

Saleem Ahmed

List of Publications by Year in descending order

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Version: 2024-02-01

8
papers

207
citations

1478505

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1720034

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docs citations

8
times ranked

623
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. <i>Nature Genetics</i> , 2017, 49, 537-549.	21.4	81
2	Exome analysis identified a novel missense mutation in the CLPP gene in a consanguineous Saudi family expanding the clinical spectrum of Perrault Syndrome type-3. <i>Journal of the Neurological Sciences</i> , 2015, 353, 149-154.	0.6	37
3	Novel nonsense mutation in the PTRF gene underlies congenital generalized lipodystrophy in a consanguineous Saudi family. <i>European Journal of Medical Genetics</i> , 2015, 58, 216-221.	1.3	31
4	A missense mutation in TRAPPC6A leads to build-up of the protein, in patients with a neurodevelopmental syndrome and dysmorphic features. <i>Scientific Reports</i> , 2018, 8, 2053.	3.3	30
5	The alkylglycerol monooxygenase (AGMO) gene previously involved in autism also causes a novel syndromic form of primary microcephaly in a consanguineous Saudi family. <i>Journal of the Neurological Sciences</i> , 2016, 363, 240-244.	0.6	18
6	Femoralâ€“facial syndrome in an infant of a diabetic mother. <i>BMJ Case Reports</i> , 2015, 2015, bcr2014208857.	0.5	7
7	Identification of Two Homozygous Sequence Variants in the <i>COL7A1</i> Gene Underlying Dystrophic Epidermolysis Bullosa by Wholeâ€“Exome Analysis in a Consanguineous Family. <i>Annals of Human Genetics</i> , 2015, 79, 350-356.	0.8	3
8	Cenani-Lenz syndrome-like limb anomaly with more severe involvement of left side. <i>BMJ Case Reports</i> , 2012, 2012, bcr0120125634-bcr0120125634.	0.5	0