Smrithi Salian

List of Publications by Year
in descending order

14	96	1478505 6	1474206 9
papers	citations	h-index	g-index

14	14	14	172
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Additional three patients with Smithâ€McCort dysplasia due to novel RAB33B mutations. American Journal of Medical Genetics, Part A, 2017, 173, 588-595.	1.2	17
2	A variant of neonatal progeroid syndrome, or Wiedemannâ€“Rautenstrauch syndrome, is associated with a nonsense variant in POLR3GL. European Journal of Human Genetics, 2020, 28, 461-468.	2.8	16
3	De Novo KAT5 Variants Cause a Syndrome with Recognizable Facial Dysmorphisms, Cerebellar Atrophy, Sleep Disturbance, and Epilepsy. American Journal of Human Genetics, 2020, 107, 564-574.	6.2	14
4	Seven additional families with spondylocarpotarsal synostosis syndrome with novel biallelic deleterious variants in FLNB. Clinical Genetics, 2018, 94, 159-164.	2.0	10
5	Variable presentation of Fraser syndrome in two fetuses and a novel mutation in FRAS1. Congenital Anomalies (discontinued), 2017, 57, 83-85.	0.6	8
6	Familial 7q11.23 duplication with variable phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 2727-2730.	1.2	6
7	Pycnodysostosis: Novel Variants in CTSK and Occurrence of Giant Cell Tumor. Journal of Pediatric Genetics, 2018, 07, 009-013.	0.7	6
8	Epileptic encephalopathy caused by <scp>ARV1</scp> deficiency: Refinement of the genotypeâ€“phenotype spectrum and functional impact on <scp>GPI</scp>â€anchored proteins. Clinical Genetics, 2021, 100, 607-614.	2.0	6
9	Severe Form of Brachydactyly Type A1 in a Child with a c.298Gâ€‰>â€‰A Mutation in IHH Gene. Journal of Pediatric Genetics, 2017, 06, 177-180.	0.7	5
10	Further evidence for causation of ischiospinal dysostosis by a pathogenic variant in BMPER and expansion of the phenotype. Congenital Anomalies (discontinued), 2019, 59, 26-27.	0.6	3
11	Focal dermal hypoplasia with a de novo mutation p.e300FNx01 of porcn gene in a male infant. Indian Journal of Dermatology, 2016, 61, 700.	0.3	3
12	PIGF deficiency causes a phenotype overlapping with DOORS syndrome. Human Genetics, 2021, 140, 879-884.	3.8	2
13	Cover Image, Volume 173A, Number 3, March 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
14	Novel ALOX12B mutation identified in parents following single nucleotide polymorphism microarray testing of banked DNA from a fatal case of congenital ichthyosis. Indian Journal of Dermatology, 2016, 61, 122.	0.3	0