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	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
2	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	8.2	89
3	Congenital Microcoria: Clinical Features and Molecular Genetics. Genes, 2021, 12, 624.	2.4	6
4	MCAT Mutations Cause Nuclear LHON-like Optic Neuropathy. Genes, 2021, 12, 521.	2.4	21
5	Expanding the <scp>KIF4A</scp>â€associated phenotype. American Journal of Medical Genetics, Part A, 2021, 185, 3728-3739.	1.2	6
6	GENETICS OF LARGE PIGMENT EPITHELIAL DETACHMENTS IN NEOVASCULAR AGE-RELATED MACULAR DEGENERATION. Retina, 2020, 40, 663-671.	1.7	4
7	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
8	RETINOCHOROIDAL ANASTOMOSIS ASSOCIATED WITH ENHANCED S-CONE SYNDROME. Retinal Cases and Brief Reports, 2019, 13, 295-299.	0.6	14
9	Genetic architecture of retinoic-acid signaling-associated ocular developmental defects. Human Genetics, 2019, 138, 937-955.	3.8	14
10	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
11	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
12	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
13	Whole-genome sequencing in patients with ciliopathies uncovers a novel recurrent tandem duplication in IFT140. Human Mutation, 2018, 39, 983-992.	2.5	21
14	Reply: The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e29-e29.	7.6	5
15	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	9.0	26
16	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
17	DPAGT1 Deficiency with Encephalopathy (DPAGT1-CDG): Clinical and Genetic Description of 11 New Patients. JIMD Reports, 2018, 44, 85-92.	1.5	16
18	Compound heterozygosity for severe and hypomorphicNDUFS2mutations cause non-syndromic LHON-like optic neuropathy. Journal of Medical Genetics, 2017, 54, 346-356.	3.2	43

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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	Mutations in TUBB4B Cause a Distinctive Sensorineural Disease. American Journal of Human Genetics, 2017, 101, 1006-1012.	6.2	30
21	Nonsyndromic Retinal Dystrophy due to Bi-Allelic Mutations in the Ciliary Transport Gene IFT140. , 2016, 57, 1053.		33
22	Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome. American Journal of Human Genetics, 2016, 98, 971-980.	6.2	113
23	Incomplete penetrance of biallelic ALDH1A3 mutations. European Journal of Medical Genetics, 2016, 59, 215-218.	1.3	13
24	Antisense Oligonucleotide Therapy for Inherited Retinal Dystrophies. Advances in Experimental Medicine and Biology, 2016, 854, 517-524.	1.6	20
25	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
26	Understanding disease pleiotropy: From puzzle to solution. Science Translational Medicine, 2015, 7, 291fs24.	12.4	9
27	Submicroscopic Deletions at 13q32.1 Cause Congenital Microcoria. American Journal of Human Genetics, 2015, 96, 631-639.	6.2	13
28	Intravitreal Injection of Splice-switching Oligonucleotides to Manipulate Splicing in Retinal Cells. Molecular Therapy - Nucleic Acids, 2015, 4, e250.	5.1	28
29	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. American Journal of Human Genetics, 2015, 97, 754-760.	6.2	54
30	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
31	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
32	Mutations in DOCK7 in Individuals with Epileptic Encephalopathy and Cortical Blindness. American Journal of Human Genetics, 2014, 94, 891-897.	6.2	44
33	Multimodal analysis of the progression of Best vitelliform macular dystrophy. Molecular Vision, 2014, 20, 575-92.	1.1	29
34	ALDH1A3 Mutations Cause Recessive Anophthalmia and Microphthalmia. American Journal of Human Genetics, 2013, 92, 265-270.	6.2	92
35	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
36	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

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37	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
38	Genetic association study of mitochondrial polymorphisms in neovascular age-related macular degeneration. Molecular Vision, 2013, 19, 1132-40.	1.1	6
39	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
40	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
41	Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy. Brain, 2012, 135, 2980-2993.	7.6	148
42	Mutations in NMNAT1 cause Leber congenital amaurosis with early-onset severe macular and optic atrophy. Nature Genetics, 2012, 44, 975-977.	21.4	123
43	Mainzer-Saldino Syndrome Is a Ciliopathy Caused by IFT140 Mutations. American Journal of Human Genetics, 2012, 90, 864-870.	6.2	173
44	No association between the T280M polymorphism of the CX3CR1 gene and exudative AMD. Experimental Eye Research, 2011, 93, 382-386.	2.6	12
45	The Spectrum of Subclinical Best Vitelliform Macular Dystrophy in Subjects with Mutations inBEST1Gene. , 2011, 52, 4678.		28
46	Spectrum ofSPATA7mutations in Leber congenital amaurosis and delineation of the associated phenotype. Human Mutation, 2010, 31, E1241-E1250.	2.5	24
47	Abnormal respiratory cilia in non-syndromic Leber congenital amaurosis with CEP290 mutations. Journal of Medical Genetics, 2010, 47, 829-834.	3.2	47
48	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
49	rs5888 Variant of SCARB1 Gene Is a Possible Susceptibility Factor for Age-Related Macular Degeneration. PLoS ONE, 2009, 4, e7341.	2.5	53
50	Expanding CEP290 mutational spectrum in ciliopathies. American Journal of Medical Genetics, Part A, 2009, 149A, 2173-2180.	1.2	38
51	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
52	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
53	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
54	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

