

# Philippe M Campeau

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	C18orf32 loss-of-function is associated with a neurodevelopmental disorder with hypotonia and contractures.. <i>Human Genetics</i> , <b>2022</b> , 1	6.3	
164	Novel diagnostic DNA methylation epesignatures expand and refine the epigenetic landscapes of Mendelian disorders.. <i>Human Genetics and Genomics Advances</i> , <b>2022</b> , 3, 100075	0.8	1
163	A Discussion With Dr. Philippe Campeau, Medical Geneticist and Clinician-Scientist.. <i>Clinical and Investigative Medicine</i> , <b>2022</b> , 45, E5-8	0.9	
162	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> , <b>2021</b> , 22, 816	2.8	0
161	International Consensus Statement on the diagnosis, multidisciplinary management and lifelong care of individuals with achondroplasia. <i>Nature Reviews Endocrinology</i> , <b>2021</b> ,	15.2	4
160	Expanding the Phenotypic Spectrum of GPI Anchoring Deficiency Due to Biallelic Variants in. <i>Neurology: Genetics</i> , <b>2021</b> , 7, e631	3.8	
159	Free GPI is the elusive Emm antigen. <i>Blood</i> , <b>2021</b> , 137, 3588-3589	2.2	0
158	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> , <b>2021</b> , 58, 653-665	5.8	2
157	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. <i>American Journal of Human Genetics</i> , <b>2021</b> , 108, 929-941 <sup>11</sup>		0
156	Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy. <i>Human Genetics</i> , <b>2021</b> , 140, 1109-1120	6.3	4
155	Rickets manifestations in a child with metaphyseal anadysplasia, report of a spontaneously resolving case. <i>BMC Pediatrics</i> , <b>2021</b> , 21, 248	2.6	
154	Response to Gao et al. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 1580-1581	8.1	
153	PIGG variant pathogenicity assessment reveals characteristic features within 19 families. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 1873-1881	8.1	1
152	Epileptic encephalopathy caused by ARV1 deficiency: Refinement of the genotype-phenotype spectrum and functional impact on GPI-anchored proteins. <i>Clinical Genetics</i> , <b>2021</b> , 100, 607-614	4	1
151	PIGH deficiency can be associated with severe neurodevelopmental and skeletal manifestations. <i>Clinical Genetics</i> , <b>2021</b> , 99, 313-317	4	4
150	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 374-383	8.1	0
149	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> , <b>2021</b> , 2, 100015	0.8	2

148	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> , <b>2021</b> , 108, 134-147	11	4
147	DOORS syndrome and a recurrent truncating ATP6V1B2 variant. <i>Genetics in Medicine</i> , <b>2021</b> , 23, 149-154	8.1	6
146	Expanding the phenotype of PIGS-associated early onset epileptic developmental encephalopathy. <i>Epilepsia</i> , <b>2021</b> , 62, e35-e41	6.4	3
145	PIGF deficiency causes a phenotype overlapping with DOORS syndrome. <i>Human Genetics</i> , <b>2021</b> , 140, 879-884	6.3	1
144	Disruption of exon-bridging interactions between the minor and major spliceosomes results in alternative splicing around minor introns. <i>Nucleic Acids Research</i> , <b>2021</b> , 49, 3524-3545	20.1	1
143	Heterozygous variant in gene in two brothers with early onset osteoporosis. <i>Bone Reports</i> , <b>2021</b> , 15, 101118	2.6	0
142	Calvarial doughnut lesions with bone fragility in a French-Canadian family; case report and review of the literature. <i>Bone Reports</i> , <b>2021</b> , 15, 101121	2.6	
141	Variant-specific effects define the phenotypic spectrum of HNRNPH2-associated neurodevelopmental disorders in males.. <i>Human Genetics</i> , <b>2021</b> , 141, 257	6.3	0
140	Variable expressivity in a family with an aggrecanopathy. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2021</b> , e1773	2.3	2
139	Clinical characteristics of patients from Quebec, Canada, with Morquio A syndrome: a longitudinal observational study. <i>Orphanet Journal of Rare Diseases</i> , <b>2020</b> , 15, 270	4.2	4
138	Further delineation of the clinical spectrum of KAT6B disorders and allelic series of pathogenic variants. <i>Genetics in Medicine</i> , <b>2020</b> , 22, 1338-1347	8.1	9
137	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. <i>European Journal of Human Genetics</i> , <b>2020</b> , 28, 1422-1431	5.3	10
136	Disrupted minor intron splicing is prevalent in Mendelian disorders. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2020</b> , 8, e1374	2.3	5
135	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. <i>Neuron</i> , <b>2020</b> , 106, 404-420.e8	13.9	49
134	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in PIGQ: Report of seven new subjects and review of the literature. <i>Journal of Inherited Metabolic Disease</i> , <b>2020</b> , 43, 1321-1332	5.4	6
133	The Canadian Rare Diseases Models and Mechanisms (RDMM) Network: Connecting Understudied Genes to Model Organisms. <i>American Journal of Human Genetics</i> , <b>2020</b> , 106, 143-152	11	16
132	Evaluation of DNA Methylation Episignatures for Diagnosis and Phenotype Correlations in 42 Mendelian Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , <b>2020</b> , 106, 356-370	11	51
131	Deficient histone H3 propionylation by BRPF1-KAT6 complexes in neurodevelopmental disorders and cancer. <i>Science Advances</i> , <b>2020</b> , 6, eaax0021	14.3	24

