Sylvain Hanein

List of Publications by Year
in descending order

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45	3,317	218592	243529
papers	citations	26	44
		h-index	g-index

47	47	47	5921
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Exome Sequencing Links Corticospinal Motor Neuron Disease to Common Neurodegenerative Disorders. Science, 2014, 343, 506-511.	6.0	466
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	Nuclear Outsourcing of RNA Interference Components to Human Mitochondria. PLoS ONE, 2011, 6, e20746.	1.1	249
4	Spectrum of NPHP6/CEP290 mutations in Leber congenital amaurosis and delineation of the associated phenotype. Human Mutation, 2007, 28, 416-416.	1.1	224
5	Identification of the SPG15 Gene, Encoding Spastizin, as a Frequent Cause of Complicated Autosomal-Recessive Spastic Paraplegia, Including Kjellin Syndrome. American Journal of Human Genetics, 2008, 82, 992-1002.	2.6	192
6	Mainzer-Saldino Syndrome Is a Ciliopathy Caused by IFT140 Mutations. American Journal of Human Genetics, 2012, 90, 864-870.	2.6	173
7	Retinal Dehydrogenase 12 (RDH12) Mutations in Leber Congenital Amaurosis. American Journal of Human Genetics, 2004, 75, 639-646.	2.6	164
8	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.	1.4	148
9	Mutations in NMNAT1 cause Leber congenital amaurosis with early-onset severe macular and optic atrophy. Nature Genetics, 2012, 44, 975-977.	9.4	123
10	Pediatric Evans syndrome is associated with a high frequency of potentially damaging variants in immune genes. Blood, 2019, 134, 9-21.	0.6	102
11	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
12	ALDH1A3 Mutations Cause Recessive Anophthalmia and Microphthalmia. American Journal of Human Genetics, 2013, 92, 265-270.	2.6	92
13	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
14	Diagnostic Yield of Next-generation Sequencing in Very Early-onset Inflammatory Bowel Diseases: A Multicentre Study. Journal of Crohn's and Colitis, 2018, 12, 1104-1112.	0.6	68
15	De novo mutation screening in childhood-onset cerebellar atrophy identifies gain-of-function mutations in the CACNA1G calcium channel gene. Brain, 2018, 141, 1998-2013.	3.7	67
16	Mutations in ACTRT1 and its enhancer RNA elements lead to aberrant activation of Hedgehog signaling in inherited and sporadic basal cell carcinomas. Nature Medicine, 2017, 23, 1226-1233.	15.2	59
17	Mutation in a primate-conserved retrotransposon reveals a noncoding RNA as a mediator of infantile encephalopathy. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 4980-4985.	3.3	58
18	A first locus for isolated autosomal recessive optic atrophy (ROA1) maps to chromosome 8q. European Journal of Human Genetics, 2003, 11, 966-971.	1.4	47

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19	Low Prevalence of GSC Gene Mutations in a Large Cohort of Predominantly Caucasian Patients with Hidradenitis Suppurativa. Journal of Investigative Dermatology, 2020, 140, 2085-2088.e14.	0.3	47
20	Prenatal human ocular degeneration occurs in Leber's congenital amaurosis (LCA2). Journal of Gene Medicine, 2002, 4, 390-396.	1.4	46
21	Compound heterozygosity for severe and hypomorphicNDUFS2mutations cause non-syndromic LHON-like optic neuropathy. Journal of Medical Genetics, 2017, 54, 346-356.	1.5	43
22	NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. American Journal of Human Genetics, 2018, 102, 460-467.	2.6	40
23	The ABCA4 Gene in Autosomal Recessive Cone-Rod Dystrophies. American Journal of Human Genetics, 2002, 71, 1480-1482.	2.6	36
24	NDP gene mutations in 14 French families with Norrie disease. Human Mutation, 2003, 22, 499-499.	1.1	33
25	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.	0.5	29
26	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.	1.1	28
27	Impact of on-site clinical genetics consultations on diagnostic rate in children and young adults with autism spectrum disorder. Molecular Autism, 2019, 10, 33.	2.6	28
28	A novel locus for autosomal dominant â€œuncomplicatedâ€•hereditary spastic paraplegia maps to chromosome 8p21.1-q13.3. Human Genetics, 2007, 122, 261-273.	1.8	27
29	A novel locus for autosomal recessive spastic ataxia on chromosome 17p. Human Genetics, 2007, 121, 413-420.	1.8	26
30	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.	1.1	24
31	Spectrum ofSPATA7mutations in Leber congenital amaurosis and delineation of the associated phenotype. Human Mutation, 2010, 31, E1241-E1250.	1.1	24
32	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
33	Mutations in LCA5 are an uncommon cause of Leber congenital amaurosis (LCA) type II. Human Mutation, 2007, 28, 1245-1245.	1.1	22
34	Refinement of the SPG15 candidate interval and phenotypic heterogeneity in three large Arab families. Neurogenetics, 2007, 8, 307-315.	0.7	16
35	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
36	Spastic paraplegia 5: Locus refinement, candidate gene analysis and clinical description. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 854-861.	1.1	13

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37	Copy number variations and founder effect underlying complete IL-10RÎ² deficiency in Portuguese kindreds. PLoS ONE, 2018, 13, e0205826.	1.1	13
38	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.	1.4	12
39	Neutropenia in Patients with Common Variable Immunodeficiency: a Rare Event Associated with Severe Outcome. Journal of Clinical Immunology, 2017, 37, 715-726.	2.0	11
40	Prenatal Human Ocular Degeneration Occurs in Leberâ€™s Congenital Amaurosis. Advances in Experimental Medicine and Biology, 2003, , 59-68.	0.8	10
41	Disease-Associated Variants of the Rod-derived Cone Viability Factor (RdCVF) in Leber Congenital Amaurosis. , 2006, 572, 9-14.		8
42	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.	1.4	5
43	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
44	A First Locus for Isolated Autosomal Recessive Optic Atrophy (ROA1) Maps to Chromosome 8q21-q22. , 2006, 572, 21-27.		1
45	Leber Congenital Amaurosis â€" Genotyping Required for Possible Inclusion in a Clinical Trial. Advances in Experimental Medicine and Biology, 2003, 533, 69-77.	0.8	1