Sarah F Smithson

List of Publications by Year
in descending order

14	134	1307594 7	1281871 11
papers	citations	h-index	g-index

15	15	15	239
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	TRPV6 compound heterozygous variants result in impaired placental calcium transport and severe undermineralization and dysplasia of the fetal skeleton. American Journal of Medical Genetics, Part A, 2018, 176, 1950-1955.	1.2	31
2	Deep phenotyping of 14 new patients with IQSEC2 variants, including monozygotic twins of discordant phenotype. Clinical Genetics, 2019, 95, 496-506.	2.0	20
3	Heterozygous ANKRD17 loss-of-function variants cause a syndrome with intellectual disability, speech delay, and dysmorphism. American Journal of Human Genetics, 2021, 108, 1138-1150.	6.2	17
4	Recurrent de novo missense variants across multiple histone H4 genes underlie a neurodevelopmental syndrome. American Journal of Human Genetics, 2022, 109, 750-758.	6.2	13
5	Disruption ofTWIST1translation by 5â€² UTR variants in Saethre-Chotzen syndrome. Human Mutation, 2018, 39, 1360-1365.	2.5	10
6	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
7	Post-mortem histology in transient receptor potential cation channel subfamily V member 6 (TRPV6) under-mineralising skeletal dysplasia suggests postnatal skeletal recovery: a case report. BMC Medical Genetics, 2020, 21, 64.	2.1	9
8	The third case of TNFRSF11A-associated dysosteosclerosis with a mutation producing elongating proteins. Journal of Human Genetics, 2021, 66, 371-377.	2.3	8
9	<scp>AIFM1</scp>â€associated Xâ€linked spondylometaphyseal dysplasia with cerebral hypomyelination. American Journal of Medical Genetics, Part A, 2021, 185, 1228-1235.	1.2	5
10	Challenges in long-term control of hypercalcaemia with denosumab after haematopoietic stem cell transplantation for TNFRSF11A osteoclast-poor autosomal recessive osteopetrosis. Bone Reports, 2021, 14, 100738.	0.4	4
11	Expanding the phenotypic spectrum of IFT81 : Associated ciliopathy syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2403-2408.	1.2	3
12	Non-collagen pathogenic variants resulting in the osteogenesis imperfecta phenotype in children: a single-country observational cohort study. Archives of Disease in Childhood, 2022, 107, 486-490.	1.9	2
13	Prenatal diagnosis and postnatal outcome of massive abdominal aortic aneurysmsâ€”a case report. Prenatal Diagnosis, 2015, 35, 923-925.	2.3	0
14	Cover Image, Volume 176A, Number 9, September 2018. American Journal of Medical Genetics, Part A, 2018, 176, .	1.2	0