Michael B Petersen

List of Publications by Year
in descending order

127	4,313	117453	128067
papers	citations	34 h-index	60 g-index

129	129	129	5529
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	A wide range of protective and predisposing variants in aggrecan influence the susceptibility for otosclerosis. Human Genetics, 2022, 141, 951-963.	1.8	6
2	First reported CABP2â€related nonâ€syndromic hearing loss in Northern Europe. Molecular Genetics & Genomic Medicine, 2021, 9, e1639.	0.6	6
3	First reported case of Doyne honeycomb retinal dystrophy (Malattia Leventinese/autosomal dominant) Tj ETQq1 1 0.784314 rgBT /Over	0.6	3
4	Recurrent arginine substitutions in the ACTG2 gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654.	1.1	27
5	Mitochondrial Disease Caused by a Novel Homozygous Mutation (Gly106del) in the SCO1 Gene. Neonatology, 2019, 116, 290-294.	0.9	7
6	Whole-exome sequencing identifies a GREB1L variant in a three-generation family with MÃ¼llerian and renal agenesis: a novel candidate gene in Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser (MRKH) syndrome. A case report. Human Reproduction, 2019, 34, 1838-1846.	0.4	34
7	Partial trisomy 21 map: Ten cases further supporting the highly restricted Down syndrome critical region (HRâ€DSCR) on human chromosome 21. Molecular Genetics & Genomic Medicine, 2019, 7, e797.	0.6	28
8	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	2.6	70
9	Genotypeâ€phenotype correlations, dystonia and disease progression in spinocerebellar ataxia type 14. Movement Disorders, 2018, 33, 1119-1129.	2.2	26
10	First reported adult patient with TARP syndrome: A case report. American Journal of Medical Genetics, Part A, 2018, 176, 2915-2918.	0.7	16
11	Ocular albinism with infertility and lateâ€onset sensorineural hearing loss. American Journal of Medical Genetics, Part A, 2018, 176, 1587-1593.	0.7	6
12	De novo KAT6B Mutation Identified with Whole-Exome Sequencing in a Girl with Say-Barber/Biesecker/Young-Simpson Syndrome. Molecular Syndromology, 2017, 8, 24-29.	0.3	4
13	Investigation of associations of ARMS2, CD14, and TLR4 gene polymorphisms with wet age-related macular degeneration in a Greek population. Clinical Ophthalmology, 2017, Volume 11, 1347-1358.	0.9	9
14	A novel deletion mutation in the <scp>ALOX</scp>12B gene in a Kurdish family with autosomal recessive congenital ichthyosis. Journal of the European Academy of Dermatology and Venereology, 2016, 30, e144-e145.	1.3	2
15	Vertebral defect, anal atresia, cardiac defect, tracheoesophageal fistula/esophageal atresia, renal defect, and limb defect association with Mayer-Rokitansky-KÃ¼ster-Hauser syndrome in co-occurrence: two case reports and a review of the literature. Journal of Medical Case Reports, 2016, 10, 374.	0.4	20
16	Prevalence and patient characteristics of Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser syndrome: a nationwide registry-based study. Human Reproduction, 2016, 31, 2384-2390.	0.4	107
17	Re-Examination of Danish Carriers of Balanced Chromosomal Inversions. Cancer Genetics, 2016, 209, 231.	0.2	0
18	The <i>PHF6</i> Mutation c.1A>G; pM1V Causes BÃ¶rjeson-Forsman-Lehmann Syndrome in a Family with Four Affected Young Boys. Molecular Syndromology, 2015, 6, 181-186.	0.3	5

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19	35-Year Follow-Up of a Case of Ring Chromosome 2: Array-CGH Analysis and Literature Review of the Ring Syndrome. Cytogenetic and Genome Research, 2015, 145, 6-13.	0.6	2
