Anna HammarsjÃ¶

List of Publications by Year
in descending order

15 papers	587 citations	840776 11 h-index	996975 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. Npj Genomic Medicine, 2022, 7, 11.	3.8	7
2	Integration of whole genome sequencing into a healthcare setting: high diagnostic rates across multiple clinical entities in 3219 rare disease patients. Genome Medicine, 2021, 13, 40.	8.2	116
3	High diagnostic yield in skeletal ciliopathies using massively parallel genome sequencing, structural variant screening and RNA analyses. Journal of Human Genetics, 2021, 66, 995-1008.	2.3	19
4	Chondrodysplasia and growth failure in children after early hematopoietic stem cell transplantation for nonâ€oncologic disorders. American Journal of Medical Genetics, Part A, 2021, 185, 517-527.	1.2	3
5	Absence of GP130 cytokine receptor signaling causes extended StÃ¼ve-Wiedemann syndrome. Journal of Experimental Medicine, 2020, 217, .	8.5	41
6	Skeletal ciliopathies: a pattern recognition approach. Japanese Journal of Radiology, 2020, 38, 193-206.	2.4	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. Genome Medicine, 2019, 11, 68.	8.2	88
8	Gain-of-function mutation of microRNA-140 in human skeletal dysplasia. Nature Medicine, 2019, 25, 583-590.	30.7	86
9	Expanding the Clinical Spectrum of Phenotypes Caused by Pathogenic Variants in PLOD2. Journal of Bone and Mineral Research, 2018, 33, 753-760.	2.8	20
10	A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation. American Journal of Human Genetics, 2018, 103, 553-567.	6.2	58
11	Alu-Alu mediated intragenic duplications in IFT81 and MATN3 are associated with skeletal dysplasias. Human Mutation, 2018, 39, 1456-1467.	2.5	16
12	Genotype-Phenotype Correlation of PLOD2 Skeletal Dysplasias Using Structural Information. Journal of Bone and Mineral Research, 2018, 33, 1377-1378.	2.8	2
13	Pathogenenic variant in the COL2A1 gene is associated with Spondyloepiphyseal dysplasia type Stanescu. American Journal of Medical Genetics, Part A, 2016, 170, 266-269.	1.2	2
14	Extending the phenotype of BMPER-related skeletal dysplasias to ischiospinal dysostosis. Orphanet Journal of Rare Diseases, 2016, 11, 1.	2.7	70
15	A novel phenotype in N-glycosylation disorders: Gillessen-Kaesbachâ€“Nishimura skeletal dysplasia due to pathogenic variants in ALG9. European Journal of Human Genetics, 2016, 24, 198-207.	2.8	29