130	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> , <b>2020</b> , 106, 484-495	11	16
129	MYSM1 maintains ribosomal protein gene expression in hematopoietic stem cells to prevent hematopoietic dysfunction. <i>JCI Insight</i> , <b>2020</b> , 5,	9.9	2
128	Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. <i>Journal of Clinical Investigation</i> , <b>2020</b> , 130, 1431-1445	15.9	17
127	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> , <b>2020</b> , 28, 461-468	5.3	6
126	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> , <b>2020</b> , 4, e10335	3.9	1
125	Retrospective analysis of fetal vertebral defects: Associated anomalies, etiologies, and outcome. <i>American Journal of Medical Genetics, Part A</i> , <b>2020</b> , 182, 664-672	2.5	3
124	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> , <b>2020</b> , 63, 103822	2.6	5
123	De Novo KAT5 Variants Cause a Syndrome with Recognizable Facial Dysmorphisms, Cerebellar Atrophy, Sleep Disturbance, and Epilepsy. <i>American Journal of Human Genetics</i> , <b>2020</b> , 107, 564-574	11	8
122	Clinicopathological Relationships in an Aged Case of DOORS Syndrome With a p.Arg506X Mutation in the Gene. <i>Frontiers in Neurology</i> , <b>2020</b> , 11, 767	4.1	4
121	A homozygous variant in the Lamin B receptor gene LBR results in a non-lethal skeletal dysplasia without Pelger-Huët anomaly. <i>Bone</i> , <b>2020</b> , 141, 115601	4.7	0
120	Biallelic variants in GLE1 with survival beyond neonatal period. <i>Clinical Genetics</i> , <b>2020</b> , 98, 622-625	4	
119	Nonsyndromic erythrodermic ichthyosis resulting from a homozygous mutation in PIGL. <i>Clinical and Experimental Dermatology</i> , <b>2020</b> , 45, 391-394	1.8	1
118	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> , <b>2020</b> , 63, 103784	2.6	2
117	The CHD4-related syndrome: a comprehensive investigation of the clinical spectrum, genotype-phenotype correlations, and molecular basis. <i>Genetics in Medicine</i> , <b>2020</b> , 22, 389-397	8.1	22
116	Bruck syndrome 2 variant lacking congenital contractures and involving a novel compound heterozygous PLOD2 mutation. <i>Bone</i> , <b>2020</b> , 130, 115047	4.7	9
115	Juvenile Paget's Disease From Heterozygous Mutation of SP7 Encoding Osterix (Specificity Protein 7, Transcription Factor SP7). <i>Bone</i> , <b>2020</b> , 137, 115364	4.7	7
114	Genetics of the patella. <i>European Journal of Human Genetics</i> , <b>2019</b> , 27, 671-680	5.3	4
113	Gain-of-Function Mutations in KCNN3 Encoding the Small-Conductance Ca-Activated K Channel SK3 Cause Zimmermann-Laband Syndrome. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 1139-1157	11	20