20	Oxidative stress in dry age-related macular degeneration and exfoliation syndrome. Critical Reviews in Clinical Laboratory Sciences, 2015, 52, 12-27.	2.7	44
21	Novel association of FCGR2A polymorphism with age-related macular degeneration (AMD) and development of a novel CFH real-time genotyping method. Clinical Chemistry and Laboratory Medicine, 2015, 53, 1521-9.	1.4	6
22	ECEL1 mutation causes fetal arthrogryposis multiplex congenita. American Journal of Medical Genetics, Part A, 2015, 167, 731-743.	0.7	28
23	A de novo 2.9ÂMb interstitial deletion at 13q12.11 in a child with developmental delay accompanied by mild dysmorphic characteristics. Molecular Cytogenetics, 2014, 7, 92.	0.4	2
24	Familial occurrence of Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser syndrome: A case report and review of the literature. American Journal of Medical Genetics, Part A, 2014, 164, 2276-2286.	0.7	61
25	Loss-of-function HDAC8 mutations cause a phenotypic spectrum of Cornelia de Lange syndrome-like features, ocular hypertelorism, large fontanelle and X-linked inheritance. Human Molecular Genetics, 2014, 23, 2888-2900.	1.4	120
26	De novo 393kb microdeletion of 7p11.2 characterized by aCGH in a boy with psychomotor retardation and dysmorphic features. Meta Gene, 2014, 2, 274-282.	0.3	6
27	Pure de novo partial trisomy 6p in a girl with craniosynostosis. American Journal of Medical Genetics, Part A, 2013, 161, 343-351.	0.7	18
28	SLITRK6 mutations cause myopia and deafness in humans and mice. Journal of Clinical Investigation, 2013, 123, 2094-2102.	3.9	50
29	Prenatal detection of an inverted duplication deletion in the long arm of chromosome 1 in a fetus with increased nuchal translucency. Molecular cytogenetic analysis and review of the literature. Clinical Dysmorphology, 2012, 21, 101-105.	0.1	1
30	Biomarkers in primary open angle glaucoma. Clinical Chemistry and Laboratory Medicine, 2012, 50, 2107-2119.	1.4	19
31	A New Multicolor Fluorescence In Situ Hybridization Probe Set Directed Against Human Heterochromatin. Journal of Histochemistry and Cytochemistry, 2012, 60, 530-536.	1.3	17
32	HDAC8 mutations in Cornelia de Lange syndrome affect the cohesin acetylation cycle. Nature, 2012, 489, 313-317.	13.7	488
33	Compound heterozygosity of the novel c.292C > T (p.R98W) and the c.35delG GJB2 mutations in postlingual, non-syndromic, sensorineural deafness. International Journal of Pediatric Otorhinolaryngology, 2012, 76, 549-551.	0.4	6
34	The novel c.247_249delTTC (p.F83del) GJB2 mutation in a family with prelingual sensorineural deafness. International Journal of Pediatric Otorhinolaryngology, 2012, 76, 969-971.	0.4	4
35	Unexpected results in the constitution of small supernumerary marker chromosomes. European Journal of Medical Genetics, 2012, 55, 185-190.	0.7	22
36	Erythrokeratodermia variabilis: Report of two cases and a novel missense variant in GJB4 encoding connexin 30.3. European Journal of Dermatology, 2012, 22, 182-186.	0.3	4

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37	Combined 22q11.1-q11.21 deletion with 15q11.2-q13.3 duplication identified by array-CGH in a 6 years old boy. Molecular Cytogenetics, 2011, 4, 6.	0.4	6
38	Homoplasmy of the G7444A mtDNA and heterozygosity of the GJB2 c.35delG mutations in a family with hearing loss. International Journal of Pediatric Otorhinolaryngology, 2011, 75, 89-94.	0.4	9
39	Detection of Deafness-Causing Mutations in the Greek Mitochondrial Genome. Disease Markers, 2011, 30, 283-289.	0.6	12
40	Deletion 2q31.2â€q31.3 in a 4â€yearâ€old girl with microcephaly and severe mental retardation. American Journal of Medical Genetics, Part A, 2011, 155, 1476-1482.	0.7	8
