Philippe M Campeau

List of Publications by Year
in descending order

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174 papers	7,956 citations	61687 45 h-index	78623 77 g-index
193 all docs	193 docs citations	193 times ranked	14846 citing authors

#	Article	IF	Citations
1	International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia. Nature Reviews Endocrinology, 2022, 18, 173-189.	4.3	44
2	Novel diagnostic DNA methylation episignatures expand and refine the epigenetic landscapes of Mendelian disorders. Human Genetics and Genomics Advances, 2022, 3, 100075.	1.0	42
3	C18orf32 loss-of-function is associated with a neurodevelopmental disorder with hypotonia and contractures. Human Genetics, 2022, , 1.	1.8	0
4	Inherited variants in CHD3 show variable expressivity in Snijders Blok-Campeau syndrome. Genetics in Medicine, 2022, 24, 1283-1296.	1.1	9
5	Variant-specific effects define the phenotypic spectrum of HNRNPH2-associated neurodevelopmental disorders in males. Human Genetics, 2022, 141, 257-272.	1.8	8
6	Variable expressivity in a family with an aggrecanopathy. Molecular Genetics & Genomic Medicine, 2022, 10, e1773.	0.6	3
7	A Discussion With Dr. Philippe Campeau, Medical Geneticist and Clinician-Scientist. Clinical and Investigative Medicine, 2022, 45, E5-8.	0.3	0
8	Ethanolamineâ€phosphate on the second mannose is a preferential bridge for some GPIâ€anchored proteins. EMBO Reports, 2022, 23, .	2.0	7
9	Hypomorphic GINS3 variants alter DNA replication and cause Meier-Gorlin syndrome. JCI Insight, 2022, 7, .	2.3	6
10	Fibronectin isoforms in skeletal development and associated disorders. American Journal of Physiology - Cell Physiology, 2022, 323, C536-C549.	2.1	7
11	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. Clinical Genetics, 2021, 99, 313-317.	1.0	7
12	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	1.1	13
13	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.	1.0	6
14	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.	2.6	15
15	DOORS syndrome and a recurrentÂtruncating ATP6V1B2 variant. Genetics in Medicine, 2021, 23, 149-154.	1.1	11
16	Expanding the phenotype of PIGSâ€associated early onset epileptic developmental encephalopathy. Epilepsia, 2021, 62, e35-e41.	2.6	11
17	PIGF deficiency causes a phenotype overlapping with DOORS syndrome. Human Genetics, 2021, 140, 879-884.	1.8	2
18	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.	6.5	18

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19	Free GPI is the elusive Emm antigen. Blood, 2021, 137, 3588-3589.	0.6	1
20	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.	1.5	12
21	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.	2.6	15
22	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. Human Genetics, 2021, 140, 1109-1120.	1.8	18
23	Rickets manifestations in a child with metaphyseal anadysplasia, report of a spontaneously resolving case. BMC Pediatrics, 2021, 21, 248.	0.7	0
24	Response to Gao et al.. Genetics in Medicine, 2021, 23, 1580-1581.	1.1	0
25	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. Genetics in Medicine, 2021, 23, 1873-1881.	1.1	5
26	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.	1.0	6
27	Heterozygous variant in WNT1 gene in two brothers with early onset osteoporosis. Bone Reports, 2021, 15, 101118.	0.2	2
28	Calvarial doughnut lesions with bone fragility in a French-Canadian family; case report and review of the literature. Bone Reports, 2021, 15, 101121.	0.2	2
29	Expanding the Phenotypic Spectrum of GPI Anchoring Deficiency Due to Biallelic Variants in GPAA1. Neurology: Genetics, 2021, 7, e631.	0.9	2
30	An all-encompassing variant classification system proposed. European Journal of Human Genetics, 2021, , .	1.4	0
31	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.	0.7	3
32	Nonsyndromic erythrodermic ichthyosis resulting from a homozygous mutation in PIGL. Clinical and Experimental Dermatology, 2020, 45, 391-394.	0.6	1
33	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.	0.7	2
34	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotypeâ€“phenotype correlations, and molecular basis. Genetics in Medicine, 2020, 22, 389-397.	1.1	53
35	Bruck syndrome 2 variant lacking congenital contractures and involving a novel compound heterozygous PLOD2 mutation. Bone, 2020, 130, 115047.	1.4	14
36	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.	1.4	16

