Taila Hartley

List of Publications by Year
in descending order

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Version: 2024-02-01

66 papers	2,892 citations	236833 25 h-index	189801 50 g-index
67 all docs	67 docs citations	67 times ranked	6388 citing authors

#	Article	IF	Citations
1	Utility of wholeâ€exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clinical Genetics, 2016, 89, 275-284.	1.0	323
2	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
3	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nature Medicine, 2019, 25, 911-919.	15.2	221
4	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. Journal of Medical Genetics, 2015, 52, 431-437.	1.5	187
5	ClinPred: Prediction Tool to Identify Disease-Relevant Nonsynonymous Single-Nucleotide Variants. American Journal of Human Genetics, 2018, 103, 474-483.	2.6	149
6	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	13.5	113
7	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. Human Mutation, 2015, 36, 931-940.	1.1	107
8	Mutations in LAMA1 Cause Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy. American Journal of Human Genetics, 2014, 95, 227-234.	2.6	92
9	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by wholeâ€exome sequencing. Clinical Genetics, 2017, 92, 281-289.	1.0	92
10	Wholeâ€exome sequencing broadens the phenotypic spectrum of rare pediatric epilepsy: a retrospective study. Clinical Genetics, 2015, 88, 34-40.	1.0	79
11	A recurrent de novo mutation in TMEM106B causes hypomyelinating leukodystrophy. Brain, 2017, 140, 3105-3111.	3.7	64
12	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. American Journal of Human Genetics, 2017, 100, 281-296.	2.6	59
13	The Matchmaker Exchange API: Automating Patient Matching Through the Exchange of Structured Phenotypic and Genotypic Profiles. Human Mutation, 2015, 36, 922-927.	1.1	50
14	Biallelic Mutations in LRRC56, Encoding a Protein Associated with Intraflagellar Transport, Cause Mucociliary Clearance and Laterality Defects. American Journal of Human Genetics, 2018, 103, 727-739.	2.6	49
15	Wholeâ€exome sequencing is a valuable diagnostic tool for inherited peripheral neuropathies: Outcomes from a cohort of 50 families. Clinical Genetics, 2018, 93, 301-309.	1.0	48
16	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an episignature of X chromosomes in females. American Journal of Human Genetics, 2021, 108, 502-516.	2.6	48
17	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	1.1	45
18	Biallelic Mutations in UNC80 Cause Persistent Hypotonia, Encephalopathy, Growth Retardation, and Severe Intellectual Disability. American Journal of Human Genetics, 2016, 98, 202-209.	2.6	45

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19	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	5.8	43
20	Matchmaking facilitates the diagnosis of an autosomal-recessive mitochondrial disease caused by biallelic mutation of the tRNA isopentenyltransferase (TRIT1) gene. Human Mutation, 2017, 38, 511-516.	1.1	39
21	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	1.4	39
22	A novel multisystem disease associated with recessive mutations in the tyrosylâ€tRNA synthetase (YARS) gene. American Journal of Medical Genetics, Part A, 2017, 173, 126-134.	0.7	36
23	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
24	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	3.7	31
25	The value of diagnostic testing for parents of children with rare genetic diseases. Genetics in Medicine, 2019, 21, 2798-2806.	1.1	31
26	Concordance between wholeâ€exome sequencing and clinical Sanger sequencing: implications for patient care. Molecular Genetics & Genomic Medicine, 2016, 4, 504-512.	0.6	30
27	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, 114-116.	0.8	29
28	Diagnostic clarity of exome sequencing following negative comprehensive panel testing in the neonatal intensive care unit. American Journal of Medical Genetics, Part A, 2018, 176, 1688-1691.	0.7	28
29	Novel ELOVL4 mutation associated with erythrokeratodermia and spinocerebellar ataxia (SCA) Tj ETQq1 1 0.784314 rgBT /Ove	0.9	27
30	Whole-transcriptome sequencing in blood provides a diagnosis of spinal muscular atrophy with progressive myoclonic epilepsy. Human Mutation, 2017, 38, 611-614.	1.1	25
31	Benchmarking outcomes in the Neonatal Intensive Care Unit: Cytogenetic and molecular diagnostic rates in a retrospective cohort. American Journal of Medical Genetics, Part A, 2017, 173, 1839-1847.	0.7	25
32	The unsolved rare genetic disease atlas? An analysis of the unexplained phenotypic descriptions in OMIMÂ®. , 2018, 178, 458-463.		25
33	Application of exome sequencing for prenatal diagnosis: a rapid scoping review. Genetics in Medicine, 2020, 22, 1925-1934.	1.1	25
34	SHORT syndrome due to a novel de novo mutation in PRKCE (Protein Kinase CÉ›) impairing TORC2-dependent AKT activation. Human Molecular Genetics, 2017, 26, 3713-3721.	1.4	22
35	A de novo mutation in RPL10 causes a rare X-linked ribosomopathy characterized by syndromic intellectual disability and epilepsy: A new case and review of the literature. European Journal of Medical Genetics, 2018, 61, 89-93.	0.7	22
36	Direct health-care costs for children diagnosed with genetic diseases are significantly higher than for children with other chronic diseases. Genetics in Medicine, 2019, 21, 1049-1057.	1.1	20

