Jean-Michel Rozet

List of Publications by Year
in descending order

116 papers	6,844 citations	71102 41 h-index	62596 80 g-index
125 all docs	125 docs citations	125 times ranked	6283 citing authors

#	Article	IF	Citations
1	Mutations in the gene encoding fibroblast growth factor receptor-3 in achondroplasia. Nature, 1994, 371, 252-254.	27.8	873
2	Retinalâ€“specific guanylate cyclase gene mutations in Leber's congenital amaurosis. Nature Genetics, 1996, 14, 461-464.	21.4	433
3	Xâ€“linked spastic paraplegia and Pelizaeusâ€“Merzbacher disease are allelic disorders at the proteolipid protein locus. Nature Genetics, 1994, 6, 257-262.	21.4	353
4	Leber congenital amaurosis: Comprehensive survey of the genetic heterogeneity, refinement of the clinical definition, and genotype-phenotype correlations as a strategy for molecular diagnosis. Human Mutation, 2004, 23, 306-317.	2.5	313
5	Spectrum of NPHP6/CEP290 mutations in Leber congenital amaurosis and delineation of the associated phenotype. Human Mutation, 2007, 28, 416-416.	2.5	224
6	Mainzer-Saldino Syndrome Is a Ciliopathy Caused by IFT140 Mutations. American Journal of Human Genetics, 2012, 90, 864-870.	6.2	173
7	A gene for Stargardt's disease (fundus flavimaculatus) maps to the short arm of chromosome 1. Nature Genetics, 1993, 5, 308-311.	21.4	169
8	Retinal Dehydrogenase 12 (RDH12) Mutations in Leber Congenital Amaurosis. American Journal of Human Genetics, 2004, 75, 639-646.	6.2	164
9	Spectrum of ABCR gene mutations in autosomal recessive macular dystrophies. European Journal of Human Genetics, 1998, 6, 291-295.	2.8	152
10	Complete exon-intron structure of the RPGR-interacting protein (RPGRIP1) gene allows the identification of mutations underlying Leber congenital amaurosis. European Journal of Human Genetics, 2001, 9, 561-571.	2.8	148
11	Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy. Brain, 2012, 135, 2980-2993.	7.6	148
12	A gene for usher syndrome type I (USH1A) maps to chromosome 14q. Genomics, 1992, 14, 979-987.	2.9	141
13	Leber Congenital Amaurosis. Molecular Genetics and Metabolism, 1999, 68, 200-208.	1.1	140
14	Mutations in NMNAT1 cause Leber congenital amaurosis with early-onset severe macular and optic atrophy. Nature Genetics, 2012, 44, 975-977.	21.4	123
15	Comprehensive survey of mutations in RP2 and RPGR in patients affected with distinct retinal dystrophies: genotypeâ€“phenotype correlations and impact on genetic counseling. Human Mutation, 2007, 28, 81-91.	2.5	114
16	Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome. American Journal of Human Genetics, 2016, 98, 971-980.	6.2	113
17	Different Functional Outcome of RetGC1 and RPE65 Gene Mutations in Leber Congenital Amaurosis. American Journal of Human Genetics, 1999, 64, 1225-1228.	6.2	112
18	Spectrum of retGC1 mutations in Leber's congenital amaurosis. European Journal of Human Genetics, 2000, 8, 578-582.	2.8	108

