Ashish Marwaha

List of Publications by Year
in descending order

13 papers	409 citations	1478505 6 h-index	1199594 12 g-index
13 all docs	13 docs citations	13 times ranked	792 citing authors

#	Article	IF	Citations
1	The utility of <scp>DNA</scp> methylation signatures in directing genome sequencing workflow: Kabuki syndrome and <scp>CDK13</scp>â€related disorder. American Journal of Medical Genetics, Part A, 2022, 188, 1368-1375.	1.2	5
2	Expansion of the neurodevelopmental phenotypic spectrum of <scp><i>CKAP2L</i></scp>â€related Filippi syndrome to include an adolescent male with normal intellect. American Journal of Medical Genetics, Part A, 2022, 188, 1928-1929.	1.2	0
3	Two cases of carbonic anhydrase <scp>VA</scp> deficiencyâ€"An ultrarare metabolic decompensation syndrome presenting with hyperammonemia, lactic acidosis, ketonuria, and good clinical outcome. JIMD Reports, 2021, 57, 9-14.	1.5	7
4	Progressive decline of T and B cell numbers and function in a patient with CDC42 deficiency. Immunologic Research, 2021, 69, 53-58.	2.9	5
5	<scp>The pointâ€ofâ€care</scp> use of a facial phenotyping tool in the genetics clinic: Enhancing diagnosis and education with machine learning. American Journal of Medical Genetics, Part A, 2021, 185, 1151-1158.	1.2	18
6	Phase II multicentre, double-blind, randomised trial of ustekinumab in adolescents with new-onset type 1 diabetes (USTEK1D): trial protocol. BMJ Open, 2021, 11, e049595.	1.9	2
7	Epidermal growth factor receptor deficiency: Expanding the phenotype beyond infancy. Journal of Dermatology, 2020, 47, 898-902.	1.2	3
8	Congenital hypothyroidism, cardiac defects, and pancreatic agenesis in an infant with <scp>GATA6</scp> mutation. American Journal of Medical Genetics, Part A, 2020, 182, 1496-1499.	1.2	8
9	Genotype-phenotype data from a case series of patients with mosaic neurofibromatosis type 1. British Journal of Dermatology, 2018, 179, 1216-1217.	1.5	6
10	Pre-diagnostic genotyping identifies T1D subjects with impaired Treg IL-2 signaling and an elevated proportion of FOXP3+IL-17+ cells. Genes and Immunity, 2017, 18, 15-21.	4.1	23
11	Targeting the IL-17/IFN-Î³ axis as a potential new clinical therapy for type 1 diabetes. Clinical Immunology, 2014, 154, 84-89.	3.2	40
12	TH17 Cells in Autoimmunity and Immunodeficiency: Protective or Pathogenic?. Frontiers in Immunology, 2012, 3, 129.	4.8	102
13	Cutting Edge: Increased IL-17â€"Secreting T Cells in Children with New-Onset Type 1 Diabetes. Journal of Immunology, 2010, 185, 3814-3818.	0.8	190