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37	Lateral meningocele (Lehman) syndrome: A child with a novel NOTCH3 mutation. American Journal of Medical Genetics, Part A, 2016, 170, 1070-1075.	0.7	19
38	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a coâ€•occurrence of multiple events. American Journal of Medical Genetics, Part A, 2016, 170, 1820-1825.	0.7	19
39	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 119-133.	0.7	17
40	A novel pathogenic variant in TNPO3 in a Hungarian family with limb-girdle muscular dystrophy 1F. European Journal of Medical Genetics, 2019, 62, 103662.	0.7	15
41	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <scp>PIGQ</scp>: Report of seven new subjects and review of the literature. Journal of Inherited Metabolic Disease, 2020, 43, 1321-1332.	1.7	15
42	Outcome of over 1500 matches through the Matchmaker Exchange for rare disease gene discovery: The 2-year experience of Care4Rare Canada. Genetics in Medicine, 2022, 24, 100-108.	1.1	15
43	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by RSPRY1 Mutations. American Journal of Human Genetics, 2015, 97, 608-615.	2.6	14
44	Evaluation of exome filtering techniques for the analysis of clinically relevant genes. Human Mutation, 2018, 39, 197-201.	1.1	13
45	p21 proteinâ€•activated kinase 1 is associated with severe regressive autism, and epilepsy. Clinical Genetics, 2019, 96, 449-455.	1.0	13
46	Clinical application of fetal genome-wide sequencing during pregnancy: position statement of the Canadian College of Medical Geneticists. Journal of Medical Genetics, 2022, 59, 931-937.	1.5	13
47	Autosomal dominant cutis laxa with progeroid features due to a novel, de novo mutation in ALDH18A1. Journal of Human Genetics, 2017, 62, 661-663.	1.1	12
48	A novel mutation in LAMC3 associated with generalized polymicrogyria of the cortex and epilepsy. Neurogenetics, 2018, 19, 61-65.	0.7	12
49	Lysosomal dysfunction in TMEM106B hypomyelinating leukodystrophy. Neurology: Genetics, 2018, 4, e288.	0.9	11
50	When to think outside the autozygome: Best practices for exome sequencing in â€œconsanguineousâ€• families. Clinical Genetics, 2020, 97, 835-843.	1.0	11
51	Intrafamilial variability of limb-girdle muscular dystrophy, LGMD1D type. European Journal of Medical Genetics, 2020, 63, 103655.	0.7	10
52	ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. American Journal of Human Genetics, 2021, 108, 2017-2023.	2.6	9
53	PhenomeCentral: 7 years of rare disease matchmaking. Human Mutation, 2022, , .	1.1	9
54	NID1 variant associated with occipital cephaloceles in a family expressing a spectrum of phenotypes. American Journal of Medical Genetics, Part A, 2019, 179, 837-841.	0.7	8

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55	Fragile X testing as a second-tier test. Genetics in Medicine, 2017, 19, 1380-1380.	1.1	7
56	Channelopathies Are a Frequent Cause of Genetic Ataxias Associated with Cerebellar Atrophy. Movement Disorders Clinical Practice, 2020, 7, 940-949.	0.8	7
57	Implementation of Epilepsy Multigene Panel Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2020, 47, 61-68.	0.3	6
58	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. American Journal of Human Genetics, 2021, 108, 749-756.	2.6	6
59	A splice site and copy number variant responsible for TTC25-related primary ciliary dyskinesia. European Journal of Medical Genetics, 2021, 64, 104193.	0.7	4
60	The complexity of diagnosing rare disease: An organizing framework for outcomes research and health economics based on real-world evidence. Genetics in Medicine, 2022, 24, 694-702.	1.1	4
61	Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery.. Human Mutation, 2022, , .	1.1	4
62	Whole genome sequencing reveals biallelic <scp>PLA2G6</scp> mutations in siblings with cerebellar atrophy and cap myopathy. Clinical Genetics, 2021, 99, 746-748.	1.0	3
63	A novel intragenic DPF2 deletion identified by genome sequencing in an adult with clinical features of <scp>Coffinâ€Siris</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2493-2496.	0.7	3
64	The Benefit of Multigene Panel Testing for the Diagnosis and Management of the Genetic Epilepsies. Genes, 2022, 13, 872.	1.0	2
65	Neurophysiological Characteristics of Allgrove (Triple A) Syndrome: Case Report and Literature Review. Child Neurology Open, 2021, 8, 2329048X2110310.	0.5	1
66	Heterozygous De Novo <scp>KPNA3</scp> Mutations Cause Complex Hereditary Spastic Paraplegia. Annals of Neurology, 2022, 91, 730-732.	2.8	1