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55	Mutations in LCA5 are an uncommon cause of Leber congenital amaurosis (LCA) type II. Human Mutation, 2007, 28, 1245-1245.	2.5	22
56	Spectrum of NPHP6/CEP290 mutations in Leber congenital amaurosis and delineation of the associated phenotype. Human Mutation, 2007, 28, 416-416.	2.5	224
57	A First Locus for Isolated Autosomal Recessive Optic Atrophy (ROA1) Maps to Chromosome 8q21-q22. , 2006, 572, 21-27.		1
58	USH1A: Chronicle of a Slow Death. American Journal of Human Genetics, 2006, 78, 357-359.	6.2	41
59	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
60	Disease-Associated Variants of the Rod-derived Cone Viability Factor (RdCVF) in Leber Congenital Amaurosis. , 2006, 572, 9-14.		8
61	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
62	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
63	A third locus for dominant optic atrophy on chromosome 22q. Journal of Medical Genetics, 2005, 42, e1-e1.	3.2	63
64	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
65	Retinal Dehydrogenase 12 (RDH12) Mutations in Leber Congenital Amaurosis. American Journal of Human Genetics, 2004, 75, 639-646.	6.2	164
66	NDP gene mutations in 14 French families with Norrie disease. Human Mutation, 2003, 22, 499-499.	2.5	33
67	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
68	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
69	Prenatal Human Ocular Degeneration Occurs in Leberâ€™s Congenital Amaurosis. Advances in Experimental Medicine and Biology, 2003, , 59-68.	1.6	10
70	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
71	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
72	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

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73	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
74	The ABCA4 Gene in Autosomal Recessive Cone-Rod Dystrophies. American Journal of Human Genetics, 2002, 71, 1480-1482.	6.2	36
75	Prenatal human ocular degeneration occurs in Leber's congenital amaurosis (LCA2). Journal of Gene Medicine, 2002, 4, 390-396.	2.8	46
76	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
77	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
78	Spectrum of retGC1 mutations in Leber's congenital amaurosis. European Journal of Human Genetics, 2000, 8, 578-582.	2.8	108
79	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
80	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
81	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
82	Age-related macular degeneration in grandparents of patients with Stargardt disease: genetic study. American Journal of Ophthalmology, 1999, 128, 173-178.	3.3	43
83	Leber Congenital Amaurosis. Molecular Genetics and Metabolism, 1999, 68, 200-208.	1.1	140
84	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
85	Spectrum of ABCR gene mutations in autosomal recessive macular dystrophies. European Journal of Human Genetics, 1998, 6, 291-295.	2.8	152
86	Novel intragenic deletions and point mutations of the ornithine transcarbamylase gene in congenital hyperammonemia. Human Mutation, 1998, 11, S81-S84.	2.5	10
87	Exclusion of five subunits of cGMP phosphodiesterase in Leber's congenital amaurosis. Human Genetics, 1998, 102, 322-326.	3.8	4
88	Exclusion of the apoE gene in autosomal dominant retinitis pigmentosa. Vision Research, 1998, 38, 3829-3831.	1.4	1
89	A retGC-1 Mutation in Autosomal Dominant Cone-Rod Dystrophy. American Journal of Human Genetics, 1998, 63, 651-654.	6.2	89
90	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

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91	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
92	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
93	Severe manifestations in carrier females in X linked retinitis pigmentosa.. Journal of Medical Genetics, 1997, 34, 793-797.	3.2	34
94	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
95	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
96	Retinalâ€“specific guanylate cyclase gene mutations in Leber's congenital amaurosis. Nature Genetics, 1996, 14, 461-464.	21.4	433
97	Mutations of the Fibroblast Growth Factor Receptor-3 Gene in Achondroplasia. Hormone Research, 1996, 45, 108-110.	1.8	47
98	Evidence for a fourth locus in Usher syndrome type I.. Journal of Medical Genetics, 1996, 33, 77-79.	3.2	4
99	Exclusion of the cone-specific ?-subunit of the transducin gene in Stargardt's disease. Human Genetics, 1995, 95, 382-4.	3.8	4
100	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
101	A gene for Leber's congenital amaurosis maps to chromosome 17p. Human Molecular Genetics, 1995, 4, 1447-1452.	2.9	56
102	No evidence of genetic heterogeneity in dominant optic atrophy.. Journal of Medical Genetics, 1995, 32, 951-953.	3.2	33
103	Dsal polymorphism at the human cone transducin Î±-subunit (GNAT2) detected by PCR. Human Molecular Genetics, 1994, 3, 1030-1030.	2.9	3
104	Five novel missense mutations of the rhodopsin gene in autosomal dominant retinitis pigmentosa. Human Molecular Genetics, 1994, 3, 1433-1434.	2.9	19
105	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
106	Mutations in the gene encoding fibroblast growth factor receptor-3 in achondroplasia. Nature, 1994, 371, 252-254.	27.8	873
107	Genetic Heterogeneity of Usher Syndrome Type 1 in French Families. Genomics, 1994, 21, 138-143.	2.9	42
108	Human Retinal Guanylate Cyclase (GUC2D) Maps to Chromosome 17p13.1. Genomics, 1994, 22, 478-481.	2.9	26

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109	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
110	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
111	Machado-Joseph Disease Is Genetically Different from Holguin Dominant Ataxia (SCA2). Genomics, 1993, 17, 556-559.	2.9	4
112	X linked spastic paraplegia (SPG2): clinical heterogeneity at a single gene locus.. Journal of Medical Genetics, 1993, 30, 381-384.	3.2	42
113	Dinucleotide repeat polymorphism at the human recoverin RCVI gene locus on chromosome 17p. Human Molecular Genetics, 1993, 2, 1081-1081.	2.9	4
114	Dinucleotide repeat polymorphism at the human protein C inhibitor (PCI) locus. Human Molecular Genetics, 1993, 2, 2201-2201.	2.9	3
115	A gene for usher syndrome type I (USH1A) maps to chromosome 14q. Genomics, 1992, 14, 979-987.	2.9	141
116	The ABCA4 2588G>C Stargardt mutation: single origin and increasing frequency from South-West to North-East Europe. , 0, .		1