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37	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.	1.3	1
38	Retrospective analysis of fetal vertebral defects: Associated anomalies, etiologies, and outcome. American Journal of Medical Genetics, Part A, 2020, 182, 664-672.	0.7	11
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.	0.7	8
40	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.	2.6	14
41	Clinicopathological Relationships in an Aged Case of DOORS Syndrome With a p.Arg506X Mutation in the ATP6V1B2 Gene. Frontiers in Neurology, 2020, 11, 767.	1.1	9
42	A homozygous variant in the Lamin B receptor gene LBR results in a non-lethal skeletal dysplasia without Pelger-HuÃ«t anomaly. Bone, 2020, 141, 115601.	1.4	1
43	Biallelic variants in <scp>GLE1</scp> with survival beyond neonatal period. Clinical Genetics, 2020, 98, 622-625.	1.0	1
44	Clinical characteristics of patients from Quebec, Canada, with Morquio A syndrome: a longitudinal observational study. Orphanet Journal of Rare Diseases, 2020, 15, 270.	1.2	6
45	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. Genetics in Medicine, 2020, 22, 1338-1347.	1.1	25
46	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.	1.4	25
47	Disrupted minor intron splicing is prevalent in Mendelian disorders. Molecular Genetics & Genomic Medicine, 2020, 8, e1374.	0.6	11
48	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	3.8	121
49	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.	1.7	15
50	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. American Journal of Human Genetics, 2020, 106, 143-152.	2.6	30
51	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
52	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. Science Advances, 2020, 6, eaax0021.	4.7	56
53	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.	2.6	22
54	Juvenile Paget's Disease From Heterozygous Mutation of SP7 Encoding Osterix (Specificity Protein 7,) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	1.4	12

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55	MYSM1 maintains ribosomal protein gene expression in hematopoietic stem cells to prevent hematopoietic dysfunction. JCI Insight, 2020, 5, .	2.3	13
56	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. Journal of Clinical Investigation, 2020, 130, 1431-1445.	3.9	40
57	Clinical variability in inherited glycosylphosphatidylinositol deficiency disorders. Clinical Genetics, 2019, 95, 112-121.	1.0	76
58	Inherited glycophosphatidylinositol deficiency variant database and analysis of pathogenic variants. Molecular Genetics & Genomic Medicine, 2019, 7, e00743.	0.6	11
59	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	2.6	46
60	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.	2.6	42
61	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.	2.6	37
62	Genetics of the patella. European Journal of Human Genetics, 2019, 27, 671-680.	1.4	7
63	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.	2.6	45
64	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. Genetics in Medicine, 2019, 21, 2521-2531.	1.1	25
65	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.	2.6	59
66	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.	2.6	32
67	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.	2.6	30
68	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	2.6	27
69	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.	1.5	43
70	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.	2.6	34
71	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. Genetics in Medicine, 2019, 21, 837-849.	1.1	47
72	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.	2.6	59

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73	Novel fibronectin mutations and expansion of the phenotype in spondylometaphyseal dysplasia with â€œcorner fracturesâ€. Bone, 2019, 121, 163-171.	1.4	13
74	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	1.1	80
75	The epilepsy-associated protein TBC1D24 is required for normal development, survival and vesicle trafficking in mammalian neurons. Human Molecular Genetics, 2019, 28, 584-597.	1.4	35
76	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. Genetics in Medicine, 2019, 21, 1058-1064.	1.1	22
77	Recessive mutations in VPS13D cause childhood onset movement disorders. Annals of Neurology, 2018, 83, 1089-1095.	2.8	104
78	APIGHmutation leading to GPI deficiency is associated with developmental delay and autism. Human Mutation, 2018, 39, 827-829.	1.1	13
79	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.	1.4	27
80	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.	3.3	28
81	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
82	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. Human Molecular Genetics, 2018, 27, 589-600.	1.4	20
83	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.	1.1	27
84	Genomic study of severe fetal anomalies and discovery of GREB1L mutations in renal agenesis. Genetics in Medicine, 2018, 20, 745-753.	1.1	60
85	Genomic Study of Severe Fetal Anomalies and Discovery of GREB1L Mutations in Renal Agenesis. Obstetrical and Gynecological Survey, 2018, 73, 677-679.	0.2	2
86	MYOD1 involvement in myopathy. European Journal of Neurology, 2018, 25, e123-e124.	1.7	8
87	BAFopathiesâ€™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Nature Communications, 2018, 9, 4885.	5.8	83
88	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nature Communications, 2018, 9, 4619.	5.8	70
89	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.	2.6	44
90	Lowry-Wood syndrome: further evidence of association with RNU4ATAC, and correlation between genotype and phenotype. Human Genetics, 2018, 137, 905-909.	1.8	11

