

Taila Hartley

List of Publications by Year in descending order

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Version: 2024-02-01

66
papers

2,892
citations

236912
25
h-index

189881
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. <i>Journal of Medical Genetics</i> , 2022, 59, 931-937.	3.2	13
2	Outcome of over 1500 matches through the Matchmaker Exchange for rare disease gene discovery: The 2-year experience of Care4Rare Canada. <i>Genetics in Medicine</i> , 2022, 24, 100-108.	2.4	15
3	Heterozygous De Novo <i>KPNA3</i> Mutations Cause Complex Hereditary Spastic Paraplegia. <i>Annals of Neurology</i> , 2022, 91, 730-732.	5.3	1
4	The complexity of diagnosing rare disease: An organizing framework for outcomes research and health economics based on real-world evidence. <i>Genetics in Medicine</i> , 2022, 24, 694-702.	2.4	4
5	PhenomeCentral: 7 years of rare disease matchmaking. <i>Human Mutation</i> , 2022, , .	2.5	9
6	Genomics4RD: An integrated platform to share Canadian deep-phenotype and multiomic data for international rare disease gene discovery.. <i>Human Mutation</i> , 2022, , .	2.5	4
7	The Benefit of Multigene Panel Testing for the Diagnosis and Management of the Genetic Epilepsies. <i>Genes</i> , 2022, 13, 872.	2.4	2
8	A novel intragenic <i>DPF2</i> deletion identified by genome sequencing in an adult with clinical features of Coffinâ€širis syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2493-2496.	1.2	3
9	Alternative genomic diagnoses for individuals with a clinical diagnosis of Dubowitz syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 119-133.	1.2	17
10	Whole genome sequencing reveals biallelic <i>PLA2G6</i> mutations in siblings with cerebellar atrophy and cap myopathy. <i>Clinical Genetics</i> , 2021, 99, 746-748.	2.0	3
11	SPEN haploinsufficiency causes a neurodevelopmental disorder overlapping proximal 1p36 deletion syndrome with an epismutation of X chromosomes in females. <i>American Journal of Human Genetics</i> , 2021, 108, 502-516.	6.2	48
12	A DNA repair disorder caused by de novo monoallelic DDB1 variants is associated with a neurodevelopmental syndrome. <i>American Journal of Human Genetics</i> , 2021, 108, 749-756.	6.2	6
13	A splice site and copy number variant responsible for TTC25-related primary ciliary dyskinesia. <i>European Journal of Medical Genetics</i> , 2021, 64, 104193.	1.3	4
14	ABHD16A deficiency causes a complicated form of hereditary spastic paraplegia associated with intellectual disability and cerebral anomalies. <i>American Journal of Human Genetics</i> , 2021, 108, 2017-2023.	6.2	9
15	Neurophysiological Characteristics of Allgrove (Triple A) Syndrome: Case Report and Literature Review. <i>Child Neurology Open</i> , 2021, 8, 2329048X2110310.	1.1	1
16	Intrafamilial variability of limb-girdle muscular dystrophy, LGMD1D type. <i>European Journal of Medical Genetics</i> , 2020, 63, 103655.	1.3	10
17	Implementation of Epilepsy Multigene Panel Testing in Ontario, Canada. <i>Canadian Journal of Neurological Sciences</i> , 2020, 47, 61-68.	0.5	6
18	Germline AGO2 mutations impair RNA interference and human neurological development. <i>Nature Communications</i> , 2020, 11, 5797.	12.8	43

