

# Jeremy A Schwartzenruber

## List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

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	An open approach to systematically prioritize causal variants and genes at all published human GWAS trait-associated loci. <i>Nature Genetics</i> , <b>2021</b> , 53, 1527-1533	36.3	22
88	A map of transcriptional heterogeneity and regulatory variation in human microglia. <i>Nature Genetics</i> , <b>2021</b> , 53, 861-868	36.3	26
87	Open Targets Genetics: systematic identification of trait-associated genes using large-scale genetics and functional genomics. <i>Nucleic Acids Research</i> , <b>2021</b> , 49, D1311-D1320	20.1	49
86	Open Targets Platform: supporting systematic drug-target identification and prioritisation. <i>Nucleic Acids Research</i> , <b>2021</b> , 49, D1302-D1310	20.1	56
85	Genome-wide meta-analysis, fine-mapping and integrative prioritization implicate new Alzheimer's disease risk genes. <i>Nature Genetics</i> , <b>2021</b> , 53, 392-402	36.3	49
84	Mapping the human genetic architecture of COVID-19. <i>Nature</i> , <b>2021</b> ,	50.4	162
83	A proteome-wide genetic investigation identifies several SARS-CoV-2-exploited host targets of clinical relevance. <i>ELife</i> , <b>2021</b> , 10,	8.9	2
82	Screening for functional transcriptional and splicing regulatory variants with GenIE. <i>Nucleic Acids Research</i> , <b>2020</b> , 48, e131	20.1	4
81	CYRI/FAM49B negatively regulates RAC1-driven cytoskeletal remodelling and protects against bacterial infection. <i>Nature Microbiology</i> , <b>2019</b> , 4, 1516-1531	26.6	18
80	Biallelic Loss-of-Function Variants in AIMP1 Cause a Rare Neurodegenerative Disease. <i>Journal of Child Neurology</i> , <b>2019</b> , 34, 74-80	2.5	5
79	A family segregating lethal neonatal coenzyme Q deficiency caused by mutations in COQ9. <i>Journal of Inherited Metabolic Disease</i> , <b>2018</b> , 41, 719-729	5.4	24
78	Molecular and functional variation in iPSC-derived sensory neurons. <i>Nature Genetics</i> , <b>2018</b> , 50, 54-61	36.3	125
77	Expansion of the clinical phenotype of the distal 10q26.3 deletion syndrome to include ataxia and hyperemia of the hands and feet. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 1611-1619	2.5	3
76	Whole genome sequencing and imputation in isolated populations identify genetic associations with medically-relevant complex traits. <i>Nature Communications</i> , <b>2017</b> , 8, 15606	17.4	60
75	Whole-Genome Sequencing Coupled to Imputation Discovers Genetic Signals for Anthropometric Traits. <i>American Journal of Human Genetics</i> , <b>2017</b> , 100, 865-884	11	74
74	A mutation in the TMEM65 gene results in mitochondrial myopathy with severe neurological manifestations. <i>European Journal of Human Genetics</i> , <b>2017</b> , 25, 744-751	5.3	8
73	Novel 25 kb Deletion of MERTK Causes Retinitis Pigmentosa With Severe Progression <b>2017</b> , 58, 1736-1742		11