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19	Mutations in DNM1L, as in OPA1, result in dominant optic atrophy despite opposite effects on mitochondrial fusion and fission. Brain, 2017, 140, 2586-2596.	7.6	100
20	AON-mediated Exon Skipping Restores Ciliation in Fibroblasts Harboring the Common Leber Congenital Amaurosis CEP290 Mutation. Molecular Therapy - Nucleic Acids, 2012, 1, e29.	5.1	94
21	ALDH1A3 Mutations Cause Recessive Anophthalmia and Microphthalmia. American Journal of Human Genetics, 2013, 92, 265-270.	6.2	92
22	A retGC-1 Mutation in Autosomal Dominant Cone-Rod Dystrophy. American Journal of Human Genetics, 1998, 63, 651-654.	6.2	89
23	A Gene for X-Linked Idiopathic Congenital Nystagmus (NYS1) Maps to Chromosome Xp11.4-p11.3. American Journal of Human Genetics, 1999, 64, 1141-1146.	6.2	89
24	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	8.2	89
25	The photoreceptor cell-specific nuclear receptor gene (PNR) accounts for retinitis pigmentosa in the Crypto-Jews from Portugal (Marranos), survivors from the Spanish Inquisition. Human Genetics, 2000, 107, 276-284.	3.8	86
26	TMEM126A, Encoding a Mitochondrial Protein, Is Mutated in Autosomal-Recessive Nonsyndromic Optic Atrophy. American Journal of Human Genetics, 2009, 84, 493-498.	6.2	85
27	Mutations in the tricarboxylic acid cycle enzyme, aconitase 2, cause either isolated or syndromic optic neuropathy with encephalopathy and cerebellar atrophy. Journal of Medical Genetics, 2014, 51, 834-838.	3.2	80
28	Complete Exonâ€“Intron Structure of the Retina-Specific ATP Binding Transporter Gene (ABCR) Allows the Identification of Novel Mutations Underlying Stargardt Disease. Genomics, 1998, 48, 139-142.	2.9	75
29	Dominant X linked retinitis pigmentosa is frequently accounted for by truncating mutations in exon ORF15 of the RPGR gene. Journal of Medical Genetics, 2002, 39, 284-285.	3.2	68
30	A third locus for dominant optic atrophy on chromosome 22q. Journal of Medical Genetics, 2005, 42, e1-e1.	3.2	63
31	A gene for Leber's congenital amaurosis maps to chromosome 17p. Human Molecular Genetics, 1995, 4, 1447-1452.	2.9	56
32	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. American Journal of Human Genetics, 2015, 97, 754-760.	6.2	54
33	rs5888 Variant of SCARB1 Gene Is a Possible Susceptibility Factor for Age-Related Macular Degeneration. PLoS ONE, 2009, 4, e7341.	2.5	53
34	Mutations of the Fibroblast Growth Factor Receptor-3 Gene in Achondroplasia. Hormone Research, 1996, 45, 108-110.	1.8	47
35	A first locus for isolated autosomal recessive optic atrophy (ROA1) maps to chromosome 8q. European Journal of Human Genetics, 2003, 11, 966-971.	2.8	47
36	Abnormal respiratory cilia in non-syndromic Leber congenital amaurosis with CEP290 mutations. Journal of Medical Genetics, 2010, 47, 829-834.	3.2	47

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37	Prenatal human ocular degeneration occurs in Leber's congenital amaurosis (LCA2). Journal of Gene Medicine, 2002, 4, 390-396.	2.8	46
38	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. European Journal of Human Genetics, 2002, 10, 197-203.	2.8	45
39	Relative Frequencies of Inherited Retinal Dystrophies and Optic Neuropathies in Southern France: Assessment of 21-year Data Management. Ophthalmic Epidemiology, 2013, 20, 13-25.	1.7	44
40	Mutations in DOCK7 in Individuals with Epileptic Encephalopathy and Cortical Blindness. American Journal of Human Genetics, 2014, 94, 891-897.	6.2	44
41	Age-related macular degeneration in grandparents of patients with Stargardt disease: genetic study. American Journal of Ophthalmology, 1999, 128, 173-178.	3.3	43
42	Compound heterozygosity for severe and hypomorphicNDUFS2mutations cause non-syndromic LHON-like optic neuropathy. Journal of Medical Genetics, 2017, 54, 346-356.	3.2	43
43	X linked spastic paraplegia (SPG2): clinical heterogeneity at a single gene locus.. Journal of Medical Genetics, 1993, 30, 381-384.	3.2	42
44	Genetic Heterogeneity of Usher Syndrome Type 1 in French Families. Genomics, 1994, 21, 138-143.	2.9	42
45	Evidence of autosomal dominant Leber congenital amaurosis (LCA) underlain by a CRX heterozygous null allele. Journal of Medical Genetics, 2003, 40, 90e-90.	3.2	41
46	USH1A: Chronicle of a Slow Death. American Journal of Human Genetics, 2006, 78, 357-359.	6.2	41
47	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
48	The ABCA4 Gene in Autosomal Recessive Cone-Rod Dystrophies. American Journal of Human Genetics, 2002, 71, 1480-1482.	6.2	36
49	Severe manifestations in carrier females in X linked retinitis pigmentosa.. Journal of Medical Genetics, 1997, 34, 793-797.	3.2	34
50	No evidence of genetic heterogeneity in dominant optic atrophy.. Journal of Medical Genetics, 1995, 32, 951-953.	3.2	33
51	NDP gene mutations in 14 French families with Norrie disease. Human Mutation, 2003, 22, 499-499.	2.5	33
52	Nonsyndromic Retinal Dystrophy due to Bi-Allelic Mutations in the Ciliary Transport Gene IFT140. , 2016, 57, 1053.		33
53	The ABCR Gene: A Major Disease Gene in Macular and Peripheral Retinal Degenerations with Onset from Early Childhood to the Elderly. Molecular Genetics and Metabolism, 1999, 68, 310-315.	1.1	32
54	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665.	3.2	32