41	Complex distal 10q rearrangement in a girl with mild intellectual disability: Follow up of the patient and review of the literature of nonâ€acrocentric satellited chromosomes. American Journal of Medical Genetics, Part A, 2011, 155, 2841-2854.	0.7	9
42	Recommendations for genetic variation data capture in developing countries to ensure a comprehensive worldwide data collection. Human Mutation, 2011, 32, 2-9.	1.1	25
43	Age-related macular degeneration: genetic and clinical findings. Clinical Chemistry and Laboratory Medicine, 2011, 49, 601-616.	1.4	43
44	Antenatal diagnosis of duodenal atresia in dizygotic twins associated with congenital hearing impairment. Journal of Maternal-Fetal and Neonatal Medicine, 2011, 24, 871-872.	0.7	0
45	A Truncating Mutation in SERPINB6 Is Associated with Autosomal-Recessive Nonsyndromic Sensorineural Hearing Loss. American Journal of Human Genetics, 2010, 86, 797-804.	2.6	56
46	Multiple enhancers located in a 1-Mb region upstream of POU3F4 promote expression during inner ear development and may be required for hearing. Human Genetics, 2010, 128, 411-419.	1.8	35
47	The effect of human embryonic stem cells (hESCs) long-term normoxic and hypoxic cultures on the maintenance of pluripotency. In Vitro Cellular and Developmental Biology - Animal, 2010, 46, 276-283.	0.7	35
48	The use of array-CGH in a cohort of Greek children with developmental delay. Molecular Cytogenetics, 2010, 3, 22.	0.4	20
49	High frequency of COH1 intragenic deletions and duplications detected by MLPA in patients with Cohen syndrome. European Journal of Human Genetics, 2010, 18, 1133-1140.	1.4	31
50	Prenatal detection of full monosomy 21 in a fetus with increased nuchal translucency: Molecular cytogenetic analysis and review of the literature. Journal of Obstetrics and Gynaecology Research, 2010, 36, 435-440.	0.6	15
51	Easy, Rapid, and Cost-Effective Methods for Identifying Carriers of Recurrent GJB2 Mutations Causing Nonsyndromic Hearing Impairment in the Greek Population. Genetic Testing and Molecular Biomarkers, 2010, 14, 189-192.	0.3	1
52	Are GJB2 mutations an aggravating factor in the phenotypic expression of mitochondrial non-syndromic deafness?. Journal of Human Genetics, 2010, 55, 265-269.	1.1	7
53	Screening of a Greek deafness population for the A7445G mitochondrial DNA mutation. Molecular Genetics and Metabolism, 2010, 100, 300-301.	0.5	4
54	Hypothesizing an Ancient Greek Origin of theGJB235delG Mutation: Can Science Meet History?. Genetic Testing and Molecular Biomarkers, 2010, 14, 183-187.	0.3	12

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55	Investigating the Impact of the Down Syndrome Related Common MTHFR 677C>T Polymorphism in the Danish Population. Disease Markers, 2009, 27, 279-285.	0.6	18
56	Genetic skeletal disorders of the fetus and infant: Pathologic and molecular findings in a series of 41 cases. Birth Defects Research Part A: Clinical and Molecular Teratology, 2009, 85, 811-821.	1.6	14
57	Prenatal diagnosis of a fetus with ring chromosome 15 characterized by arrayâ€CGH. Prenatal Diagnosis, 2009, 29, 884-888.	1.1	7
58	Detailed molecular and clinical investigation of a child with a partial deletion of chromosome 11 (Jacobsen syndrome). Molecular Cytogenetics, 2009, 2, 26.	0.4	9
59	Phenotypic variability of patients homozygous for the GJB2 mutation 35delG cannot be explained by the influence of one major modifier gene. European Journal of Human Genetics, 2009, 17, 517-524.	1.4	46
