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	Primary immunodeficiency diseases: Genomic approaches delineate heterogeneous Mendelian disorders. Journal of Allergy and Clinical Immunology, 2017, 139, 232-245.	2.9	261
2	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
3	Clinical exome sequencing reveals locus heterogeneity and phenotypic variability of cohesinopathies. Genetics in Medicine, 2019, 21, 663-675.	2.4	52
4	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
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	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
8	Clinical, neuroradiological, and biochemical features of SLC35A2â€CDG patients. Journal of Inherited Metabolic Disease, 2019, 42, 553-564.	3.6	32
9	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. PLoS ONE, 2016, 11, e0150555.	2.5	32
10	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
11	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
12	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
13	Neurofibromatosis type 2: Multiple intraâ€dermal tumors in a toddler. American Journal of Medical Genetics, Part A, 2017, 173, 1447-1449.	1.2	2
14	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