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55	Basal exon skipping and nonsense-associated altered splicing allows bypassing complete CEP290 loss-of-function in individuals with unusually mild retinal disease. Human Molecular Genetics, 2018, 27, 2689-2702.	2.9	31
56	Mutations in TUBB4B Cause a Distinctive Sensorineural Disease. American Journal of Human Genetics, 2017, 101, 1006-1012.	6.2	30
57	A novel mutation disrupting the cytoplasmic domain of CRB1 in a large consanguineous family of Palestinian origin affected with Leber congenital amaurosis. Ophthalmic Genetics, 2002, 23, 225-235.	1.2	29
58	Multimodal analysis of the progression of Best vitelliform macular dystrophy. Molecular Vision, 2014, 20, 575-92.	1.1	29
59	Evidence of a founder effect for the RETGC1 (GUCY2D) 2943DelG mutation in Leber congenital amaurosis pedigrees of Finnish origin. Human Mutation, 2002, 20, 322-323.	2.5	28
60	The Spectrum of Subclinical Best Vitelliform Macular Dystrophy in Subjects with Mutations inBEST1Gene. , 2011, 52, 4678.		28
61	Intravitreal Injection of Splice-switching Oligonucleotides to Manipulate Splicing in Retinal Cells. Molecular Therapy - Nucleic Acids, 2015, 4, e250.	5.1	28
62	Human Retinal Guanylate Cyclase (GUC2D) Maps to Chromosome 17p13.1. Genomics, 1994, 22, 478-481.	2.9	26
63	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
64	A novel mutation in the GUCY2D gene responsible for an early onset severe RP different from the usual GUCY2Dâ€ŁCA phenotype. Human Mutation, 2005, 25, 222-222.	2.5	24
65	Spectrum ofSPATA7mutations in Leber congenital amaurosis and delineation of the associated phenotype. Human Mutation, 2010, 31, E1241-E1250.	2.5	24
66	TMEM126A is a mitochondrial located mRNA (MLR) protein of the mitochondrial inner membrane. Biochimica Et Biophysica Acta - General Subjects, 2013, 1830, 3719-3733.	2.4	23
67	Mutations in LCA5 are an uncommon cause of Leber congenital amaurosis (LCA) type II. Human Mutation, 2007, 28, 1245-1245.	2.5	22
68	Loss of Function of RIMS2 Causes a Syndromic Congenital Cone-Rod Synaptic Disease with Neurodevelopmental and Pancreatic Involvement. American Journal of Human Genetics, 2020, 106, 859-871.	6.2	22
69	Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in IFT140. Human Mutation, 2018, 39, 983-992.	2.5	21
70	MCAT Mutations Cause Nuclear LHON-like Optic Neuropathy. Genes, 2021, 12, 521.	2.4	21
71	Structure and refinement of the physical mapping of the Î³-glutamylcysteine ligase regulatory subunit (GLCLR) gene to chromosome 1p22.1 within the critically deleted region of human malignant mesothelioma. Cytogenetic and Genome Research, 1998, 82, 91-94.	1.1	20
72	Antisense Oligonucleotide Therapy for Inherited Retinal Dystrophies. Advances in Experimental Medicine and Biology, 2016, 854, 517-524.	1.6	20

