Jeremy A Schwartzentruber

List of Publications by Citations

89 papers	10,186 citations	45 h-index	95 g-index
95 ext. papers	12,606 ext. citations	13.8 avg, IF	4.98 L-index

#	Paper	IF	Citations
89	Driver mutations in histone H3.3 and chromatin remodelling genes in paediatric glioblastoma. *Nature*, **2012**, 482, 226-31	50.4	1655
88	Hotspot mutations in H3F3A and IDH1 define distinct epigenetic and biological subgroups of glioblastoma. *Cancer Cell*, **2012**, 22, 425-37	24.3	1243
87	K27M mutation in histone H3.3 defines clinically and biologically distinct subgroups of pediatric diffuse intrinsic pontine gliomas. *Acta Neuropathologica*, **2012**, 124, 439-47	14.3	629
86	Recurrent somatic alterations of FGFR1 and NTRK2 in pilocytic astrocytoma. *Nature Genetics*, **2013**, 45, 927-32	36.3	550
85	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. *Nature Genetics*, **2012**, 44, 934-40	36.3	521
84	Clonal selection drives genetic divergence of metastatic medulloblastoma. *Nature*, **2012**, 482, 529-33	50.4	322
83	Recurrent somatic mutations in ACVR1 in pediatric midline high-grade astrocytoma. *Nature Genetics*, **2014**, 46, 462-6	36.3	296
82	Frequent ATRX mutations and loss of expression in adult diffuse astrocytic tumors carrying IDH1/IDH2 and TP53 mutations. *Acta Neuropathologica*, **2012**, 124, 615-25	14.3	295
81	What can exome sequencing do for you?. *Journal of Medical Genetics*, **2011**, 48, 580-9	5.8	263
80	Biallelic mutations in BRCA1 cause a new Fanconi anemia subtype. *Cancer Discovery*, **2015**, 5, 135-42	24.4	215
79	Mutations in SETD2 and genes affecting histone H3K36 methylation target hemispheric high-grade gliomas. *Acta Neuropathologica*, **2013**, 125, 659-69	14.3	201
78	FORGE Canada Consortium: outcomes of a 2-year national rare-disease gene-discovery project. *American Journal of Human Genetics*, **2014**, 94, 809-17	11	174
77	Mapping the human genetic architecture of COVID-19. *Nature*, **2021**,	50.4	162
76	Mutations in NMNAT1 cause Leber congenital amaurosis and identify a new disease pathway for retinal degeneration. *Nature Genetics*, **2012**, 44, 1035-9	36.3	147
75	Haploinsufficiency of a spliceosomal GTPase encoded by EFTUD2 causes mandibulofacial dysostosis with microcephaly. *American Journal of Human Genetics*, **2012**, 90, 369-77	11	143
74	Haploinsufficiency of SF3B4, a component of the pre-mRNA spliceosomal complex, causes Nager syndrome. *American Journal of Human Genetics*, **2012**, 90, 925-33	11	135
73	CTP synthase 1 deficiency in humans reveals its central role in lymphocyte proliferation. *Nature*, **2014**, 510, 288-92	50.4	131