60	The A1555G mitochondrial DNA mutation in Greek patients with non-syndromic, sensorineural hearing loss. Biochemical and Biophysical Research Communications, 2009, 390, 755-757.	1.0	12
61	Copy number variation and genomic alterations in health and disease. Genome Medicine, 2009, 1, 21.	3.6	4
62	Complex chromosome rearrangement in a child with microcephaly, dysmorphic facial features and mosaicism for a terminal deletion del(18)(q21.32-qter) investigated by FISH and array-CGH: Case report. Molecular Cytogenetics, 2008, 1, 24.	0.4	4
63	Pathologic, radiographic and molecular findings in three fetuses diagnosed with HEM/Greenberg skeletal dysplasia. Prenatal Diagnosis, 2008, 28, 309-312.	1.1	45
64	Prenatal diagnosis of a fetus with 7q11.23 deletion detected by multiplex ligationâ€dependent probe amplification (MLPA) screening. Prenatal Diagnosis, 2008, 28, 556-558.	1.1	13
65	Cohen syndrome resulting from a novel large intragenic COH1 deletion segregating in an isolated Greek island population. American Journal of Medical Genetics, Part A, 2008, 146A, 2221-2226.	0.7	26
66	Strong linkage disequilibrium for the frequent GJB2 35delG mutation in the Greek population. American Journal of Medical Genetics, Part A, 2008, 146A, 2879-2884.	0.7	22
67	Sudden hearing loss in a family with GJB2 related progressive deafness. International Journal of Pediatric Otorhinolaryngology, 2008, 72, 1735-1740.	0.4	10
68	Non-disjunction of chromosome 13. Human Molecular Genetics, 2007, 16, 2004-2010.	1.4	38
69	A report on 10 new patients with heterozygous mutations in theCOL11A1 gene and a review of genotypeâ€“phenotype correlations in type XI collagenopathies. American Journal of Medical Genetics, Part A, 2007, 143A, 258-264.	0.7	75
70	A seventh locus for otosclerosis, OTSC7, maps to chromosome 6q13â€“16.1. European Journal of Human Genetics, 2007, 15, 362-368.	1.4	53
71	Juvenile Paget's Disease: The Second Reported, Oldest Patient Is Homozygous for the TNFRSF11B â€œBalkanâ€•Mutation (966_969delTGACinsCTT), Which Elevates Circulating Immunoreactive Osteoprotegerin Levels. Journal of Bone and Mineral Research, 2007, 22, 938-946.	3.1	36
72	Monogenic nonsyndromic otosclerosis: Audiological and linkage analysis in a large Greek pedigree. International Journal of Pediatric Otorhinolaryngology, 2006, 70, 631-637.	0.4	10

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73	A LargeGLC1CGreek Family with a Myocilin T377M Mutation: Inheritance and Phenotypic Variability. , 2006, 47, 620.		17
74	Prenatal diagnosis of achondroplasia presenting with multiple-suture synostosis: a novel association. Prenatal Diagnosis, 2006, 26, 258-261.	1.1	13
75	Prenatal diagnosis of glycogen storage disease type IV. Prenatal Diagnosis, 2006, 26, 951-955.	1.1	32
76	3C syndrome with cryptorchidism and posterior embryotoxon. Clinical Dysmorphology, 2005, 14, 97-100.	0.1	5
77	Hellenic National Mutation Database: a prototype database for mutations leading to inherited disorders in the Hellenic population. Human Mutation, 2005, 25, 327-333.	1.1	53
78	Different mechanisms preclude mutant CLDN14 proteins from forming tight junctions in vitro. Human Mutation, 2005, 25, 543-549.	1.1	45
79	Clinical, cytogenetic, and molecular genetic characterization of two unrelated patients with different duplications of 21q. American Journal of Medical Genetics Part A, 2005, 37, 104-109.	2.4	8
80	Trisomy 13 due to rea(13q;13q) is caused by i(13) and not rob(13;13)(q10;q10) in the majority of cases. , 2005, 132A, 310-313.		9
81	Prenatal Diagnosis of Trisomy 2 Mosaicism: A Case Report. Fetal Diagnosis and Therapy, 2004, 19, 488-490.	0.6	10