112	Biallelic variants in the transcription factor PAX7 are a new genetic cause of myopathy. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 2521-2531	8.1	17
111	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 815-834	11	21
110	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> , <b>2019</b> , 104, 596-610	11	22
109	Clinical variability in inherited glycosylphosphatidylinositol deficiency disorders. <i>Clinical Genetics</i> , <b>2019</b> , 95, 112-121	4	52
108	Inherited glycosylphosphatidylinositol deficiency variant database and analysis of pathogenic variants. <i>Molecular Genetics &amp; Genomic Medicine</i> , <b>2019</b> , 7, e00743	2.3	9
107	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. <i>American Journal of Human Genetics</i> , <b>2019</b> , 105, 534-548	11	25
106	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> , <b>2019</b> , 105, 625-630	11	22
105	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> , <b>2019</b> , 105, 384-394	11	29
104	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 530-541	11	17
103	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. <i>American Journal of Human Genetics</i> , <b>2019</b> , 104, 422-438	11	10
102	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> , <b>2019</b> , 56, 701-710	5.8	22
101	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> , <b>2019</b> , 105, 1237-1253	11	17
100	IQSEC2-related encephalopathy in males and females: a comparative study including 37 novel patients. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 837-849	8.1	32
99	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> , <b>2019</b> , 104, 164-178	11	27
98	Novel fibronectin mutations and expansion of the phenotype in spondylometaphyseal dysplasia with "corner fractures". <i>Bone</i> , <b>2019</b> , 121, 163-171	4.7	7
97	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffin-Siris syndrome. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 1295-1307	8.1	36
96	The epilepsy-associated protein TBC1D24 is required for normal development, survival and vesicle trafficking in mammalian neurons. <i>Human Molecular Genetics</i> , <b>2019</b> , 28, 584-597	5.6	23
95	A new microdeletion syndrome involving TBC1D24, ATP6V0C, and PDPK1 causes epilepsy, microcephaly, and developmental delay. <i>Genetics in Medicine</i> , <b>2019</b> , 21, 1058-1064	8.1	12

94	Recessive mutations in VPS13D cause childhood onset movement disorders. <i>Annals of Neurology</i> , <b>2018</b> , 83, 1089-1095	9.4	61
93	A PIGH mutation leading to GPI deficiency is associated with developmental delay and autism. <i>Human Mutation</i> , <b>2018</b> , 39, 827-829	4.7	10
92	Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La Réunion Island, in patients with Fryns syndrome. <i>European Journal of Human Genetics</i> , <b>2018</b> , 26, 340-349	5.3	18
91	Dysregulation of cotranscriptional alternative splicing underlies CHARGE syndrome. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , <b>2018</b> , 115, E620-E629	11.5	19
90	Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. <i>American Journal of Human Genetics</i> , <b>2018</b> , 102, 156-174	11.4	75
89	FRMPD4 mutations cause X-linked intellectual disability and disrupt dendritic spine morphogenesis. <i>Human Molecular Genetics</i> , <b>2018</b> , 27, 589-600	5.6	8
88	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> , <b>2018</b> , 20, 294-302	8.1	20
87	Genomic study of severe fetal anomalies and discovery of GREB1L mutations in renal agenesis. <i>Genetics in Medicine</i> , <b>2018</b> , 20, 745-753	8.1	38
86	Arginase overexpression in neurons and its effect on traumatic brain injury. <i>Molecular Genetics and Metabolism</i> , <b>2018</b> , 125, 112-117	3.7	13
85	BCL11B mutations in patients affected by a neurodevelopmental disorder with reduced type 2 innate lymphoid cells. <i>Brain</i> , <b>2018</b> , 141, 2299-2311	11.2	36
84	MYOD1 involvement in myopathy. <i>European Journal of Neurology</i> , <b>2018</b> , 25, e123-e124	6	6
83	BAFopathies and DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffin-Siris and Nicolaides-Baraitser syndromes. <i>Nature Communications</i> , <b>2018</b> , 9, 4885	17.4	48
82	CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. <i>Nature Communications</i> , <b>2018</b> , 9, 4619	17.4	39
81	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> , <b>2018</b> , 103, 602-611	11	32
80	Lowry-Wood syndrome: further evidence of association with RNU4ATAC, and correlation between genotype and phenotype. <i>Human Genetics</i> , <b>2018</b> , 137, 905-909	6.3	5
79	Hot water epilepsy and SYN1 variants. <i>Epilepsia</i> , <b>2018</b> , 59, 2162-2163	6.4	13
78	Kaufman oculo-cerebro-facial syndrome in a child with small and absent terminal phalanges and absent nails. <i>Journal of Human Genetics</i> , <b>2017</b> , 62, 465-471	4.3	11
77	MicroRNA miR-23a cluster promotes osteocyte differentiation by regulating TGF- $\beta$ signalling in osteoblasts. <i>Nature Communications</i> , <b>2017</b> , 8, 15000	17.4	63