72	Fusion of TTYH1 with the C19MC microRNA cluster drives expression of a brain-specific DNMT3B isoform in the embryonal brain tumor ETMR. *Nature Genetics*, **2014**, 46, 39-44	36.3	131
71	Mutations in SRCAP, encoding SNF2-related CREBBP activator protein, cause Floating-Harbor syndrome. *American Journal of Human Genetics*, **2012**, 90, 308-13	11	130
70	Mutations in DDHD2, encoding an intracellular phospholipase A(1), cause a recessive form of complex hereditary spastic paraplegia. *American Journal of Human Genetics*, **2012**, 91, 1073-81	11	128
69	Mutations in SYNGAP1 cause intellectual disability, autism, and a specific form of epilepsy by inducing haploinsufficiency. *Human Mutation*, **2013**, 34, 385-94	4.7	126
68	Mutations in PIK3R1 cause SHORT syndrome. *American Journal of Human Genetics*, **2013**, 93, 158-66	11	125
67	Molecular and functional variation in iPSC-derived sensory neurons. *Nature Genetics*, **2018**, 50, 54-61	36.3	125
66	A recurrent PDGFRB mutation causes familial infantile myofibromatosis. *American Journal of Human Genetics*, **2013**, 92, 996-1000	11	108
65	Mutations in FLNC are Associated with Familial Restrictive Cardiomyopathy. *Human Mutation*, **2016**, 37, 269-79	4.7	101
64	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. *Nature Genetics*, **2014**, 46, 510-515	36.3	100
63	Mutations in KAT6B, encoding a histone acetyltransferase, cause Genitopatellar syndrome. *American Journal of Human Genetics*, **2012**, 90, 282-9	11	99
62	Mutations in C5ORF42 cause Joubert syndrome in the French Canadian population. *American Journal of Human Genetics*, **2012**, 90, 693-700	11	95
61	Mutations in NOTCH2 in families with Hajdu-Cheney syndrome. *Human Mutation*, **2011**, 32, 1114-7	4.7	79
60	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. *American Journal of Human Genetics*, **2017**, 100, 865-884	11	74
59	Exome sequencing as a diagnostic tool for pediatric-onset ataxia. *Human Mutation*, **2014**, 35, 45-9	4.7	72
58	Mutations in STAMBP, encoding a deubiquitinating enzyme, cause microcephaly-capillary malformation syndrome. *Nature Genetics*, **2013**, 45, 556-62	36.3	69
57	Mutations in CSPP1, encoding a core centrosomal protein, cause a range of ciliopathy phenotypes in humans. *American Journal of Human Genetics*, **2014**, 94, 73-9	11	63
56	Whole genome sequencing and imputation in isolated populations identify genetic associations with medically-relevant complex traits. *Nature Communications*, **2017**, 8, 15606	17.4	60
55	Specific combination of compound heterozygous mutations in 17β-hydroxysteroid dehydrogenase type 4 (HSD17B4) defines a new subtype of D-bifunctional protein deficiency. *Orphanet Journal of Rare Diseases*, **2012**, 7, 90	4.2	58

54	Differential stability of 2'F-ANA*RNA and ANA*RNA hybrid duplexes: roles of structure, pseudohydrogen bonding, hydration, ion uptake and flexibility. *Nucleic Acids Research*, **2010**, 38, 2498-511	20.1	58
53	Mutation in the nuclear-encoded mitochondrial isoleucyl-tRNA synthetase IARS2 in patients with cataracts, growth hormone deficiency with short stature, partial sensorineural deafness, and peripheral neuropathy or with Leigh syndrome. *Human Mutation*, **2014**, 35, 1285-9	4.7	57
52	Mutations in NFKB2 and potential genetic heterogeneity in patients with DAVID syndrome, having variable endocrine and immune deficiencies. *BMC Medical Genetics*, **2014**, 15, 139	2.1	56
51	Open Targets Platform: supporting systematic drug-target identification and prioritisation. *Nucleic Acids Research*, **2021**, 49, D1302-D1310	20.1	56
50	Mutations in TMEM231 cause Joubert syndrome in French Canadians. *Journal of Medical Genetics*, **2012**, 49, 636-41	5.8	55
49	Novel inborn error of folate metabolism: identification by exome capture and sequencing of mutations in the MTHFD1 gene in a single proband. *Journal of Medical Genetics*, **2011**, 48, 590-2	5.8	52
48	Open Targets Genetics: systematic identification of trait-associated genes using large-scale genetics and functional genomics. *Nucleic Acids Research*, **2021**, 49, D1311-D1320	20.1	49
47	Genome-wide meta-analysis, fine-mapping and integrative prioritization implicate new Alzheimer's disease risk genes. *Nature Genetics*, **2021**, 53, 392-402	36.3	49
46	Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. *Journal of Medical Genetics*, **2014**, 51, 470-4	5.8	45
45	GeneMatcher aids in the identification of a new malformation syndrome with intellectual disability, unique facial dysmorphisms, and skeletal and connective tissue abnormalities caused by de novo variants in HNRNPK. *Human Mutation*, **2015**, 36, 1009-1014	4.7	45
44	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. *American Journal of Human Genetics*, **2015**, 97, 744-53	11	41
43	Compound heterozygous mutations in glycyl-tRNA synthetase are a proposed cause of systemic mitochondrial disease. *BMC Medical Genetics*, **2014**, 15, 36	2.1	40
42	Disrupted auto-regulation of the spliceosomal gene SNRPB causes cerebro-costo-mandibular syndrome. *Nature Communications*, **2014**, 5, 4483	17.4	40
41	Congenital Visual Impairment and Progressive Microcephaly Due to Lysyl-Transfer Ribonucleic Acid (RNA) Synthetase (KARS) Mutations: The Expanding Phenotype of Aminoacyl-Transfer RNA Synthetase Mutations in Human Disease. *Journal of Child Neurology*, **2015**, 30, 1037-43	2.5	39
40	Bioinactive ACTH causing glucocorticoid deficiency. *Journal of Clinical Endocrinology and Metabolism*, **2013**, 98, 736-42	5.6	39
39	Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. *Neurobiology of Aging*, **2015**, 36, 1222.e1-5	5.6	35
38	Intellectual disability associated with a homozygous missense mutation in THOC6. *Orphanet Journal of Rare Diseases*, **2013**, 8, 62	4.2	34
37	CCDC88B is a novel regulator of maturation and effector functions of T cells during pathological inflammation. *Journal of Experimental Medicine*, **2014**, 211, 2519-35	16.6	32