82	DNA studies of mono- and pseudodicentric isochromosomes 18q. American Journal of Medical Genetics Part A, 2004, 127A, 230-233.	2.4	7
83	Double supernumerary isodicentric chromosomes derived from 15 resulting in partial hexasomy. American Journal of Medical Genetics Part A, 2003, 116A, 356-359.	2.4	17
84	Prevalence of GJB2 mutations in prelingual deafness in the Greek population. International Journal of Pediatric Otorhinolaryngology, 2002, 65, 101-108.	0.4	63
85	Human CCS gene: genomic organization and exclusion as a candidate for amyotrophic lateral sclerosis (ALS). BMC Genetics, 2002, 3, 5.	2.7	13
86	Mutations in the TMPRSS3 gene are a rare cause of childhood nonsyndromic deafness in Caucasian patients. Journal of Molecular Medicine, 2002, 80, 124-131.	1.7	65
87	Maternal uniparental isodisomy 20 in a foetus with trisomy 20 mosaicism: clinical, cytogenetic and molecular analysis. European Journal of Human Genetics, 2002, 10, 694-698.	1.4	40
88	Prenatal diagnosis of two rarede novo structural aberrations of the Y chromosome: cytogenetic and molecular analysis. Prenatal Diagnosis, 2001, 21, 484-487.	1.1	7
89	Prenatal diagnosis of prelingual deafness: carrier testing and prenatal diagnosis of the common GJB2 35delG mutation. Prenatal Diagnosis, 2001, 21, 10-13.	1.1	15
90	Detection of mutations in theCOL4A5gene by SSCP in X-linked Alport syndrome. Human Mutation, 2001, 18, 141-148.	1.1	38

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91	Genetic linkage of autosomal dominant primary open angle glaucoma to chromosome 3q in a Greek pedigree. European Journal of Human Genetics, 2001, 9, 452-457.	1.4	29
92	Molecular cytogenetic characterization and origin of two de novo duplication 9p cases. , 2000, 91, 102-106.		21
93	Association between presenilin-1 polymorphism and maternal meiosis II errors in Down syndrome. American Journal of Medical Genetics Part A, 2000, 93, 366-372.	2.4	26
94	BRCA1 mutation analysis in breast/ovarian cancer families from Greece. Human Mutation, 2000, 16, 272-273.	1.1	27
95	Genotyping of presenilin-1 polymorphism in amyotrophic lateral sclerosis. Journal of Neurology, 2000, 247, 940-942.	1.8	8
96	Patterns of Meiotic Recombination on the Long Arm of Human Chromosome 21. Genome Research, 2000, 10, 1319-1332.	2.4	47
97	Apolipoprotein E and presenilin-1 genotypes in Huntington's disease. Journal of Neurology, 1999, 246, 574-577.	1.8	35
98	Molecular and clinical correlation study of Williams-Beuren syndrome: No evidence of molecular factors in the deletion region or imprinting affecting clinical outcome. , 1999, 86, 34-43.		34
99	Origin of nondisjunction in trisomy 8 and trisomy 8 mosaicism. European Journal of Human Genetics, 1998, 6, 432-438.	1.4	59
100	FRAXA and FRAXE prevalence in patients with nonspecific mental retardation in the Hellenic population. Genetic Epidemiology, 1998, 15, 103-109.	0.6	25
101	Cytogenetic and molecular study of 32 Down syndrome families: potential leukaemia predisposing role of the most proximal segment of chromosome 21q. British Journal of Haematology, 1998, 103, 213-216.	1.2	23
102	Analysis of the Origin of the Extra Chromosome in Trisomy 8 in 4 Cases of Spontaneous Abortions. Fetal Diagnosis and Therapy, 1998, 13, 42-45.	0.6	4
103	Apolipoprotein E alleles in mothers of trisomy 18 conceptuses. Clinical Genetics, 1998, 53, 321-322.	1.0	2
104	Apolipoprotein E îœˆ4 Allele as a Genetic Risk Factor for Left Ventricular Failure in Homozygous Î²-Thalassemia. Blood, 1998, 92, 3455-3459.	0.6	3