72	A novel multisystem disease associated with recessive mutations in the tyrosyl-tRNA synthetase (YARS) gene. <i>American Journal of Medical Genetics, Part A</i> , <b>2017</b> , 173, 126-134	2.5	27
71	Severe Neonatal Presentation of Mitochondrial Citrate Carrier (SLC25A1) Deficiency. <i>JIMD Reports</i> , <b>2016</b> , 30, 73-79	1.9	17
70	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a co-occurrence of multiple events. <i>American Journal of Medical Genetics, Part A</i> , <b>2016</b> , 170, 1820-5	2.5	14
69	Mutations in FLNC are Associated with Familial Restrictive Cardiomyopathy. <i>Human Mutation</i> , <b>2016</b> , 37, 269-79	4.7	101
68	Biallelic mutations in BRCA1 cause a new Fanconi anemia subtype. <i>Cancer Discovery</i> , <b>2015</b> , 5, 135-42	24.4	215
67	Whole exome sequencing identifies the TNNI3K gene as a cause of familial conduction system disease and congenital junctional ectopic tachycardia. <i>International Journal of Cardiology</i> , <b>2015</b> , 185, 114-6	3.2	18
66	THEMIS is required for pathogenesis of cerebral malaria and protection against pulmonary tuberculosis. <i>Infection and Immunity</i> , <b>2015</b> , 83, 759-68	3.7	14
65	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. <i>American Journal of Human Genetics</i> , <b>2015</b> , 97, 744-53	11	41
64	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. <i>Journal of Child Neurology</i> , <b>2015</b> , 30, 1037-43	2.5	39
63	A novel CCBE1 mutation leading to a mild form of hennekam syndrome: case report and review of the literature. <i>BMC Medical Genetics</i> , <b>2015</b> , 16, 28	2.1	12
62	An inherited immunoglobulin class-switch recombination deficiency associated with a defect in the INO80 chromatin remodeling complex. <i>Journal of Allergy and Clinical Immunology</i> , <b>2015</b> , 135, 998-1007.e6	11.5	30
61	Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. <i>Neurobiology of Aging</i> , <b>2015</b> , 36, 1222.e1-5	5.6	35
60	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. <i>Human Mutation</i> , <b>2015</b> , 36, 1009-1014	4.7	45
59	An N-terminal formyl methionine on COX 1 is required for the assembly of cytochrome c oxidase. <i>Human Molecular Genetics</i> , <b>2015</b> , 24, 4103-13	5.6	18
58	The utility of exome sequencing for genetic diagnosis in a familial microcephaly epilepsy syndrome. <i>BMC Neurology</i> , <b>2014</b> , 14, 22	3.1	11
57	CTP synthase 1 deficiency in humans reveals its central role in lymphocyte proliferation. <i>Nature</i> , <b>2014</b> , 510, 288-92	50.4	131
56	Fusion of TTYH1 with the C19MC microRNA cluster drives expression of a brain-specific DNMT3B isoform in the embryonal brain tumor ETMR. <i>Nature Genetics</i> , <b>2014</b> , 46, 39-44	36.3	131
55	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. <i>Human Mutation</i> , <b>2014</b> , 35, 1285-9	4.7	57

54	CCDC88B is a novel regulator of maturation and effector functions of T cells during pathological inflammation. <i>Journal of Experimental Medicine</i> , <b>2014</b> , 211, 2519-35	16.6	32
53	De novo CCND2 mutations leading to stabilization of cyclin D2 cause megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome. <i>Nature Genetics</i> , <b>2014</b> , 46, 510-515	36.3	100
52	Compound heterozygous mutations in glycyl-tRNA synthetase are a proposed cause of systemic mitochondrial disease. <i>BMC Medical Genetics</i> , <b>2014</b> , 15, 36	2.1	40
51	Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. <i>Journal of Medical Genetics</i> , <b>2014</b> , 51, 470-4	5.8	45
50	FORGE Canada Consortium: outcomes of a 2-year national rare-disease gene-discovery project. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 809-17	11	174
49	Mutations in riboflavin transporter present with severe sensory loss and deafness in childhood. <i>Muscle and Nerve</i> , <b>2014</b> , 50, 775-9	3.4	15
48	Mutations in NFKB2 and potential genetic heterogeneity in patients with DAVID syndrome, having variable endocrine and immune deficiencies. <i>BMC Medical Genetics</i> , <b>2014</b> , 15, 139	2.1	56
47	Recurrent somatic mutations in ACVR1 in pediatric midline high-grade astrocytoma. <i>Nature Genetics</i> , <b>2014</b> , 46, 462-6	36.3	296
46	Disrupted auto-regulation of the spliceosomal gene SNRNPB causes cerebro-costo-mandibular syndrome. <i>Nature Communications</i> , <b>2014</b> , 5, 4483	17.4	40
45	Whole-exome sequencing in an individual with severe global developmental delay and intractable epilepsy identifies a novel, de novo GRIN2A mutation. <i>Epilepsia</i> , <b>2014</b> , 55, e75-9	6.4	26
44	Exome sequencing as a diagnostic tool for pediatric-onset ataxia. <i>Human Mutation</i> , <b>2014</b> , 35, 45-9	4.7	72
43	Histone H3 mutations in pediatric brain tumors. <i>Cold Spring Harbor Perspectives in Biology</i> , <b>2014</b> , 6, a0186892	6.9	23
42	Altered IFN- $\gamma$ -mediated immunity and transcriptional expression patterns in N-Ethyl-N-nitrosourea-induced STAT4 mutants confer susceptibility to acute typhoid-like disease. <i>Journal of Immunology</i> , <b>2014</b> , 192, 259-70	5.3	15
41	Neuropathologic features of pontocerebellar hypoplasia type 6. <i>Journal of Neuropathology and Experimental Neurology</i> , <b>2014</b> , 73, 1009-25	3.1	26
40	Mutations in CSPP1, encoding a core centrosomal protein, cause a range of ciliopathy phenotypes in humans. <i>American Journal of Human Genetics</i> , <b>2014</b> , 94, 73-9	11	63
39	Mutations in ALDH6A1 encoding methylmalonate semialdehyde dehydrogenase are associated with dysmyelination and transient methylmalonic aciduria. <i>Orphanet Journal of Rare Diseases</i> , <b>2013</b> , 8, 98	4.2	28
38	Intellectual disability associated with a homozygous missense mutation in THOC6. <i>Orphanet Journal of Rare Diseases</i> , <b>2013</b> , 8, 62	4.2	34
37	Recurrent somatic alterations of FGFR1 and NTRK2 in pilocytic astrocytoma. <i>Nature Genetics</i> , <b>2013</b> , 45, 927-32	36.3	550

