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	HDAC8 mutations in Cornelia de Lange syndrome affect the cohesin acetylation cycle. Nature, 2012, 489, 313-317.	13.7	488
2	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
3	Molecular genetic approach to the characterization of the ?Down syndrome region? of chromosome 21. Genomics, 1989, 5, 325-331.	1.3	205
4	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
5	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
6	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
7	Prevalence and patient characteristics of Mayerâ€“Rokitanskyâ€“KÃ¼sterâ€“Hauser syndrome: a nationwide registry-based study. Human Reproduction, 2016, 31, 2384-2390.	0.4	107
8	A genetic linkage map of 27 markers on human chromosome 21. Genomics, 1991, 9, 407-419.	1.3	82
9	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
10	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
11	The spectrum of intermediate <scp>SCN</scp>8Aâ€related epilepsy. Epilepsia, 2019, 60, 830-844.	2.6	70
12	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
13	Uniparental isodisomy due to duplication of chromosome 21 occuring in somatic cells monosomic for chromosome 21. Genomics, 1992, 13, 269-274.	1.3	64
14	Prevalence of GJB2 mutations in prelingual deafness in the Greek population. International Journal of Pediatric Otorhinolaryngology, 2002, 65, 101-108.	0.4	63
15	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
16	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
17	Origin of nondisjunction in trisomy 8 and trisomy 8 mosaicism. European Journal of Human Genetics, 1998, 6, 432-438.	1.4	59
18	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

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19	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
20	A seventh locus for otosclerosis, OTSC7, maps to chromosome 6q13â€“16.1. European Journal of Human Genetics, 2007, 15, 362-368.	1.4	53
21	SLITRK6 mutations cause myopia and deafness in humans and mice. Journal of Clinical Investigation, 2013, 123, 2094-2102.	3.9	50
22	Patterns of Meiotic Recombination on the Long Arm of Human Chromosome 21. Genome Research, 2000, 10, 1319-1332.	2.4	47
23	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
24	Different mechanisms preclude mutant CLDN14 proteins from forming tight junctions in vitro. Human Mutation, 2005, 25, 543-549.	1.1	45
25	Pathologic, radiographic and molecular findings in three fetuses diagnosed with HEM/Greenberg skeletal dysplasia. Prenatal Diagnosis, 2008, 28, 309-312.	1.1	45
26	Apolipoprotein E polymorphism in the Greek population. Clinical Genetics, 1997, 52, 216-218.	1.0	44
27	Oxidative stress in dry age-related macular degeneration and exfoliation syndrome. Critical Reviews in Clinical Laboratory Sciences, 2015, 52, 12-27.	2.7	44
28	Age-related macular degeneration: genetic and clinical findings. Clinical Chemistry and Laboratory Medicine, 2011, 49, 601-616.	1.4	43
29	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
30	Detection of mutations in theCOL4A5gene by SSCP in X-linked Alport syndrome. Human Mutation, 2001, 18, 141-148.	1.1	38
31	Non-disjunction of chromosome 13. Human Molecular Genetics, 2007, 16, 2004-2010.	1.4	38
32	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
33	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
34	Apolipoprotein E and presenilin-1 genotypes in Huntington's disease. Journal of Neurology, 1999, 246, 574-577.	1.8	35
35	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
36	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

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37	Tetrasomy 18p de novo: Parental Origin and Different Mechanisms of Formation. European Journal of Human Genetics, 1996, 4, 160-167.	1.4	35
38	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
39	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
40	Prenatal diagnosis of glycogen storage disease type IV. Prenatal Diagnosis, 2006, 26, 951-955.	1.1	32
41	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
42	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
43	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
44	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
45	ECEL1 mutation causes fetal arthrogryposis multiplex congenita. American Journal of Medical Genetics, Part A, 2015, 167, 731-743.	0.7	28
46	Partial trisomy 21 map: Ten cases further supporting the highly restricted Down syndrome critical region (HRâ€ÐSCR) on human chromosome 21. Molecular Genetics & Genomic Medicine, 2019, 7, e797.	0.6	28
47	BRCA1 mutation analysis in breast/ovarian cancer families from Greece. Human Mutation, 2000, 16, 272-273.	1.1	27
48	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
49	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
50	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
51	Genotypeâ€phenotype correlations, dystonia and disease progression in spinocerebellar ataxia type 14. Movement Disorders, 2018, 33, 1119-1129.	2.2	26
52	A 48,XXY,+21 Down syndrome patient with additional paternal X and maternal 21. Human Genetics, 1991, 87, 54-56.	1.8	25
53	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
54	FRAXA and FRAXE prevalence in patients with nonspecific mental retardation in the Hellenic population. Genetic Epidemiology, 1998, 15, 103-109.	0.6	25

