

Anna Hammarsjö

List of Publications by Year in descending order

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Version: 2024-02-01

15
papers

587
citations

840119

11
h-index

996533

15
g-index

15
all docs

15
docs citations

15
times ranked

1279
citing authors

#	ARTICLE	IF	CITATIONS
1	Expanding the mutation and phenotype spectrum of MYH3-associated skeletal disorders. <i>Npj Genomic Medicine</i> , 2022, 7, 11.	1.7	7
2	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. <i>Genome Medicine</i> , 2021, 13, 40.	3.6	116
3	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. <i>Journal of Human Genetics</i> , 2021, 66, 995-1008.	1.1	19
4	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for non-oncologic disorders. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 517-527.	0.7	3
5	Absence of GP130 cytokine receptor signaling causes extended StÅ¼ve-Wiedemann syndrome. <i>Journal of Experimental Medicine</i> , 2020, 217, .	4.2	41
6	Skeletal ciliopathies: a pattern recognition approach. <i>Japanese Journal of Radiology</i> , 2020, 38, 193-206.	1.0	30
7	From cytogenetics to cytogenomics: whole-genome sequencing as a first-line test comprehensively captures the diverse spectrum of disease-causing genetic variation underlying intellectual disability. <i>Genome Medicine</i> , 2019, 11, 68.	3.6	88
8	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. <i>Nature Medicine</i> , 2019, 25, 583-590.	15.2	86
9	Expanding the Clinical Spectrum of Phenotypes Caused by Pathogenic Variants in <i>PLOD2</i> . <i>Journal of Bone and Mineral Research</i> , 2018, 33, 753-760.	3.1	20
10	A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. <i>American Journal of Human Genetics</i> , 2018, 103, 553-567.	2.6	58
11	<i>Alu-Alu</i> mediated intragenic duplications in <i>IFT81</i> and <i>MATN3</i> are associated with skeletal dysplasias. <i>Human Mutation</i> , 2018, 39, 1456-1467.	1.1	16
12	Genotype-Phenotype Correlation of PLOD2 Skeletal Dysplasias Using Structural Information. <i>Journal of Bone and Mineral Research</i> , 2018, 33, 1377-1378.	3.1	2
13	Pathogenic variant in the <i>COL2A1</i> gene is associated with Spondyloepiphyseal dysplasia type Stanescu. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 266-269.	0.7	2
14	Extending the phenotype of BMPER-related skeletal dysplasias to ischiopinal dysostosis. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 1.	1.2	70
15	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbachâ€Nishimura skeletal dysplasia due to pathogenic variants in ALC9. <i>European Journal of Human Genetics</i> , 2016, 24, 198-207.	1.4	29