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73	Five novel missense mutations of the rhodopsin gene in autosomal dominant retinitis pigmentosa. Human Molecular Genetics, 1994, 3, 1433-1434.	2.9	19
74	R102G polymorphism of the C3 gene associated with exudative age-related macular degeneration in a French population. Molecular Vision, 2010, 16, 1324-30.	1.1	19
75	Structure and Physical Mapping of DR1, a TATA-Binding Protein-Associated Phosphoprotein Gene, to Chromosome 1p22.1 and Its Exclusion in Stargardt Disease (STGD). Genomics, 1996, 36, 554-556.	2.9	16
76	Partial duplication [dup. TCAC (178)] and novel point mutations (T125M, G188R, A209V, and H302L) of the ornithine transcarbamylase gene in congenital hyperammonemia. , 1996, 8, 74-76.		16
77	DPAGT1 Deficiency with Encephalopathy (DPAGT1-CDG): Clinical and Genetic Description of 11 New Patients. JIMD Reports, 2018, 44, 85-92.	1.5	16
78	Union Makes Strength: A Worldwide Collaborative Genetic and Clinical Study to Provide a Comprehensive Survey of RD3 Mutations and Delineate the Associated Phenotype. PLoS ONE, 2013, 8, e51622.	2.5	16
79	Two Novel Missense Mutations in the Peripherin/RDS Gene in two Unrelated French Patients with Autosomal Dominant Retinitis Pigmentosa. European Journal of Ophthalmology, 1998, 8, 98-101.	1.3	15
80	AON-Mediated Exon Skipping to Bypass Protein Truncation in Retinal Dystrophies Due to the Recurrent CEP290 c.4723A > T Mutation. Fact or Fiction?. Genes, 2019, 10, 368.	2.4	15
81	RETINOCHOROIDAL ANASTOMOSIS ASSOCIATED WITH ENHANCED S-CONE SYNDROME. Retinal Cases and Brief Reports, 2019, 13, 295-299.	0.6	14
82	Genetic architecture of retinoic-acid signaling-associated ocular developmental defects. Human Genetics, 2019, 138, 937-955.	3.8	14
83	Leber Congenital Amaurosis: Survey of the Genetic Heterogeneity, Refinement of the Clinical Definition and Phenotype-Genotype Correlations as a Strategy for Molecular Diagnosis. , 2006, 572, 15-20.		14
84	Three different ABCA4 mutations in the same large family with several consanguineous loops affected with autosomal recessive coneâ€“rod dystrophy. European Journal of Human Genetics, 2006, 14, 1269-1273.	2.8	13
85	Submicroscopic Deletions at 13q32.1 Cause Congenital Microcoria. American Journal of Human Genetics, 2015, 96, 631-639.	6.2	13
86	Incomplete penetrance of biallelic ALDH1A3 mutations. European Journal of Medical Genetics, 2016, 59, 215-218.	1.3	13
87	Population history and infrequent mutations: how old is a rare mutation? GUCY2D as a worked example. European Journal of Human Genetics, 2008, 16, 115-123.	2.8	12
88	No association between the T280M polymorphism of the CX3CR1 gene and exudative AMD. Experimental Eye Research, 2011, 93, 382-386.	2.6	12
89	Novel intragenic deletions and point mutations of the ornithine transcarbamylase gene in congenital hyperammonemia. Human Mutation, 1998, 11, S81-S84.	2.5	10
90	Prenatal Human Ocular Degeneration Occurs in Leberâ€™s Congenital Amaurosis. Advances in Experimental Medicine and Biology, 2003, , 59-68.	1.6	10