76	Yunis-Varfi syndrome caused by biallelic VAC14 mutations. <i>European Journal of Human Genetics</i> , <b>2017</b> , 25, 1049-1054	5.3	16
75	An exome sequencing study of Moebius syndrome including atypical cases reveals an individual with CFEOM3A and a mutation. <i>Journal of Physical Education and Sports Management</i> , <b>2017</b> , 3, a000984	2.8	12
74	A Novel PGM3 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> , <b>2017</b> , 32, 1853-1859	6.3	20
73	Retrospective Analysis of Congenital Scoliosis: Associated Anomalies and Genetic Diagnoses. <i>Spine</i> , <b>2017</b> , 42, E841-E847	3.3	8
72	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. <i>Human Molecular Genetics</i> , <b>2017</b> , 26, 1706-1715	5.6	33
71	Mutations in the phosphatidylinositol glycan C () gene are associated with epilepsy and intellectual disability. <i>Journal of Medical Genetics</i> , <b>2017</b> , 54, 196-201	5.8	36
70	The spectrum of infantile myofibromatosis includes both non-penetrance and adult recurrence. <i>European Journal of Medical Genetics</i> , <b>2017</b> , 60, 353-358	2.6	19
69	Mutations in the Chromatin Regulator Gene BRPF1 Cause Syndromic Intellectual Disability and Deficient Histone Acetylation. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 91-104	11	43
68	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 664-685	11	214
67	Mutations in GPAA1, Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 856-865	11	35
66	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. <i>Genetics</i> , <b>2017</b> , 207, 9-27	4	99
65	A non-mosaic mutation in a male with severe congenital anomalies overlapping focal dermal hypoplasia. <i>Molecular Genetics and Metabolism Reports</i> , <b>2017</b> , 12, 57-61	1.8	7
64	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with "Corner Fractures". <i>American Journal of Human Genetics</i> , <b>2017</b> , 101, 815-823	11	22
63	Heterozygous variants in ACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. <i>Human Mutation</i> , <b>2017</b> , 38, 1365-1371	4.7	17
62	Genetic Testing in a Cohort of Complex Esophageal Atresia. <i>Molecular Syndromology</i> , <b>2017</b> , 8, 236-243	1.5	5
61	Chondrodysplasia with multiple dislocations: comprehensive study of a series of 30 cases. <i>Clinical Genetics</i> , <b>2017</b> , 91, 868-880	4	8
60	Genomic approaches to diagnose rare bone disorders. <i>Bone</i> , <b>2017</b> , 102, 5-14	4.7	12
59	Corner fracture type spondylometaphyseal dysplasia: Overlap with type II collagenopathies. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 733-739	2.5	5