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91	Hot water epilepsy and <scp>SYN</scp>1 variants. Epilepsia, 2018, 59, 2162-2163.	2.6	23
92	Arginase overexpression in neurons and its effect on traumatic brain injury. Molecular Genetics and Metabolism, 2018, 125, 112-117.	0.5	22
93	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. Brain, 2018, 141, 2299-2311.	3.7	81
94	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.	1.1	14
95	MicroRNA miR-23a cluster promotes osteocyte differentiation by regulating TGF-Î² signalling in osteoblasts. Nature Communications, 2017, 8, 15000.	5.8	91
96	Yunis-VarÃ³n syndrome caused by biallelic VAC14 mutations. European Journal of Human Genetics, 2017, 25, 1049-1054.	1.4	21
97	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.	0.5	18
98	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.	3.1	28
99	Retrospective Analysis of Congenital Scoliosis. Spine, 2017, 42, E841-E847.	1.0	13
100	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	1.4	39
101	Mutations in the phosphatidylinositol glycan C (PIGC) gene are associated with epilepsy and intellectual disability. Journal of Medical Genetics, 2017, 54, 196-201.	1.5	44
102	The spectrum of infantile myofibromatosis includes both non-penetrance and adult recurrence. European Journal of Medical Genetics, 2017, 60, 353-358.	0.7	23
103	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. American Journal of Human Genetics, 2017, 100, 91-104.	2.6	72
104	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
105	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.	2.6	49
106	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27.	1.2	165
107	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.	0.4	13
108	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€. American Journal of Human Genetics, 2017, 101, 815-823.	2.6	37

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109	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	1.1	27
110	Genetic Testing in a Cohort of Complex Esophageal Atresia. Molecular Syndromology, 2017, 8, 236-243.	0.3	7
111	Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases. Clinical Genetics, 2017, 91, 868-880.	1.0	14
112	Genomic approaches to diagnose rare bone disorders. Bone, 2017, 102, 5-14.	1.4	15
113	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. American Journal of Medical Genetics, Part A, 2017, 173, 733-739.	0.7	8
114	Spondyloepimetaphysial Dysplasia with Joint Laxity in Three Siblings with B3GALT6 Mutations. Molecular Syndromology, 2017, 8, 303-307.	0.3	7
115	Loss of DDRGK1 modulates SOX9 ubiquitination in spondyloepimetaphyseal dysplasia. Journal of Clinical Investigation, 2017, 127, 1475-1484.	3.9	46
116	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.	0.8	24
117	Epilepsy in KCNH1â€related syndromes. Epileptic Disorders, 2016, 18, 123-136.	0.7	34
118	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.	2.6	111
119	FHF1 (FGF12) epileptic encephalopathy. Neurology: Genetics, 2016, 2, e115.	0.9	32
120	TBC1D24 genotypeâ€“phenotype correlation. Neurology, 2016, 87, 77-85.	1.5	97
121	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.	2.6	36
122	Biosynthesis of glycosaminoglycans: associated disorders and biochemical tests. Journal of Inherited Metabolic Disease, 2016, 39, 173-188.	1.7	45
123	LMX1B and the Nail-Patella Syndrome. , 2016, , 741-746.		0
124	Adult presentation of Xâ€linked Conradiâ€HÃ¼nermannâ€Happle syndrome. American Journal of Medical Genetics, Part A, 2015, 167, 1309-1314.	0.7	6
125	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.	1.5	27
126	A crossâ€sectional multicenter study ofÂosteogenesis imperfecta in North AmericaÂâ€“Âresults from the linked clinical research centers. Clinical Genetics, 2015, 87, 133-140.	1.0	59

