Jean illustrates the importance of genetic screening test availability. Journal of Medical Genetics, 2021, 58, 653-665.	3.2	12

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127	Lowry-Wood syndrome: further evidence of association with RNU4ATAC, and correlation between genotype and phenotype. Human Genetics, 2018, 137, 905-909.	3.8	11
128	Inherited glycophosphatidylinositol deficiency variant database and analysis of pathogenic variants. Molecular Genetics & Genomic Medicine, 2019, 7, e00743.	1.2	11
129	Retrospective analysis of fetal vertebral defects: Associated anomalies, etiologies, and outcome. American Journal of Medical Genetics, Part A, 2020, 182, 664-672.	1.2	11
130	Disrupted minor intron splicing is prevalent in Mendelian disorders. Molecular Genetics & Genomic Medicine, 2020, 8, e1374.	1.2	11
131	DOORS syndrome and a recurrentÂtruncating ATP6V1B2 variant. Genetics in Medicine, 2021, 23, 149-154.	2.4	11
132	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	5.1	11
133	Early childhood presentation of Czech dysplasia. Clinical Dysmorphology, 2013, 22, 76-80.	0.3	9
134	Clinicopathological Relationships in an Aged Case of DOORS Syndrome With a p.Arg506X Mutation in the ATP6V1B2 Gene. Frontiers in Neurology, 2020, 11, 767.	2.4	9
135	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	2.4	9
136	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. American Journal of Medical Genetics, Part A, 2017, 173, 733-739.	1.2	8
137	MYOD1 involvement in myopathy. European Journal of Neurology, 2018, 25, e123-e124.	3.3	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. European Journal of Medical Genetics, 2020, 63, 103822.	1.3	8
139	Variant-specific effects define the phenotypic spectrum of HNRNPH2-associated neurodevelopmental disorders in males. Human Genetics, 2022, 141, 257-272.	3.8	8
140	Genetic Testing in a Cohort of Complex Esophageal Atresia. Molecular Syndromology, 2017, 8, 236-243.	0.8	7
141	Spondyloepimetaphysial Dysplasia with Joint Laxity in Three Siblings with B3GALT6 Mutations. Molecular Syndromology, 2017, 8, 303-307.	0.8	7
142	Genetics of the patella. European Journal of Human Genetics, 2019, 27, 671-680.	2.8	7
143	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317.	2.0	7
144	Ethanolamineâ€phosphate on the second mannose is a preferential bridge for some GPIâ€anchored proteins. EMBO Reports, 2022, 23, .	4.5	7

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145	Fibronectin isoforms in skeletal development and associated disorders. American Journal of Physiology - Cell Physiology, 2022, 323, C536-C549.	4.6	7
146	Adult presentation of Xâ€linked Conradiâ€HÃ¼nermannâ€Happle syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1309-1314.	1.2	6
147	Clinical characteristics of patients from Quebec, Canada, with Morquio A syndrome: a longitudinal observational study. Orphanet Journal of Rare Diseases, 2020, 15, 270.	2.7	6
148	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.	1.7	6
149	Epileptic encephalopathy caused by <scp>ARV1</scp> deficiency: Refinement of the genotypeâ€“phenotype spectrum and functional impact on <scp>GPI</scp>â€anchored proteins. Clinical Genetics, 2021, 100, 607-614.	2.0	6
150	Hypomorphic GINS3 variants alter DNA replication and cause Meier-Gorlin syndrome. JCI Insight, 2022, 7, .	5.0	6
151	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. Genetics in Medicine, 2021, 23, 1873-1881.	2.4	5
152	Neurotransmitter diseases and related conditions. Molecular Genetics and Metabolism, 2007, 92, 189-197.	1.1	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. Trials, 2021, 22, 816.	1.6	3
154	Variable expressivity in a family with an aggrecanopathy. Molecular Genetics & Genomic Medicine, 2022, 10, e1773.	1.2	3
155	Genotype-Phenotype Correlation. Obstetrical and Gynecological Survey, 2014, 69, 728-730.	0.4	2
156	Genomic Study of Severe Fetal Anomalies and Discovery of GREB1L Mutations in Renal Agenesis. Obstetrical and Gynecological Survey, 2018, 73, 677-679.	0.4	2
157	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.	1.3	2
158	PIGF deficiency causes a phenotype overlapping with DOORS syndrome. Human Genetics, 2021, 140, 879-884.	3.8	2
159	Heterozygous variant in WNT1 gene in two brothers with early onset osteoporosis. Bone Reports, 2021, 15, 101118.	0.4	2
160	Calvarial doughnut lesions with bone fragility in a French-Canadian family; case report and review of the literature. Bone Reports, 2021, 15, 101121.	0.4	2
161	Expanding the Phenotypic Spectrum of GPI Anchoring Deficiency Due to Biallelic Variants in GPAA1. Neurology: Genetics, 2021, 7, e631.	1.9	2
162	Nonsyndromic erythrodermic ichthyosis resulting from a homozygous mutation in PIGL. Clinical and Experimental Dermatology, 2020, 45, 391-394.	1.3	1

#	Article	IF	Citations
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