58	Spondyloepimetaphysial Dysplasia with Joint Laxity in Three Siblings with Mutations. <i>Molecular Syndromology</i> , <b>2017</b> , 8, 303-307	1.5	5
57	Loss of DDRGK1 modulates SOX9 ubiquitination in spondyloepimetaphyseal dysplasia. <i>Journal of Clinical Investigation</i> , <b>2017</b> , 127, 1475-1484	15.9	20
56	Case Report: Novel mutations in are associated with autosomal dominant tonic-clonic and myoclonic epilepsy and recessive Parkinsonism, psychosis, and intellectual disability. <i>F1000Research</i> , <b>2017</b> , 6, 553	3.6	20
55	De Novo Mutations in CHD4, an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. <i>American Journal of Human Genetics</i> , <b>2016</b> , 99, 934-941	11	68
54	FHF1 (FGF12) epileptic encephalopathy. <i>Neurology: Genetics</i> , <b>2016</b> , 2, e115	3.8	22
53	TBC1D24 genotype-phenotype correlation: Epilepsies and other neurologic features. <i>Neurology</i> , <b>2016</b> , 87, 77-85	6.5	75
52	Small 6q16.1 Deletions Encompassing POU3F2 Cause Susceptibility to Obesity and Variable Developmental Delay with Intellectual Disability. <i>American Journal of Human Genetics</i> , <b>2016</b> , 98, 363-72	11	26
51	Biosynthesis of glycosaminoglycans: associated disorders and biochemical tests. <i>Journal of Inherited Metabolic Disease</i> , <b>2016</b> , 39, 173-88	5.4	30
50	Epilepsy in KCNH1-related syndromes. <i>Epileptic Disorders</i> , <b>2016</b> , 18, 123-36	1.9	21
49	A cross-sectional multicenter study of osteogenesis imperfecta in North America - results from the linked clinical research centers. <i>Clinical Genetics</i> , <b>2015</b> , 87, 133-40	4	45
48	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> , <b>2015</b> , 96, 816-25	11	75
47	Mutations in KCNH1 and ATP6V1B2 cause Zimmermann-Laband syndrome. <i>Nature Genetics</i> , <b>2015</b> , 47, 661-7	36.3	128
46	FBN1 contributing to familial congenital diaphragmatic hernia. <i>American Journal of Medical Genetics, Part A</i> , <b>2015</b> , 167A, 831-6	2.5	19
45	Adult presentation of X-linked Conradi-Hünermann-Happle syndrome. <i>American Journal of Medical Genetics, Part A</i> , <b>2015</b> , 167, 1309-14	2.5	5
44	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> , <b>2015</b> , 52, 438-45	5.8	23
43	A longitudinal study of urea cycle disorders. <i>Molecular Genetics and Metabolism</i> , <b>2014</b> , 113, 127-30	3.7	106
42	Argininosuccinate lyase in enterocytes protects from development of necrotizing enterocolitis. <i>American Journal of Physiology - Renal Physiology</i> , <b>2014</b> , 307, G347-54	5.1	17
41	Genotype-phenotype correlation--promiscuity in the era of next-generation sequencing. <i>New England Journal of Medicine</i> , <b>2014</b> , 371, 593-6	59.2	68



40	Diagnosis of ALG12-CDG by exome sequencing in a case of severe skeletal dysplasia. <i>Molecular Genetics and Metabolism Reports</i> , <b>2014</b> , 1, 213-219	1.8	14
39	Myhre and LAPS syndromes: clinical and molecular review of 32 patients. <i>European Journal of Human Genetics</i> , <b>2014</b> , 22, 1272-7	5.3	32
38	The undernourished neonatal mouse metabolome reveals evidence of liver and biliary dysfunction, inflammation, and oxidative stress. <i>Journal of Nutrition</i> , <b>2014</b> , 144, 273-81	4.1	26
37	DOORS syndrome: phenotype, genotype and comparison with Coffin-Siris syndrome. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , <b>2014</b> , 166C, 327-32	3.1	33
36	Exome sequencing identifies a novel homozygous mutation in the phosphate transporter SLC34A1 in hypophosphatemia and nephrocalcinosis. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2014</b> , 99, E2451-6	5.6	29
35	The genetic basis of DOORS syndrome: an exome-sequencing study. <i>Lancet Neurology</i> , <b>2014</b> , 13, 44-58	24.1	96
34	Branched-chain amino acid metabolism: from rare Mendelian diseases to more common disorders. <i>Human Molecular Genetics</i> , <b>2014</b> , 23, R1-8	5.6	73
33	Next-generation sequencing for disorders of low and high bone mineral density. <i>Osteoporosis International</i> , <b>2013</b> , 24, 2253-9	5.3	42
32	Phenotypic variability of osteogenesis imperfecta type V caused by an IFITM5 mutation. <i>Journal of Bone and Mineral Research</i> , <b>2013</b> , 28, 1523-30	6.3	55
31	Identification of novel mutations confirms PDE4D as a major gene causing acrodysostosis. <i>Human Mutation</i> , <b>2013</b> , 34, 97-102	4.7	46
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