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91	Understanding disease pleiotropy: From puzzle to solution. Science Translational Medicine, 2015, 7, 291fs24.	12.4	9
92	Disease-Associated Variants of the Rod-derived Cone Viability Factor (RdCVF) in Leber Congenital Amaurosis. , 2006, 572, 9-14.		8
93	Congenital Microcoria: Clinical Features and Molecular Genetics. Genes, 2021, 12, 624.	2.4	6
94	Expanding the <scp>KIF4A</scp>â€associated phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3728-3739.	1.2	6
95	Genetic association study of mitochondrial polymorphisms in neovascular age-related macular degeneration. Molecular Vision, 2013, 19, 1132-40.	1.1	6
96	Physical and Genetic Localization of the Î³ Subunit of the Cyclic GMP Phosphodiesterase on the Long Arm of Chromosome 17 (17q25). Genomics, 1993, 17, 526-528.	2.9	5
97	Intellectual disability associated with retinal dystrophy in the Xp11.3 deletion syndrome: ZNF674 on trial. Guilty or innocent?. European Journal of Human Genetics, 2012, 20, 352-356.	2.8	5
98	Reply: The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e29-e29.	7.6	5
99	Machado-Joseph Disease Is Genetically Different from Holguin Dominant Ataxia (SCA2). Genomics, 1993, 17, 556-559.	2.9	4
100	Dinucleotide repeat polymorphism at the human recoverin RCVI gene locus on chromosome 17p. Human Molecular Genetics, 1993, 2, 1081-1081.	2.9	4
101	Exclusion of the cone-specific ?-subunit of the transducin gene in Stargardt's disease. Human Genetics, 1995, 95, 382-4.	3.8	4
102	Evidence for a fourth locus in Usher syndrome type I.. Journal of Medical Genetics, 1996, 33, 77-79.	3.2	4
103	Exclusion of five subunits of cGMP phosphodiesterase in Leber's congenital amaurosis. Human Genetics, 1998, 102, 322-326.	3.8	4
104	GENETICS OF LARGE PIGMENT EPITHELIAL DETACHMENTS IN NEOVASCULAR AGE-RELATED MACULAR DEGENERATION. Retina, 2020, 40, 663-671.	1.7	4
105	Dinucleotide repeat polymorphism at the human protein C inhibitor (PCI) locus. Human Molecular Genetics, 1993, 2, 2201-2201.	2.9	3
106	Dsal polymorphism at the human cone transducin Î±-subunit (GNAT2) detected by PCR. Human Molecular Genetics, 1994, 3, 1030-1030.	2.9	3
107	Apparent segregation of null alleles ascribed to deletions of the ornithine transcarbamylase gene in congenital hyperammonaemia. Prenatal Diagnosis, 1995, 15, 757-761.	2.3	3
108	Whole Locus Sequencing Identifies a Prevalent Founder Deep Intronic RPGRIP1 Pathologic Variant in the French Leber Congenital Amaurosis Cohort. Genes, 2021, 12, 287.	2.4	3

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109	Description of Two Siblings with Apparently Severe CEP290 Mutations and Unusually Mild Retinal Disease Unrelated to Basal Exon Skipping or Nonsense-Associated Altered Splicing. Advances in Experimental Medicine and Biology, 2019, 1185, 189-195.	1.6	3
110	Genetic Deciphering of Early-Onset and Severe Retinal Dystrophy Associated with Sensorineural Hearing Loss. Advances in Experimental Medicine and Biology, 2019, 1185, 233-238.	1.6	3
111	Exclusion of the apoE gene in autosomal dominant retinitis pigmentosa. Vision Research, 1998, 38, 3829-3831.	1.4	1
112	A First Locus for Isolated Autosomal Recessive Optic Atrophy (ROA1) Maps to Chromosome 8q21-q22. , 2006, 572, 21-27.		1
113	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. , 0, .		1
114	Leber Congenital Amaurosis â€" Genotyping Required for Possible Inclusion in a Clinical Trial. Advances in Experimental Medicine and Biology, 2003, 533, 69-77.	1.6	1
115	ISDN2014_0400: Mutations in DOCK7 in individuals with epileptic encephalopathy and cortical blindness. International Journal of Developmental Neuroscience, 2015, 47, 119-120.	1.6	0
116	Identification des bases molÃ©culaires des dystrophies rÃ©tiniennes hÃ©rÃ©ditaires : dÃ©couverte de vÃ©ritÃ©s contraires et consÃ©quences. Bulletin De L'Academie Nationale De Medecine, 2009, 193, 163-178.	0.0	0