36	An inherited immunoglobulin class-switch recombination deficiency associated with a defect in the INO80 chromatin remodeling complex. *Journal of Allergy and Clinical Immunology*, **2015**, 135, 998-1007.e6	11.5	30
35	Mutations in ALDH6A1 encoding methylmalonate semialdehyde dehydrogenase are associated with dysmyelination and transient methylmalonic aciduria. *Orphanet Journal of Rare Diseases*, **2013**, 8, 98	4.2	28
34	A novel multisystem disease associated with recessive mutations in the tyrosyl-tRNA synthetase (YARS) gene. *American Journal of Medical Genetics, Part A*, **2017**, 173, 126-134	2.5	27
33	Whole-exome sequencing in an individual with severe global developmental delay and intractable epilepsy identifies a novel, de novo GRIN2A mutation. *Epilepsia*, **2014**, 55, e75-9	6.4	26
32	Neuropathologic features of pontocerebellar hypoplasia type 6. *Journal of Neuropathology and Experimental Neurology*, **2014**, 73, 1009-25	3.1	26
31	A map of transcriptional heterogeneity and regulatory variation in human microglia. *Nature Genetics*, **2021**, 53, 861-868	36.3	26
30	A family segregating lethal neonatal coenzyme Q deficiency caused by mutations in COQ9. *Journal of Inherited Metabolic Disease*, **2018**, 41, 719-729	5.4	24
29	Metaphyseal dysplasia with maxillary hypoplasia and brachydactyly is caused by a duplication in RUNX2. *American Journal of Human Genetics*, **2013**, 92, 252-8	11	23
28	Histone H3 mutations in pediatric brain tumors. *Cold Spring Harbor Perspectives in Biology*, **2014**, 6, a018689	10.2	23
27	An open approach to systematically prioritize causal variants and genes at all published human GWAS trait-associated loci. *Nature Genetics*, **2021**, 53, 1527-1533	36.3	22
26	Whole exome sequencing identifies the TNNI3K gene as a cause of familial conduction system disease and congenital junctional ectopic tachycardia. *International Journal of Cardiology*, **2015**, 185, 114-6	3.2	18
25	CYRI/FAM49B negatively regulates RAC1-driven cytoskeletal remodelling and protects against bacterial infection. *Nature Microbiology*, **2019**, 4, 1516-1531	26.6	18
24	An N-terminal formyl methionine on COX 1 is required for the assembly of cytochrome c oxidase. *Human Molecular Genetics*, **2015**, 24, 4103-13	5.6	18
23	Severe Neonatal Presentation of Mitochondrial Citrate Carrier (SLC25A1) Deficiency. *JIMD Reports*, **2016**, 30, 73-79	1.9	17
22	Genome-wide mouse mutagenesis reveals CD45-mediated T cell function as critical in protective immunity to HSV-1. *PLoS Pathogens*, **2013**, 9, e1003637	7.6	17
21	Molecular genetics of achromatopsia in Newfoundland reveal genetic heterogeneity, founder effects and the first cases of Jalili syndrome in North America. *Ophthalmic Genetics*, **2013**, 34, 119-29	1.2	16
20	Mutations in riboflavin transporter present with severe sensory loss and deafness in childhood. *Muscle and Nerve*, **2014**, 50, 775-9	3.4	15
19	Altered IFN-γ-mediated immunity and transcriptional expression patterns in N-Ethyl-N-nitrosourea-induced STAT4 mutants confer susceptibility to acute typhoid-like disease. *Journal of Immunology*, **2014**, 192, 259-70	5.3	15

