Farrah Rajabi

List of Publications by Year
in descending order

17 papers	88 citations	1684188 5 h-index	1474206 9 g-index
17 all docs	17 docs citations	17 times ranked	168 citing authors

#	Article	IF	Citations
1	Gainâ€ofâ€function variants in the ODC1 gene cause a syndromic neurodevelopmental disorder associated with macrocephaly, alopecia, dysmorphic features, and neuroimaging abnormalities. American Journal of Medical Genetics, Part A, 2018, 176, 2554-2560.	1.2	26
2	Genotypeâ€“phenotype correlations and novel molecular insights into the DHX30-associated neurodevelopmental disorders. Genome Medicine, 2021, 13, 90.	8.2	16
3	Pontocerebellar hypoplasia due to bi-allelic variants in MINPP1. European Journal of Human Genetics, 2021, 29, 411-421.	2.8	13
4	Semaphorin-Plexin Signaling: From Axonal Guidance to a New X-Linked Intellectual Disability Syndrome. Pediatric Neurology, 2022, 126, 65-73.	2.1	8
5	Phenylalanine hydroxylase genotype-phenotype associations in the United States: A single center study. Molecular Genetics and Metabolism, 2019, 128, 415-421.	1.1	7
6	Liver Failure as the Presentation of Ornithine Transcarbamylase Deficiency in a 13-Month-Old Female. JIMD Reports, 2017, 40, 17-22.	1.5	4
7	Imprinted genes in clinical exome sequencing: Review of 538 cases and exploration of mouse-human conservation in the identification of novel human disease loci. European Journal of Medical Genetics, 2020, 63, 103903.	1.3	4
8	Gravesâ€™ disease in a five-month-old boy with an unusual treatment course. Journal of Pediatric Endocrinology and Metabolism, 2021, 34, 401-406.	0.9	3
9	Genetic Study in a Cohort of Children With ROHHAD Syndrome. Journal of the Endocrine Society, 2021, 5, A503-A504.	0.2	2
10	New Innovations: Therapies for Genetic Conditions. Current Genetic Medicine Reports, 2014, 2, 113-123.	1.9	1
11	Hyperphenylalaninemia and the genomic revolution. Molecular Genetics and Metabolism, 2015, 114, 380-381.	1.1	1
12	Expansion and Implications of Newborn Screening. Current Genetic Medicine Reports, 2015, 3, 110-117.	1.9	1
13	A novel variant in the TSPAN12 gene-presenting as unilateral myopia, pediatric cataract, and heterochromia in a patient with familial exudative vitreoretinopathy. European Journal of Ophthalmology, 2022, 32, NP6-NP9.	1.3	1
14	Bilateral consecutive choroidal neovascularization in Best vitelliform macular dystrophy. Baylor University Medical Center Proceedings, 0, , 1-3.	0.5	1
15	Confounding factors in identification of disease-resilient individuals. Nature Biotechnology, 2016, 34, 1103-1104.	17.5	0
16	Acute Pancreatitis in a Patient with Maple Syrup Urine Disease: A Management Paradox. Journal of Pediatrics, 2018, 198, 313-316.	1.8	0
17	OR33-07 ARNT2: A Potential Novel Candidate Gene for Monogenic Obesity in Humans. Journal of the Endocrine Society, 2020, 4, .	0.2	0