105	A large family with subtelomeric translocation t(18;21)(q23;q22.1) and molecular breakpoint in the Down syndrome critical region. Human Genetics, 1997, 100, 669-675.	1.8	17
106	A patient with Edwards syndrome caused by a rare pseudodicentric chromosome 18 of paternal origin. Clinical Genetics, 1997, 52, 56-60.	1.0	10
107	Apolipoprotein E polymorphism in the Greek population. Clinical Genetics, 1997, 52, 216-218.	1.0	44
108	Investigation of deletions at 7q11.23 in 44 patients referred for Williams-Beuren syndrome, using FISH and four DNA polymorphisms. Human Genetics, 1996, 99, 56-61.	1.8	25

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109	Susceptible chiasmate configurations of chromosome 21 predispose to nonâ€“disjunction in both maternal meiosis I and meiosis II. Nature Genetics, 1996, 14, 400-405.	9.4	362
110	Tetrasomy 18p de novo: Parental Origin and Different Mechanisms of Formation. European Journal of Human Genetics, 1996, 4, 160-167.	1.4	35
111	Clinical, cytogenetic, and molecular diagnosis of Angelman syndrome: Estimated prevalence rate in a Danish county. American Journal of Medical Genetics Part A, 1995, 60, 261-262.	2.4	121
112	Non-disjunction of chromosome 21 in maternal meiosis I: evidence for a maternal age-dependent mechanism involving reduced recombination. Human Molecular Genetics, 1994, 3, 1529-1535.	1.4	165
113	Monozygotic twins discordant for gastroschisis: Case report and review of the literature of twins and familial occurrence of gastroschisis. American Journal of Medical Genetics Part A, 1994, 52, 223-226.	2.4	30
114	Unbalanced translocation, t(18;21), detected by fluorescence in situ hybridization (FISH) in a child with 18q- syndrome and a ring chromosome 21. American Journal of Medical Genetics Part A, 1993, 46, 647-651.	2.4	3
115	A Linkage Map of Human Chromosome 21: 43 PCR Markers at Average Intervals of 2.5 cM. Genomics, 1993, 16, 562-571.	1.3	73
116	Uniparental isodisomy due to duplication of chromosome 21 occuring in somatic cells monosomic for chromosome 21. Genomics, 1992, 13, 269-274.	1.3	64
117	D21S215 is a (GT)n polymorphic marker close to centromeric alphoid sequences on chromosome 21. Genomics, 1992, 13, 1365-1367.	1.3	14
118	Dinucleotide repeat (GT)n markers on chromosome 21. Genomics, 1992, 14, 818-819.	1.3	13
119	A genetic linkage map of 27 markers on human chromosome 21. Genomics, 1991, 9, 407-419.	1.3	82
120	Linkage mapping of highly informative DNA polymorphisms within the human interferon-Î± receptor gene on chromosome 21. Genomics, 1991, 11, 573-576.	1.3	35
121	A 48,XXY,+21 Down syndrome patient with additional paternal X and maternal 21. Human Genetics, 1991, 87, 54-56.	1.8	25
122	PvuII and XhoI/EcoRV polymorphisms adjacent to the Î±- A-crystallin (CRYA1) gene on human chromosome 21. Nucleic Acids Research, 1990, 18, 4300-4300.	6.5	1
123	Linkage analysis of the human HMG14 gene on chromosome 21 using a GT dinucleotide repeat as polymorphic marker. Genomics, 1990, 7, 136-138.	1.3	60
124	Linkage mapping of the highly informative DNA marker D21S156 to human chromosome 21 using a polymorphic GT dinucleotide repeat. Genomics, 1990, 8, 400-402.	1.3	28
125	Two PstI DNA polymorphisms adjacent to the human gene for the interferon-induced p78 protein (MX1) Tj ETQq1 1 0.784314 rgBT /O	6.5	3
126	Molecular genetic approach to the characterization of the ?Down syndrome region? of chromosome 21. Genomics, 1989, 5, 325-331.	1.3	205

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127	Rare chromosome 20 variants encountered during prenatal diagnosis. Prenatal Diagnosis, 1986, 6, 363-367.	1.1	12