Cecilie F Rustad

List of Publications by Year
in descending order

14	754	840776 11	1058476 14
papers	citations	h-index	g-index

15	15	15	1892
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	6.2	19
2	Positive response to imatinib in <scp>PDGFRB</scp>â€related Kosaki overgrowth syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 2597-2601.	1.2	1
3	High prevalence of symptomatic spinal stenosis in Norwegian adults with achondroplasia: a population-based study. Orphanet Journal of Rare Diseases, 2020, 15, 123.	2.7	25
4	PAPSS2â€related brachyolmia: Clinical and radiological phenotype in 18 new cases. American Journal of Medical Genetics, Part A, 2019, 179, 1884-1894.	1.2	9
5	De novo substitutions of TRPM3 cause intellectual disability and epilepsy. European Journal of Human Genetics, 2019, 27, 1611-1618.	2.8	45
6	The Discovery of a LEMD2-Associated Nuclear Envelopathy with Early Progeroid Appearance Suggests Advanced Applications for AI-Driven Facial Phenotyping. American Journal of Human Genetics, 2019, 104, 749-757.	6.2	41
7	Nordic Guidelines for Germline Predisposition to Myeloid Neoplasms in Adults: Recommendations for Genetic Diagnosis, Clinical Management and Follow-up. HemaSphere, 2019, 3, e321.	2.7	51
8	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
9	Clinical, neuroradiological, and biochemical features of SLC35A2â€CDG patients. Journal of Inherited Metabolic Disease, 2019, 42, 553-564.	3.6	32
10	Heterozygous mutations affecting the protein kinase domain of CDK13 cause a syndromic form of developmental delay and intellectual disability. Journal of Medical Genetics, 2018, 55, 28-38.	3.2	36
11	Neurofibromatosis type 2: Multiple intraâ€dermal tumors in a toddler. American Journal of Medical Genetics, Part A, 2017, 173, 1447-1449.	1.2	2
12	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	2.9	261
13	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. PLoS ONE, 2016, 11, e0150555.	2.5	32
14	PGM3 Mutations Cause a Congenital Disorder of Glycosylation with Severe Immunodeficiency and Skeletal Dysplasia. American Journal of Human Genetics, 2014, 95, 96-107.	6.2	148