Resham Ejaz

List of Publications by Year
in descending order

10 papers	374 citations	1684188 5 h-index	1720034 7 g-index
10 all docs	10 docs citations	10 times ranked	1242 citing authors

#	Article	IF	Citations
1	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
2	Lateral meningocele (Lehman) syndrome: A child with a novel NOTCH3 mutation. American Journal of Medical Genetics, Part A, 2016, 170, 1070-1075.	1.2	19
3	Riboflavin transporter deficiency mimicking mitochondrial myopathy caused by complex II deficiency. American Journal of Medical Genetics, Part A, 2018, 176, 399-403.	1.2	18
4	De novo pathogenic variant in TUBB2A presenting with arthrogryposis multiplex congenita, brain abnormalities, and severe developmental delay. , 2017, 173, 2725-2730.		15
5	Stable transmission of an unbalanced chromosome 21 derived from chromoanasynthesis in a patient with a SYNGAP1 likely pathogenic variant. Molecular Cytogenetics, 2018, 11, 50.	0.9	11
6	Primary mediastinal paraganglioma associated with a familial variant in the succinate dehydrogenase B subunit gene. Journal of Surgical Oncology, 2018, 117, 160-162.	1.7	6
7	Child Neurology: Diencephalic syndromeâ€“like presentation of a cervicomedullary brainstem tumor. Neurology, 2016, 87, e248-e251.	1.1	5
8	Impact of Mobility Device Use on Quality of Life in Children With Friedreich Ataxia. Journal of Child Neurology, 2018, 33, 397-404.	1.4	5
9	MG-119â€...The evolving features of nicolaides-baraitser syndrome â€“ a case report of a twenty-years follow-up. Journal of Medical Genetics, 2015, 52, A5.1-A5.	3.2	0
10	Urgent Management of Bleeding in Immune Thrombocytopenia: Towards a Standardized Protocol in the Emergency Department. Blood, 2018, 132, 3517-3517.	1.4	0