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127	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.	2.6	102
128	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. Nature Genetics, 2015, 47, 661-667.	9.4	177
129	FBN1 contributing to familial congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2015, 167, 831-836.	0.7	24
130	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. European Journal of Human Genetics, 2014, 22, 1272-1277.	1.4	38
131	The Undernourished Neonatal Mouse Metabolome Reveals Evidence of Liver and Biliary Dysfunction, Inflammation, and Oxidative Stress. Journal of Nutrition, 2014, 144, 273-281.	1.3	38
132	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.	0.7	40
133	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.	1.8	34
134	Genotype-Phenotype Correlation. Obstetrical and Gynecological Survey, 2014, 69, 728-730.	0.2	2
135	The genetic basis of DOORS syndrome: an exome-sequencing study. Lancet Neurology, The, 2014, 13, 44-58.	4.9	108
136	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. Human Molecular Genetics, 2014, 23, R1-R8.	1.4	234
137	A longitudinal study of urea cycle disorders. Molecular Genetics and Metabolism, 2014, 113, 127-130.	0.5	153
138	Argininosuccinate lyase in enterocytes protects from development of necrotizing enterocolitis. American Journal of Physiology - Renal Physiology, 2014, 307, G347-G354.	1.6	21
139	Genotypeâ€“Phenotype Correlation â€” Promiscuity in the Era of Next-Generation Sequencing. New England Journal of Medicine, 2014, 371, 593-596.	13.9	86
140	Diagnosis of ALG12-CDG by exome sequencing in a case of severe skeletal dysplasia. Molecular Genetics and Metabolism Reports, 2014, 1, 213-219.	0.4	16
141	Urea Cycle. , 2014, , 134-151.		0
142	Next-generation sequencing for disorders of low and high bone mineral density. Osteoporosis International, 2013, 24, 2253-2259.	1.3	46
143	Phenotypic Variability of Osteogenesis Imperfecta Type V Caused by an IFITM 5 Mutation. Journal of Bone and Mineral Research, 2013, 28, 1523-1530.	3.1	67
144	Identification of Novel Mutations ConfirmsPde4das a Major Gene Causing Acrodysostosis. Human Mutation, 2013, 34, 97-102.	1.1	49

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145	Yunis-VarÃ³n Syndrome Is Caused by Mutations in FIG4, Encoding a Phosphoinositide Phosphatase. American Journal of Human Genetics, 2013, 92, 781-791.	2.6	124
146	A Recurrent PDGFRB Mutation Causes Familial Infantile Myofibromatosis. American Journal of Human Genetics, 2013, 92, 996-1000.	2.6	135
147	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.	0.7	79
148	WNT1 Mutations in Early-Onset Osteoporosis and Osteogenesis Imperfecta. New England Journal of Medicine, 2013, 368, 1809-1816.	13.9	308
149	MCTP2 is a dosage-sensitive gene required for cardiac outflow tract development. Human Molecular Genetics, 2013, 22, 4339-4348.	1.4	40
150	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.	3.3	58
151	Early childhood presentation of Czech dysplasia. Clinical Dysmorphology, 2013, 22, 76-80.	0.1	9
152	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.	3.1	26
153	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.	2.0	18
154	Whole-exome sequencing identifies mutations in the nucleoside transporter gene SLC29A3 in dysosteosclerosis, a form of osteopetrosis. Human Molecular Genetics, 2012, 21, 4904-4909.	1.4	81
155	TheKAT6B-related disorders genitopatellar syndrome and Ohdo/SBBYS syndrome have distinct clinical features reflecting distinct molecular mechanisms. Human Mutation, 2012, 33, 1520-1525.	1.1	68
156	miRNA-34c regulates Notch signaling during bone development. Human Molecular Genetics, 2012, 21, 2991-3000.	1.4	210
157	Mutations in KAT6B, Encoding a Histone Acetyltransferase, Cause Genitopatellar Syndrome. American Journal of Human Genetics, 2012, 90, 282-289.	2.6	112
158	Nitric-Oxide Supplementation for Treatment of Long-Term Complications in Argininosuccinic Aciduria. American Journal of Human Genetics, 2012, 90, 836-846.	2.6	73
159	Requirement of argininosuccinate lyase for systemic nitric oxide production. Nature Medicine, 2011, 17, 1619-1626.	15.2	189
160	Management of West Syndrome in a Patient With Methylmalonic Aciduria. Journal of Child Neurology, 2010, 25, 94-97.	0.7	12
161	Early orthotopic liver transplantation in urea cycle defects: Follow up of a developmental outcome study. Molecular Genetics and Metabolism, 2010, 100, S84-S87.	0.5	53
162	Selective Inhibition of CCR2 Expressing Lymphomyeloid Cells in Experimental Autoimmune Encephalomyelitis by a GM-CSF-MCP1 Fusokine. Journal of Immunology, 2009, 182, 2620-2627.	0.4	21

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163	Mesenchymal Stromal Cells Engineered to Express Erythropoietin Induce Anti-erythropoietin Antibodies and Anemia in Allorecipients. Molecular Therapy, 2009, 17, 369-372.	3.7	34
164	Clinical Heterogeneity in Ethylmalonic Encephalopathy. Journal of Child Neurology, 2009, 24, 991-996.	0.7	40
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