36	Metaphyseal dysplasia with maxillary hypoplasia and brachydactyly is caused by a duplication in RUNX2. <i>American Journal of Human Genetics</i> , <b>2013</b> , 92, 252-8	11	23
35	Mutations in SYNGAP1 cause intellectual disability, autism, and a specific form of epilepsy by inducing haploinsufficiency. <i>Human Mutation</i> , <b>2013</b> , 34, 385-94	4.7	126
34	Mutations in PIK3R1 cause SHORT syndrome. <i>American Journal of Human Genetics</i> , <b>2013</b> , 93, 158-66	11	125
33	A recurrent PDGFRB mutation causes familial infantile myofibromatosis. <i>American Journal of Human Genetics</i> , <b>2013</b> , 92, 996-1000	11	108
32	Molecular genetics of achromatopsia in Newfoundland reveal genetic heterogeneity, founder effects and the first cases of Jalili syndrome in North America. <i>Ophthalmic Genetics</i> , <b>2013</b> , 34, 119-29	1.2	16
31	Mutations in SETD2 and genes affecting histone H3K36 methylation target hemispheric high-grade gliomas. <i>Acta Neuropathologica</i> , <b>2013</b> , 125, 659-69	14.3	201
30	Iron refractory iron deficiency anemia: presentation with hyperferritinemia and response to oral iron therapy. <i>Pediatrics</i> , <b>2013</b> , 131, e620-5	7.4	15
29	Bioinactive ACTH causing glucocorticoid deficiency. <i>Journal of Clinical Endocrinology and Metabolism</i> , <b>2013</b> , 98, 736-42	5.6	39
28	Genome-wide mouse mutagenesis reveals CD45-mediated T cell function as critical in protective immunity to HSV-1. <i>PLoS Pathogens</i> , <b>2013</b> , 9, e1003637	7.6	17
27	Mutations in STAMBP, encoding a deubiquitinating enzyme, cause microcephaly-capillary malformation syndrome. <i>Nature Genetics</i> , <b>2013</b> , 45, 556-62	36.3	69
26	Bridging the gap between single molecule and ensemble methods for measuring lateral dynamics in the plasma membrane. <i>PLoS ONE</i> , <b>2013</b> , 8, e78096	3.7	8
25	Mutations in NMNAT1 cause Leber congenital amaurosis and identify a new disease pathway for retinal degeneration. <i>Nature Genetics</i> , <b>2012</b> , 44, 1035-9	36.3	147
24	Frequent ATRX mutations and loss of expression in adult diffuse astrocytic tumors carrying IDH1/IDH2 and TP53 mutations. <i>Acta Neuropathologica</i> , <b>2012</b> , 124, 615-25	14.3	295
23	Mutations in DDHD2, encoding an intracellular phospholipase A(1), cause a recessive form of complex hereditary spastic paraplegia. <i>American Journal of Human Genetics</i> , <b>2012</b> , 91, 1073-81	11	128
22	K27M mutation in histone H3.3 defines clinically and biologically distinct subgroups of pediatric diffuse intrinsic pontine gliomas. <i>Acta Neuropathologica</i> , <b>2012</b> , 124, 439-47	14.3	629
21	Hotspot mutations in H3F3A and IDH1 define distinct epigenetic and biological subgroups of glioblastoma. <i>Cancer Cell</i> , <b>2012</b> , 22, 425-37	24.3	1243
20	Specific combination of compound heterozygous mutations in 17 $\beta$ -hydroxysteroid dehydrogenase type 4 (HSD17B4) defines a new subtype of D-bifunctional protein deficiency. <i>Orphanet Journal of Rare Diseases</i> , <b>2012</b> , 7, 90	4.2	58
19	Driver mutations in histone H3.3 and chromatin remodelling genes in paediatric glioblastoma. <i>Nature</i> , <b>2012</b> , 482, 226-31	50.4	1655