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55	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
56	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
57	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
58	Unexpected results in the constitution of small supernumerary marker chromosomes. European Journal of Medical Genetics, 2012, 55, 185-190.	0.7	22
59	Molecular cytogenetic characterization and origin of two de novo duplication 9p cases. , 2000, 91, 102-106.		21
60	The use of array-CGH in a cohort of Greek children with developmental delay. Molecular Cytogenetics, 2010, 3, 22.	0.4	20
61	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
62	Biomarkers in primary open angle glaucoma. Clinical Chemistry and Laboratory Medicine, 2012, 50, 2107-2119.	1.4	19
63	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
64	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
65	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
66	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
67	A LargeGLC1CGreek Family with a Myocilin T377M Mutation: Inheritance and Phenotypic Variability. , 2006, 47, 620.		17
68	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
69	First reported adult patient with TARP syndrome: A case report. American Journal of Medical Genetics, Part A, 2018, 176, 2915-2918.	0.7	16
70	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
71	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
72	D21S215 is a (GT)n polymorphic marker close to centromeric alphoid sequences on chromosome 21. Genomics, 1992, 13, 1365-1367.	1.3	14

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73	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
74	Dinucleotide repeat (GT)n markers on chromosome 21. Genomics, 1992, 14, 818-819.	1.3	13
75	Human CCS gene: genomic organization and exclusion as a candidate for amyotrophic lateral sclerosis (ALS). BMC Genetics, 2002, 3, 5.	2.7	13
76	Prenatal diagnosis of achondroplasia presenting with multiple-suture synostosis: a novel association. Prenatal Diagnosis, 2006, 26, 258-261.	1.1	13
77	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
78	Rare chromosome 20 variants encountered during prenatal diagnosis. Prenatal Diagnosis, 1986, 6, 363-367.	1.1	12
79	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
80	Hypothesizing an Ancient Greek Origin of theGJB235delG Mutation: Can Science Meet History?. Genetic Testing and Molecular Biomarkers, 2010, 14, 183-187.	0.3	12
81	Detection of Deafness-Causing Mutations in the Greek Mitochondrial Genome. Disease Markers, 2011, 30, 283-289.	0.6	12
82	Prenatal Diagnosis of Trisomy 2 Mosaicism: A Case Report. Fetal Diagnosis and Therapy, 2004, 19, 488-490.	0.6	10
83	Monogenic nonsyndromic otosclerosis: Audiological and linkage analysis in a large Greek pedigree. International Journal of Pediatric Otorhinolaryngology, 2006, 70, 631-637.	0.4	10
84	A patient with Edwards syndrome caused by a rare pseudodicentric chromosome 18 of paternal origin. Clinical Genetics, 1997, 52, 56-60.	1.0	10
85	Sudden hearing loss in a family with GJB2 related progressive deafness. International Journal of Pediatric Otorhinolaryngology, 2008, 72, 1735-1740.	0.4	10
86	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
87	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
88	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
89	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
90	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

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91	Genotyping of presenilin-1 polymorphism in amyotrophic lateral sclerosis. Journal of Neurology, 2000, 247, 940-942.	1.8	8
92	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
93	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
94	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
95	DNA studies of mono- and pseudodicentric isochromosomes 18q. American Journal of Medical Genetics Part A, 2004, 127A, 230-233.	2.4	7
96	Prenatal diagnosis of a fetus with ring chromosome 15 characterized by arrayâ€CGH. Prenatal Diagnosis, 2009, 29, 884-888.	1.1	7
97	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
98	Mitochondrial Disease Caused by a Novel Homozygous Mutation (Gly106del) in the SCO1 Gene. Neonatology, 2019, 116, 290-294.	0.9	7
99	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
100	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
101	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
102	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
103	Ocular albinism with infertility and lateâ€onset sensorineural hearing loss. American Journal of Medical Genetics, Part A, 2018, 176, 1587-1593.	0.7	6
104	First reported CABP2â€related nonâ€syndromic hearing loss in Northern Europe. Molecular Genetics & Genomic Medicine, 2021, 9, e1639.	0.6	6
105	A wide range of protective and predisposing variants in aggrecan influence the susceptibility for otosclerosis. Human Genetics, 2022, 141, 951-963.	1.8	6
106	3C syndrome with cryptorchidism and posterior embryotoxon. Clinical Dysmorphology, 2005, 14, 97-100.	0.1	5
107	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
108	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

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109	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
110	Copy number variation and genomic alterations in health and disease. Genome Medicine, 2009, 1, 21.	3.6	4
111	Screening of a Greek deafness population for the A7445G mitochondrial DNA mutation. Molecular Genetics and Metabolism, 2010, 100, 300-301.	0.5	4
112	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
113	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
114	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
115	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
116	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
117	First reported case of Doyne honeycomb retinal dystrophy (Malattia Leventinese/autosomal dominant) Tj ETQq1 1 0.784314 rgBT /O	0.6	3
118	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
119	Apolipoprotein E alleles in mothers of trisomy 18 conceptuses. Clinical Genetics, 1998, 53, 321-322.	1.0	2
120	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
121	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
122	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
123	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
124	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
125	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
126	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

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127	Re-Examination of Danish Carriers of Balanced Chromosomal Inversions. Cancer Genetics, 2016, 209, 231.	0.2	0