Josseline Kaplan

List of Publications by Year
in descending order

35 papers	3,300 citations	279701 23 h-index	360920 35 g-index
35 all docs	35 docs citations	35 times ranked	4516 citing authors

#	Article	IF	Citations
1	Nuclear gene OPA1, encoding a mitochondrial dynamin-related protein, is mutated in dominant optic atrophy. Nature Genetics, 2000, 26, 207-210.	9.4	1,275
2	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.	1.1	313
3	Spectrum of NPHP6/CEP290 mutations in Leber congenital amaurosis and delineation of the associated phenotype. Human Mutation, 2007, 28, 416-416.	1.1	224
4	Spastic paraplegia gene 7 in patients with spasticity and/or optic neuropathy. Brain, 2012, 135, 2980-2993.	3.7	148
5	Leber Congenital Amaurosis. Molecular Genetics and Metabolism, 1999, 68, 200-208.	0.5	140
6	Recessive and Dominant De Novo ITPR1 Mutations Cause Gillespie Syndrome. American Journal of Human Genetics, 2016, 98, 971-980.	2.6	113
7	Mutations in DNM1L, as in OPA1, result in dominant optic atrophy despite opposite effects on mitochondrial fusion and fission. Brain, 2017, 140, 2586-2596.	3.7	100
8	AON-mediated Exon Skipping Restores Ciliation in Fibroblasts Harboring the Common Leber Congenital Amaurosis CEP290 Mutation. Molecular Therapy - Nucleic Acids, 2012, 1, e29.	2.3	94
9	Impaired complex I repair causes recessive Leberâ€™s hereditary optic neuropathy. Journal of Clinical Investigation, 2021, 131, .	3.9	89
10	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.	1.8	86
11	TMEM126A, Encoding a Mitochondrial Protein, Is Mutated in Autosomal-Recessive Nonsyndromic Optic Atrophy. American Journal of Human Genetics, 2009, 84, 493-498.	2.6	85
12	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.	1.5	80
13	Recessive Mutations in RTN4IP1 Cause Isolated and Syndromic Optic Neuropathies. American Journal of Human Genetics, 2015, 97, 754-760.	2.6	54
14	FDXR Mutations Cause Sensorial Neuropathies and Expand the Spectrum of Mitochondrial Fe-S-Synthesis Diseases. American Journal of Human Genetics, 2017, 101, 630-637.	2.6	52
15	Relative Frequencies of Inherited Retinal Dystrophies and Optic Neuropathies in Southern France: Assessment of 21-year Data Management. Ophthalmic Epidemiology, 2013, 20, 13-25.	0.8	44
16	Mutations in DOCK7 in Individuals with Epileptic Encephalopathy and Cortical Blindness. American Journal of Human Genetics, 2014, 94, 891-897.	2.6	44
17	Compound heterozygosity for severe and hypomorphicNDUFS2mutations cause non-syndromic LHON-like optic neuropathy. Journal of Medical Genetics, 2017, 54, 346-356.	1.5	43
18	Dopachrome tautomerase variants in patients with oculocutaneous albinism. Genetics in Medicine, 2021, 23, 479-487.	1.1	33

#	Article	IF	Citations
19	Leber Congenital Amaurosis: From Darkness to Spotlight. Ophthalmic Genetics, 2008, 29, 92-98.	0.5	31
20	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.	1.4	31
21	Mutations in TUBB4B Cause a Distinctive Sensorineural Disease. American Journal of Human Genetics, 2017, 101, 1006-1012.	2.6	30
22	Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurology, 2018, 75, 105.	4.5	26
23	TMEM126A is a mitochondrial located mRNA (MLR) protein of the mitochondrial inner membrane. Biochimica Et Biophysica Acta - General Subjects, 2013, 1830, 3719-3733.	1.1	23
24	Highâ€resolution arrayâ€<scp>CGH</scp> in patients with oculocutaneous albinism identifies new deletions of the <scp>TYR</scp>,<scp> OCA</scp>2, and <scp>SLC</scp>45A2 genes and a complex rearrangement of the <scp>OCA</scp>2 gene. Pigment Cell and Melanoma Research, 2014, 27, 59-71.	1.5	23
25	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.	2.6	22
26	MCAT Mutations Cause Nuclear LHON-like Optic Neuropathy. Genes, 2021, 12, 521.	1.0	21
27	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.	1.0	15
28	RETINOCHOROIDAL ANASTOMOSIS ASSOCIATED WITH ENHANCED S-CONE SYNDROME. Retinal Cases and Brief Reports, 2019, 13, 295-299.	0.3	14
29	Submicroscopic Deletions at 13q32.1 Cause Congenital Microcoria. American Journal of Human Genetics, 2015, 96, 631-639.	2.6	13
30	Clinicoâ€molecular analysis of eleven patients with Hermanskyâ€“Pudlak type 5 syndrome, a mild form of <scp>HPS</scp>. Pigment Cell and Melanoma Research, 2017, 30, 563-570.	1.5	13
31	A neuropathological study of novel RTTN gene mutations causing a familial microcephaly with simplified gyral pattern. Birth Defects Research, 2018, 110, 598-602.	0.8	7
32	Reply: The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e29-e29.	3.7	5
33	Whole Locus Sequencing Identifies a Prevalent Founder Deep Intronic RPGRIP1 Pathologic Variant in the French Leber Congenital Amaurosis Cohort. Genes, 2021, 12, 287.	1.0	3
34	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.	0.8	3
35	Genetic Deciphering of Early-Onset and Severe Retinal Dystrophy Associated with Sensorineural Hearing Loss. Advances in Experimental Medicine and Biology, 2019, 1185, 233-238.	0.8	3