

# 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	The Human Phenotype Ontology in 2017. <i>Nucleic Acids Research</i> , 2017, 45, D865-D876.	6.5	699
2	Expansion of the Human Phenotype Ontology (HPO) knowledge base and resources. <i>Nucleic Acids Research</i> , 2019, 47, D1018-D1027.	6.5	539
3	RD-Connect: An Integrated Platform Connecting Databases, Registries, Biobanks and Clinical Bioinformatics for Rare Disease Research. <i>Journal of General Internal Medicine</i> , 2014, 29, 780-787.	1.3	159
4	Life expectancy at birth in Duchenne muscular dystrophy: a systematic review and meta-analysis. <i>European Journal of Epidemiology</i> , 2020, 35, 643-653.	2.5	132
5	Limb-girdle muscular dystrophies – international collaborations for translational research. <i>Nature Reviews Neurology</i> , 2016, 12, 294-309.	4.9	81
6	Mapping the differences in care for 5,000 Spinal Muscular Atrophy patients, a survey of 24 national registries in North America, Australasia and Europe. <i>Journal of Neurology</i> , 2014, 261, 152-163.	1.8	76
7	European Cross-Sectional Survey of Current Care Practices for Duchenne Muscular Dystrophy Reveals Regional and Age-Dependent Differences. <i>Journal of Neuromuscular Diseases</i> , 2016, 3, 517-527.	1.1	55
8	RD-Connect, NeurOmics and EURenOmics: collaborative European initiative for rare diseases. <i>European Journal of Human Genetics</i> , 2018, 26, 778-785.	1.4	55
9	Targeted therapies for congenital myasthenic syndromes: systematic review and steps towards a treatabolome. <i>Emerging Topics in Life Sciences</i> , 2019, 3, 19-37.	1.1	47
10	Advances in the diagnosis of inherited neuromuscular diseases and implications for therapy development. <i>Lancet Neurology</i> , The, 2020, 19, 522-532.	4.9	36
11	Solving patients with rare diseases through programmatic reanalysis of genome-phenome data. <i>European Journal of Human Genetics</i> , 2021, 29, 1337-1347.	1.4	34
12	Intersection of Proteomics and Genomics to “Solve the Unsolved” in Rare Disorders such as Neurodegenerative and Neuromuscular Diseases. <i>Proteomics - Clinical Applications</i> , 2018, 12, 1700073.	0.8	33
13	Linked Registries: Connecting Rare Diseases Patient Registries through a Semantic Web Layer. <i>BioMed Research International</i> , 2017, 2017, 1-13.	0.9	28
14	Predictors of Health-Related Quality of Life in boys with Duchenne muscular dystrophy from six European countries. <i>Journal of Neurology</i> , 2017, 264, 709-723.	1.8	25
15	Recessive variants of <i>MUSK</i> are associated with late onset CMS and predominant limb girdle weakness. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1594-1601.	0.7	25
16	The UK Myotonic Dystrophy Patient Registry: facilitating and accelerating clinical research. <i>Journal of Neurology</i> , 2017, 264, 979-988.	1.8	23
17	Increasing phenotypic annotation improves the diagnostic rate of exome sequencing in a rare neuromuscular disorder. <i>Human Mutation</i> , 2019, 40, 1797-1812.	1.1	22
18	A guide to writing systematic reviews of rare disease treatments to generate FAIR-compliant datasets: building a Treatabolome. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 206.	1.2	21

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19	Improved Diagnosis and Care for Rare Diseases through Implementation of Precision Public Health Framework. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 55-94.	0.8	20
20	Improved Criteria for the Classification of Titin Variants in Inherited Skeletal Myopathies. <i>Journal of Neuromuscular Diseases</i> , 2020, 7, 153-166.	1.1	18
21	The RDConnect GenomePhenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. <i>Human Mutation</i> , 2022, , .	1.1	18
22	A nomenclature and classification for the congenital myasthenic syndromes: preparing for FAIR data in the genomic era. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 211.	1.2	17
23	Natural History, Trial Readiness and Gene Discovery: Advances in Patient Registries for Neuromuscular Disease. <i>Advances in Experimental Medicine and Biology</i> , 2017, 1031, 97-124.	0.8	16
24	Improved Diagnosis of Rare Disease Patients through Systematic Detection of Runs of Homozygosity. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1205-1215.	1.2	14
25	Congenital myasthenic syndrome: Correlation between clinical features and molecular diagnosis. <i>European Journal of Neurology</i> , 2022, 29, 833-842.	1.7	14
26	Overview of existing initiatives to develop and improve access and data sharing in rare disease registries and biobanks worldwide. <i>Expert Opinion on Orphan Drugs</i> , 2016, 4, 729-739.	0.5	6
27	Severe neurodevelopmental disease caused by a homozygous TLK2 variant. <i>European Journal of Human Genetics</i> , 2020, 28, 383-387.	1.4	6
28	The impact of integrated omics technologies for patients with rare diseases. <i>Expert Opinion on Orphan Drugs</i> , 2014, 2, 1211-1219.	0.5	5
29	Critical points for an accurate human genome analysis. <i>Human Mutation</i> , 2017, 38, 912-921.	1.1	5