18	Clonal selection drives genetic divergence of metastatic medulloblastoma. <i>Nature</i> , <b>2012</b> , 482, 529-33	50.4	322
17	De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related megalencephaly syndromes. <i>Nature Genetics</i> , <b>2012</b> , 44, 934-40	36.3	521
16	Mutations in KAT6B, encoding a histone acetyltransferase, cause Genitopatellar syndrome. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 282-9	11	99
15	Mutations in SRCAP, encoding SNF2-related CREBBP activator protein, cause Floating-Harbor syndrome. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 308-13	11	130
14	Haploinsufficiency of a spliceosomal GTPase encoded by EFTUD2 causes mandibulofacial dysostosis with microcephaly. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 369-77	11	143
13	Mutations in C5ORF42 cause Joubert syndrome in the French Canadian population. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 693-700	11	95
12	Haploinsufficiency of SF3B4, a component of the pre-mRNA spliceosomal complex, causes Nager syndrome. <i>American Journal of Human Genetics</i> , <b>2012</b> , 90, 925-33	11	135
11	Mutations in TMEM231 cause Joubert syndrome in French Canadians. <i>Journal of Medical Genetics</i> , <b>2012</b> , 49, 636-41	5.8	55
10	What can exome sequencing do for you?. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 580-9	5.8	263
9	Mutations in NOTCH2 in families with Hajdu-Cheney syndrome. <i>Human Mutation</i> , <b>2011</b> , 32, 1114-7	4.7	79
8	Novel inborn error of folate metabolism: identification by exome capture and sequencing of mutations in the MTHFD1 gene in a single proband. <i>Journal of Medical Genetics</i> , <b>2011</b> , 48, 590-2	5.8	52
7	Differential stability of 2'F-ANA*RNA and ANA*RNA hybrid duplexes: roles of structure, pseudohydrogen bonding, hydration, ion uptake and flexibility. <i>Nucleic Acids Research</i> , <b>2010</b> , 38, 2498-511	20.1	58
6	Molecular and functional variation in iPSC-derived sensory neurons		2
5	Very low depth whole genome sequencing in complex trait association studies		9
4	A map of transcriptional heterogeneity and regulatory variation in human microglia		12
3	Genome-wide meta-analysis, fine-mapping, and integrative prioritization identify new Alzheimer's disease risk genes		5
2	Open Targets Genetics: An open approach to systematically prioritize causal variants and genes at all published human GWAS trait-associated loci		10
1	CYRI1-mediated inhibition of RAC1 signalling restricts Salmonella Typhimurium infection		1

