

List of Publications by Year in descending order

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8
papers

109
citations

1684188

5
h-index

1588992

8
g-index

8
all docs

8
docs citations

8
times ranked

227
citing authors

#	ARTICLE	IF	CITATIONS
1	High prevalence of variants in skeletal dysplasia associated genes in individuals with short stature and minor skeletal anomalies. <i>European Journal of Endocrinology</i> , 2021, 185, 691-705.	3.7	13
2	Spondyloepiphyseal dysplasia type Stanescu: Expanding the clinical and molecular spectrum of a very rare type II collagenopathy. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2715-2721.	1.2	2
3	Genomic sequencing highlights the diverse molecular causes of Perrault syndrome: a peroxisomal disorder (PEX6), metabolic disorders (CLPP, GGPS1), and mtDNA maintenance/translation disorders (LARS2, TFAM). <i>Human Genetics</i> , 2020, 139, 1325-1343.	3.8	21
4	Upper limb phocomelia: A prenatal case of thrombocytopenia-absent radius (TAR) syndrome illustrating the importance of chromosomal microarray in limb reduction defects. <i>Taiwanese Journal of Obstetrics and Gynecology</i> , 2020, 59, 318-322.	1.3	5
5	Clinical and Molecular Description of 16 Families With Heterozygous <i>IHH</i> Variants. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2020, 105, 2654-2666.	3.6	6
6	Reporting and methodological quality of clinical trials on exercise therapy for Parkinson's disease. <i>Parkinsonism and Related Disorders</i> , 2019, 69, 150-156.	2.2	5
7	Prenatal diagnosis of holoprosencephaly associated with Smith-Lemli-Opitz syndrome (SLOS) in a 46,XX fetus. <i>Taiwanese Journal of Obstetrics and Gynecology</i> , 2017, 56, 541-544.	1.3	7
8	Fifteen Years of Clinical Trials in Huntington's Disease: A Very Low Clinical Drug Development Success Rate. <i>Journal of Huntington's Disease</i> , 2017, 6, 157-163.	1.9	50