Rachel Thompson

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/5692744/publications.pdf

Version: 2024-02-01

29 papers	2,266 citations	430442 18 h-index	433756 31 g-index
32 all docs	32 docs citations	32 times ranked	5332 citing authors

#	Article	IF	Citations
1	Congenital myasthenic syndrome: Correlation between clinical features and molecular diagnosis. European Journal of Neurology, 2022, 29, 833-842.	1.7	14
2	The RDâ€Connect Genomeâ€Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. Human Mutation, 2022, , .	1.1	18
3	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. European Journal of Human Genetics, 2021, 29, 1337-1347.	1.4	34
4	Severe neurodevelopmental disease caused by a homozygous TLK2 variant. European Journal of Human Genetics, 2020, 28, 383-387.	1.4	6
5	A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. Orphanet Journal of Rare Diseases, 2020, 15, 206.	1.2	21
6	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. Lancet Neurology, The, 2020, 19, 522-532.	4.9	36
7	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. Journal of Neuromuscular Diseases, 2020, 7, 153-166.	1.1	18
8	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. Journal of Molecular Diagnostics, 2020, 22, 1205-1215.	1.2	14
9	Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. European Journal of Epidemiology, 2020, 35, 643-653.	2.5	132
10	Increasing phenotypic annotation improves the diagnostic rate of exome sequencing in a rare neuromuscular disorder. Human Mutation, 2019, 40, 1797-1812.	1.1	22
11	Targeted therapies for congenital myasthenic syndromes: systematic review and steps towards a treatabolome. Emerging Topics in Life Sciences, 2019, 3, 19-37.	1.1	47
12	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. Nucleic Acids Research, 2019, 47, D1018-D1027.	6.5	539
13	RD-Connect, NeurOmics and EURenOmics: collaborative European initiative for rare diseases. European Journal of Human Genetics, 2018, 26, 778-785.	1.4	55
14	Recessive variants of MuSK are associated with late onset CMS and predominant limb girdle weakness. American Journal of Medical Genetics, Part A, 2018, 176, 1594-1601.	0.7	25
15	Intersection of Proteomics and Genomics to â€œSolve the Unsolvedâ€• in Rare Disorders such as Neurodegenerative and Neuromuscular Diseases. Proteomics - Clinical Applications, 2018, 12, 1700073.	0.8	33
16	A nomenclature and classification for the congenital myasthenic syndromes: preparing for FAIR data in the genomic era. Orphanet Journal of Rare Diseases, 2018, 13, 211.	1.2	17
17	The Human Phenotype Ontology in 2017. Nucleic Acids Research, 2017, 45, D865-D876.	6.5	699
18	Predictors of Health-Related Quality of Life in boys with Duchenne muscular dystrophy from six European countries. Journal of Neurology, 2017, 264, 709-723.	1.8	25

#	Article	IF	Citations
19	Critical points for an accurate human genome analysis. Human Mutation, 2017, 38, 912-921.	1.1	5
20	The UK Myotonic Dystrophy Patient Registry: facilitating and accelerating clinical research. Journal of Neurology, 2017, 264, 979-988.	1.8	23
21	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. Advances in Experimental Medicine and Biology, 2017, 1031, 55-94.	0.8	20
22	Natural History, Trial Readiness and Gene Discovery: Advances in Patient Registries for Neuromuscular Disease. Advances in Experimental Medicine and Biology, 2017, 1031, 97-124.	0.8	16
23	Linked Registries: Connecting Rare Diseases Patient Registries through a Semantic Web Layer. BioMed Research International, 2017, 2017, 1-13.	0.9	28
24	European Cross-Sectional Survey ofÂCurrent Care Practices for Duchenne Muscular Dystrophy Reveals Regional andÂAge-Dependent Differences. Journal of Neuromuscular Diseases, 2016, 3, 517-527.	1.1	55
25	Limb-girdle muscular dystrophies â€" international collaborations for translational research. Nature Reviews Neurology, 2016, 12, 294-309.	4.9	81
26	Overview of existing initiatives to develop and improve access and data sharing in rare disease registries and biobanks worldwide. Expert Opinion on Orphan Drugs, 2016, 4, 729-739.	0.5	6
27	The impact of integrated omics technologies for patients with rare diseases. Expert Opinion on Orphan Drugs, 2014, 2, 1211-1219.	0.5	5
28	Mapping the differences in care for 5,000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe. Journal of Neurology, 2014, 261, 152-163.	1.8	76
29	RD-Connect: An Integrated Platform Connecting Databases, Registries, Biobanks and Clinical Bioinformatics for Rare Disease Research. Journal of General Internal Medicine, 2014, 29, 780-787.	1.3	159