Taila Hartley

List of Publications by Year
in descending order

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66 papers	2,892 citations	236612 25 h-index	189595 50 g-index
67 all docs	67 docs citations	67 times ranked	6388 citing authors

#	Article	IF	Citations
1	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
2	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
3	Heterozygous De Novo <scp>KPNA3</scp> Mutations Cause Complex Hereditary Spastic Paraplegia. Annals of Neurology, 2022, 91, 730-732.	2.8	1
4	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
5	PhenomeCentral: 7 years of rare disease matchmaking. Human Mutation, 2022, , .	1.1	9
6	Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery.. Human Mutation, 2022, , .	1.1	4
7	The Benefit of Multigene Panel Testing for the Diagnosis and Management of the Genetic Epilepsies. Genes, 2022, 13, 872.	1.0	2
8	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
9	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
10	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
11	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
12	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
13	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
14	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
15	Neurophysiological Characteristics of Allgrove (Triple A) Syndrome: Case Report and Literature Review. Child Neurology Open, 2021, 8, 2329048X2110310.	0.5	1
16	Intrafamilial variability of limb-girdle muscular dystrophy, LGMD1D type. European Journal of Medical Genetics, 2020, 63, 103655.	0.7	10
17	Implementation of Epilepsy Multigene Panel Testing in Ontario, Canada. Canadian Journal of Neurological Sciences, 2020, 47, 61-68.	0.3	6
18	Germline AGO2 mutations impair RNA interference and human neurological development. Nature Communications, 2020, 11, 5797.	5.8	43

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19	Application of exome sequencing for prenatal diagnosis: a rapid scoping review. Genetics in Medicine, 2020, 22, 1925-1934.	1.1	25
20	Channelopathies Are a Frequent Cause of Genetic Ataxias Associated with Cerebellar Atrophy. Movement Disorders Clinical Practice, 2020, 7, 940-949.	0.8	7
21	When to think outside the autozygome: Best practices for exome sequencing in â€œconsanguineousâ€• families. Clinical Genetics, 2020, 97, 835-843.	1.0	11
22	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
23	p21 proteinâ€activated kinase 1 is associated with severe regressive autism, and epilepsy. Clinical Genetics, 2019, 96, 449-455.	1.0	13
24	The value of diagnostic testing for parents of children with rare genetic diseases. Genetics in Medicine, 2019, 21, 2798-2806.	1.1	31
25	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. Nature Medicine, 2019, 25, 911-919.	15.2	221
26	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
27	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. Cell, 2019, 177, 32-37.	13.5	113
28	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
29	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
30	A novel mutation in LAMC3 associated with generalized polymicrogyria of the cortex and epilepsy. Neurogenetics, 2018, 19, 61-65.	0.7	12
31	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
32	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
33	Evaluation of exome filtering techniques for the analysis of clinically relevant genes. Human Mutation, 2018, 39, 197-201.	1.1	13
34	Lysosomal dysfunction in TMEM106B hypomyelinating leukodystrophy. Neurology: Genetics, 2018, 4, e288.	0.9	11
35	The unsolved rare genetic disease atlas? An analysis of the unexplained phenotypic descriptions in OMIMÂ®. , 2018, 178, 458-463.		25
36	Novel ELOVL4 mutation associated with erythrokeratodermia and spinocerebellar ataxia (SCA) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	0.9	27

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37	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
38	ClinPred: Prediction Tool to Identify Disease-Relevant Nonsynonymous Single-Nucleotide Variants. American Journal of Human Genetics, 2018, 103, 474-483.	2.6	149
39	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	2.6	36
40	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
41	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. American Journal of Human Genetics, 2017, 100, 281-296.	2.6	59
42	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
43	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by wholeâ€exome sequencing. Clinical Genetics, 2017, 92, 281-289.	1.0	92
44	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
45	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
46	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	2.6	305
47	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
48	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	3.7	31
49	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	1.4	39
50	Fragile X testing as a second-tier test. Genetics in Medicine, 2017, 19, 1380-1380.	1.1	7
51	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
52	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
53	A recurrent de novo mutation in TMEM106B causes hypomyelinating leukodystrophy. Brain, 2017, 140, 3105-3111.	3.7	64
54	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	1.1	45

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55	Concordance between wholeâ€exome sequencing and clinical Sanger sequencing: implications for patient care. Molecular Genetics & Genomic Medicine, 2016, 4, 504-512.	0.6	30
56	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
57	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
58	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
59	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
60	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. Human Mutation, 2015, 36, 931-940.	1.1	107
61	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
62	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
63	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
64	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by RSPRY1 Mutations. American Journal of Human Genetics, 2015, 97, 608-615.	2.6	14
65	Wholeâ€exome sequencing broadens the phenotypic spectrum of rare pediatric epilepsy: a retrospective study. Clinical Genetics, 2015, 88, 34-40.	1.0	79
66	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