Malak Ali Alghamdi

List of Publications by Year
in descending order

19	404	1162889 8	839398 18
papers	citations	h-index	g-index

19	19	19	998
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. American Journal of Human Genetics, 2019, 104, 1182-1201.	2.6	184
2	JAK Inhibitor Therapy in a Child with Inherited USP18 Deficiency. New England Journal of Medicine, 2020, 382, 256-265.	13.9	69
3	Combining exome/genome sequencing with data repository analysis reveals novel geneâ€“disease associations for a wide range of genetic disorders. Genetics in Medicine, 2021, 23, 1551-1568.	1.1	30
4	Phenotypic and molecular spectrum of pyridoxamineâ€5â€²â€phosphate oxidase deficiency: A scoping review of 87 cases of pyridoxamineâ€5â€²â€phosphate oxidase deficiency. Clinical Genetics, 2021, 99, 99-110.	1.0	25
5	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. Frontiers in Pediatrics, 2021, 9, 633385.	0.9	15
6	Effect of common medications on the expression of SARS-CoV-2 entry receptors in liver tissue. Archives of Toxicology, 2020, 94, 4037-4041.	1.9	12
7	Expanding the phenotype and the genotype of Stromme syndrome: A novel variant of the CENPF gene and literature review. European Journal of Medical Genetics, 2020, 63, 103844.	0.7	12
8	A Novel Biallelic STING1 Gene Variant Causing SAVI in Two Siblings. Frontiers in Immunology, 2020, 11, 599564.	2.2	12
9	Diversity of Phenotype and Genetic Etiology of 23 Cystinuria Saudi Patients: A Retrospective Study. Frontiers in Pediatrics, 2020, 8, 569389.	0.9	9
10	Unusual Prominent Pulmonary Involvement in a Homozygous PRF1 Gene Variant in a Female Patient. Journal of Clinical Immunology, 2021, 41, 217-220.	2.0	6
11	Molecular autopsy by proxy in preconception counseling. Clinical Genetics, 2021, 100, 678-691.	1.0	6
12	A Novel TBX1 Variant Causing Hypoparathyroidism and Deafness. Journal of the Endocrine Society, 2020, 4, bvz028.	0.1	5
13	Biallelic lossâ€ofâ€function HACD1 variants are a bona fide cause of congenital myopathy. Clinical Genetics, 2021, 99, 513-518.	1.0	5
14	Clinical and Genetic Characterization of Craniosynostosis in Saudi Arabia. Frontiers in Pediatrics, 2021, 9, 582816.	0.9	5
15	A disorder clinically resembling cystic fibrosis caused by biallelic variants in the AGR2 gene. Journal of Medical Genetics, 2022, 59, 993-1001.	1.5	5
16	Pyridox(am)ine 5â€²-Phosphate Oxidase Deficiency: Severe Prenatal Presentation with Hypoxic Ischemic Encephalopathy. Journal of Pediatric Epilepsy, 2019, 08, 049-055.	0.1	2
17	Heterotopic respiratory mucosa in the scalp overlying abnormal bony island in the skull linked to maternal misoprostol use, literature review and surgical experience. International Journal of Surgery Case Reports, 2019, 59, 115-119.	0.2	1
18	Succinic semialdehyde dehydrogenase deficiency presenting with central hypothyroidism. Clinical Case Reports (discontinued), 2021, 9, 229-235.	0.2	1

#	Article	IF	Citations
19	Implications of mosaicism in variant interpretation: A case of a de novo homozygous NF1 variant. European Journal of Medical Genetics, 2021, 64, 104236.	0.7	0