18	Iron refractory iron deficiency anemia: presentation with hyperferritinemia and response to oral iron therapy. *Pediatrics*, **2013**, 131, e620-5	7.4	15
17	THEMIS is required for pathogenesis of cerebral malaria and protection against pulmonary tuberculosis. *Infection and Immunity*, **2015**, 83, 759-68	3.7	14
16	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a co-occurrence of multiple events. *American Journal of Medical Genetics, Part A*, **2016**, 170, 1820-5	2.5	14
15	A novel CCBE1 mutation leading to a mild form of hennekam syndrome: case report and review of the literature. *BMC Medical Genetics*, **2015**, 16, 28	2.1	12
14	A map of transcriptional heterogeneity and regulatory variation in human microglia		12
13	Novel 25 kb Deletion of MERTK Causes Retinitis Pigmentosa With Severe Progression **2017**, 58, 1736-1742		11
12	The utility of exome sequencing for genetic diagnosis in a familial microcephaly epilepsy syndrome. *BMC Neurology*, **2014**, 14, 22	3.1	11
11	Open Targets Genetics: An open approach to systematically prioritize causal variants and genes at all published human GWAS trait-associated loci		10
10	Very low depth whole genome sequencing in complex trait association studies		9
9	A mutation in the TMEM65 gene results in mitochondrial myopathy with severe neurological manifestations. *European Journal of Human Genetics*, **2017**, 25, 744-751	5.3	8
8	Bridging the gap between single molecule and ensemble methods for measuring lateral dynamics in the plasma membrane. *PLoS ONE*, **2013**, 8, e78096	3.7	8
7	Genome-wide meta-analysis, fine-mapping, and integrative prioritization identify new Alzheimer disease risk genes		5
6	Biallelic Loss-of-Function Variants in AIMP1 Cause a Rare Neurodegenerative Disease. *Journal of Child Neurology*, **2019**, 34, 74-80	2.5	5
5	Screening for functional transcriptional and splicing regulatory variants with GenIE. *Nucleic Acids Research*, **2020**, 48, e131	20.1	4
4	Expansion of the clinical phenotype of the distal 10q26.3 deletion syndrome to include ataxia and hyperemia of the hands and feet. *American Journal of Medical Genetics, Part A*, **2017**, 173, 1611-1619	2.5	3
3	Molecular and functional variation in iPSC-derived sensory neurons		2
2	A proteome-wide genetic investigation identifies several SARS-CoV-2-exploited host targets of clinical relevance. *ELife*, **2021**, 10,	8.9	2
1	CYRI1-mediated inhibition of RAC1 signalling restricts Salmonella Typhimurium infection		1