Wafaa Eyaid

List of Publications by Year
in descending order

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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	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. European Journal of Human Genetics, 2017, 25, 176-182.	2.8	291
2	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
3	Molecular autopsy in maternalâ€“fetal medicine. Genetics in Medicine, 2018, 20, 420-427.	2.4	84
4	Expanded Newborn Screening Program in Saudi Arabia: Incidence of screened disorders. Journal of Paediatrics and Child Health, 2017, 53, 585-591.	0.8	83
5	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
6	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
7	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
8	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
9	NCKAP1L defects lead to a novel syndrome combining immunodeficiency, lymphoproliferation, and hyperinflammation. Journal of Experimental Medicine, 2020, 217, .	8.5	48
10	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
11	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
12	Phenotypic and Molecular Spectrum of Aicardi-GoutiÃ¨res Syndrome: A Study of 24 Patients. Pediatric Neurology, 2018, 78, 35-40.	2.1	40
13	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
14	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
15	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
16	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
17	6-Pyruvoyltetrahydropterin Synthase Deficiency: Review and Report of 28 Arab Subjects. Pediatric Neurology, 2019, 96, 40-47.	2.1	12
18	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

#	Article	IF	Citations
19	<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
20	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
21	What is the right sequencing approach? Solo VS extended family analysis in consanguineous populations. BMC Medical Genomics, 2020, 13, 103.	1.5	10
22	A new association between CDK5RAP2 microcephaly and congenital cataracts. Annals of Human Genetics, 2018, 82, 165-170.	0.8	9
23	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
24	Phenotypic delineation of the retinal arterial macroaneurysms with supravalvular pulmonic stenosis syndrome. Clinical Genetics, 2020, 97, 447-456.	2.0	7
25	Early onset of Fazio-Londe syndrome: the first case report from the Arabian Peninsula. Human Genome Variation, 2017, 4, 17018.	0.7	6
26	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