Wafaa Eyaid

List of Publications by Year
in descending order

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Version: 2024-02-01

26 papers	1,185 citations	623734 14 h-index	526287 27 g-index
29 all docs	29 docs citations	29 times ranked	2903 citing authors

#	Article	IF	Citations
1	Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort. European Journal of Human Genetics, 2021, 29, 141-153.	2.8	66
2	A recurrent, homozygous EMC10 frameshift variant is associated with a syndrome of developmental delay with variable seizures and dysmorphic features. Genetics in Medicine, 2021, 23, 1158-1162.	2.4	13
3	Loss of C2orf69 defines a fatal autoinflammatory syndrome in humans and zebrafish that evokes a glycogen-storage-associated mitochondriopathy. American Journal of Human Genetics, 2021, 108, 1301-1317.	6.2	11
4	Long-term effectiveness of carglumic acid in patients with propionic acidemia (PA) and methylmalonic acidemia (MMA): a randomized clinical trial. Orphanet Journal of Rare Diseases, 2021, 16, 422.	2.7	12
5	Phenotypic delineation of the retinal arterial macroaneurysms with supravalvular pulmonic stenosis syndrome. Clinical Genetics, 2020, 97, 447-456.	2.0	7
6	What is the right sequencing approach? Solo VS extended family analysis in consanguineous populations. BMC Medical Genomics, 2020, 13, 103.	1.5	10
7	NCKAP1L defects lead to a novel syndrome combining immunodeficiency, lymphoproliferation, and hyperinflammation. Journal of Experimental Medicine, 2020, 217, .	8.5	48
8	MEFV c.2230G>T p.(Ala744Ser) rs61732874 previously misclassified as pathogenic variant due to lack of a population specific database. Annals of Human Genetics, 2020, 84, 370-379.	0.8	1
9	<scp>PDCD6IP</scp>, encoding a regulator of the <scp>ESCRT</scp> complex, is mutated in microcephaly. Clinical Genetics, 2020, 98, 80-85.	2.0	11
10	Evaluation of long-term effectiveness of the use of carglumic acid in patients with propionic acidemia (PA) or methylmalonic acidemia (MMA): study protocol for a randomized controlled trial. BMC Pediatrics, 2019, 19, 195.	1.7	9
11	Phenotypic and biochemical analysis of an international cohort of individuals with variants in NAA10 and NAA15. Human Molecular Genetics, 2019, 28, 2900-2919.	2.9	46
12	6-Pyruvoyltetrahydropterin Synthase Deficiency: Review and Report of 28 Arab Subjects. Pediatric Neurology, 2019, 96, 40-47.	2.1	12
13	A new association between CDK5RAP2 microcephaly and congenital cataracts. Annals of Human Genetics, 2018, 82, 165-170.	0.8	9
14	Whole-genome sequencing offers additional but limited clinical utility compared with reanalysis of whole-exome sequencing. Genetics in Medicine, 2018, 20, 1328-1333.	2.4	118
15	Molecular autopsy in maternalâ€“fetal medicine. Genetics in Medicine, 2018, 20, 420-427.	2.4	84
16	Phenotypic and Molecular Spectrum of Aicardi-GoutiÃ¨res Syndrome: A Study of 24 Patients. Pediatric Neurology, 2018, 78, 35-40.	2.1	40
17	A multicenter clinical exome study in unselected cohorts from a consanguineous population of Saudi Arabia demonstrated a high diagnostic yield. Molecular Genetics and Metabolism, 2017, 121, 91-95.	1.1	68
18	Expanded Newborn Screening Program in Saudi Arabia: Incidence of screened disorders. Journal of Paediatrics and Child Health, 2017, 53, 585-591.	0.8	83

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19	Clinical, Biochemical, and Molecular Features in 37 Saudi Patients with Very Long Chain Acyl CoA Dehydrogenase Deficiency. JIMD Reports, 2017, 40, 47-53.	1.5	15
20	Early onset of Fazio-Londe syndrome: the first case report from the Arabian Peninsula. Human Genome Variation, 2017, 4, 17018.	0.7	6
21	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. European Journal of Human Genetics, 2017, 25, 176-182.	2.8	291
22	Thirteen year retrospective review of the spectrum of inborn errors of metabolism presenting in a tertiary center in Saudi Arabia. Orphanet Journal of Rare Diseases, 2016, 11, 126.	2.7	61
23	Clinical, biochemical, cellular and molecular characterization of mitochondrial DNA depletion syndrome due to novel mutations in the MPV17 gene. European Journal of Human Genetics, 2014, 22, 184-191.	2.8	52
24	Clinical and Molecular Characteristics of Mitochondrial DNA Depletion Syndrome Associated with Neonatal Cholestasis and Liver Failure. Journal of Pediatrics, 2014, 164, 553-559.e2.	1.8	44
25	Transaldolase deficiency: report of 12 new cases and further delineation of the phenotype. Journal of Inherited Metabolic Disease, 2013, 36, 997-1004.	3.6	36
26	A novel homozygous missense mutation (c.610G>A, p.Gly204Ser) in the WNT7A gene causes tetraâ€amelia in two Saudi families. American Journal of Medical Genetics, Part A, 2011, 155, 599-604.	1.2	27