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19	Application of exome sequencing for prenatal diagnosis: a rapid scoping review. <i>Genetics in Medicine</i> , 2020, 22, 1925-1934.	2.4	25
20	Channelopathies Are a Frequent Cause of Genetic Ataxias Associated with Cerebellar Atrophy. <i>Movement Disorders Clinical Practice</i> , 2020, 7, 940-949.	1.5	7
21	When to think outside the autozygome: Best practices for exome sequencing in "consanguineous" families. <i>Clinical Genetics</i> , 2020, 97, 835-843.	2.0	11
22	Early infantile epileptic encephalopathy due to biallelic pathogenic variants in <i>PIGQ</i> : Report of seven new subjects and review of the literature. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 1321-1332.	3.6	15
23	p21 protein-activated kinase 1 is associated with severe regressive autism, and epilepsy. <i>Clinical Genetics</i> , 2019, 96, 449-455.	2.0	13
24	The value of diagnostic testing for parents of children with rare genetic diseases. <i>Genetics in Medicine</i> , 2019, 21, 2798-2806.	2.4	31
25	Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts. <i>Nature Medicine</i> , 2019, 25, 911-919.	30.7	221
26	A novel pathogenic variant in <i>TNPO3</i> in a Hungarian family with limb-girdle muscular dystrophy 1F. <i>European Journal of Medical Genetics</i> , 2019, 62, 103662.	1.3	15
27	A Diagnosis for All Rare Genetic Diseases: The Horizon and the Next Frontiers. <i>Cell</i> , 2019, 177, 32-37.	28.9	113
28	<i>NID1</i> variant associated with occipital cephaloceles in a family expressing a spectrum of phenotypes. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 837-841.	1.2	8
29	Direct health-care costs for children diagnosed with genetic diseases are significantly higher than for children with other chronic diseases. <i>Genetics in Medicine</i> , 2019, 21, 1049-1057.	2.4	20
30	A novel mutation in <i>LAMC3</i> associated with generalized polymicrogyria of the cortex and epilepsy. <i>Neurogenetics</i> , 2018, 19, 61-65.	1.4	12
31	Whole-exome sequencing is a valuable diagnostic tool for inherited peripheral neuropathies: Outcomes from a cohort of 50 families. <i>Clinical Genetics</i> , 2018, 93, 301-309.	2.0	48
32	A de novo mutation in <i>RPL10</i> causes a rare X-linked ribosomopathy characterized by syndromic intellectual disability and epilepsy: A new case and review of the literature. <i>European Journal of Medical Genetics</i> , 2018, 61, 89-93.	1.3	22
33	Evaluation of exome filtering techniques for the analysis of clinically relevant genes. <i>Human Mutation</i> , 2018, 39, 197-201.	2.5	13
34	Lysosomal dysfunction in <i>TMEM106B</i> hypomyelinating leukodystrophy. <i>Neurology: Genetics</i> , 2018, 4, e288.	1.9	11
35	The unsolved rare genetic disease atlas? An analysis of the unexplained phenotypic descriptions in OMIM®. , 2018, 178, 458-463.		25
36	Novel <i>ELOVL4</i> mutation associated with erythrokeratoderma and spinocerebellar ataxia (SCA) Tj ETQq0 0 0,rgBT /Overlock 10 Tf	1.9	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.	6.2	49
38	ClinPred: Prediction Tool to Identify Disease-Relevant Nonsynonymous Single-Nucleotide Variants. American Journal of Human Genetics, 2018, 103, 474-483.	6.2	149
39	De Novo Truncating Mutations in WASF1 Cause Intellectual Disability with Seizures. American Journal of Human Genetics, 2018, 103, 144-153.	6.2	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.	1.2	28
41	Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome. American Journal of Human Genetics, 2017, 100, 281-296.	6.2	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.	2.3	12
43	Debunking Occam's razor: Diagnosing multiple genetic diseases in families by whole-exome sequencing. Clinical Genetics, 2017, 92, 281-289.	2.0	92
44	Whole-transcriptome sequencing in blood provides a diagnosis of spinal muscular atrophy with progressive myoclonic epilepsy. Human Mutation, 2017, 38, 611-614.	2.5	25
45	Matchmaking facilitates the diagnosis of an autosomal-recessive mitochondrial disease caused by biallelic mutation of the tRNA isopentenyltransferase (<i>TRIT1</i>) gene. Human Mutation, 2017, 38, 511-516.	2.5	39
46	International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases. American Journal of Human Genetics, 2017, 100, 695-705.	6.2	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.	1.2	25
48	MCM3AP in recessive Charcot-Marie-Tooth neuropathy and mild intellectual disability. Brain, 2017, 140, 2093-2103.	7.6	31
49	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	2.9	39
50	Fragile X testing as a second-tier test. Genetics in Medicine, 2017, 19, 1380-1380.	2.4	7
51	A novel multisystem disease associated with recessive mutations in the tyrosyl-tRNA synthetase (<i>YARS</i>) gene. American Journal of Medical Genetics, Part A, 2017, 173, 126-134.	1.2	36
52	SHORT syndrome due to a novel de novo mutation in PRKCE (Protein Kinase C ϵ) impairing TORC2-dependent AKT activation. Human Molecular Genetics, 2017, 26, 3713-3721.	2.9	22
53	A recurrent de novo mutation in TMEM106B causes hypomyelinating leukodystrophy. Brain, 2017, 140, 3105-3111.	7.6	64
54	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	2.5	45

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55	Concordance between whole-exome sequencing and clinical Sanger sequencing: implications for patient care. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 504-512.	1.2	30
56	Lateral meningocele (Lehman) syndrome: A child with a novel <i>NOTCH3</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1070-1075.	1.2	19
57	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a co-occurrence of multiple events. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1820-1825.	1.2	19
58	Biallelic Mutations in <i>UNC80</i> Cause Persistent Hypotonia, Encephalopathy, Growth Retardation, and Severe Intellectual Disability. <i>American Journal of Human Genetics</i> , 2016, 98, 202-209.	6.2	45
59	Utility of whole-exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. <i>Clinical Genetics</i> , 2016, 89, 275-284.	2.0	323
60	PhenomeCentral: A Portal for Phenotypic and Genotypic Matchmaking of Patients with Rare Genetic Diseases. <i>Human Mutation</i> , 2015, 36, 931-940.	2.5	107
61	The Matchmaker Exchange API: Automating Patient Matching Through the Exchange of Structured Phenotypic and Genotypic Profiles. <i>Human Mutation</i> , 2015, 36, 922-927.	2.5	50
62	Whole exome sequencing identifies the <i>TNNI3K</i> gene as a cause of familial conduction system disease and congenital junctional ectopic tachycardia. <i>International Journal of Cardiology</i> , 2015, 185, 114-116.	1.7	29
63	The clinical application of genome-wide sequencing for monogenic diseases in Canada: Position Statement of the Canadian College of Medical Geneticists. <i>Journal of Medical Genetics</i> , 2015, 52, 431-437.	3.2	187
64	Identification of a Recognizable Progressive Skeletal Dysplasia Caused by <i>RSPRY1</i> Mutations. <i>American Journal of Human Genetics</i> , 2015, 97, 608-615.	6.2	14
65	Whole-exome sequencing broadens the phenotypic spectrum of rare pediatric epilepsy: a retrospective study. <i>Clinical Genetics</i> , 2015, 88, 34-40.	2.0	79
66	Mutations in <i>LAMA1</i> Cause Cerebellar Dysplasia and Cysts with and without Retinal Dystrophy. <i>American Journal of Human Genetics</i> , 2014, 95, 227-234.	6.2	92