Tzipora C Falik-Zaccai

List of Publications by Year
in descending order

31 papers	614 citations	687363 13 h-index	642732 23 g-index
34 all docs	34 docs citations	34 times ranked	1093 citing authors

#	Article	IF	Citations
1	Chronic Diarrhea and Juvenile Cataracts: Think Cerebrotendinous Xanthomatosis and Treat. Pediatrics, 2009, 123, 143-147.	2.1	78
2	Whole Exome Sequencing Reveals Mutations in Known Retinal Disease Genes in 33 out of 68 Israeli Families with Inherited Retinopathies. Scientific Reports, 2015, 5, 13187.	3.3	66
3	PNPO deficiency: An under diagnosed inborn error of pyridoxine metabolism. Molecular Genetics and Metabolism, 2008, 94, 431-434.	1.1	54
4	Pathogenic variants in glutamyl-tRNAGln amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. Nature Communications, 2018, 9, 4065.	12.8	44
5	De Novo Variants in MAPK8IP3 Cause Intellectual Disability with Variable Brain Anomalies. American Journal of Human Genetics, 2019, 104, 203-212.	6.2	44
6	Population screening in a Druze community: the challenge and the reward. Genetics in Medicine, 2008, 10, 903-909.	2.4	37
7	Two separate functions of NME3 critical for cell survival underlie a neurodegenerative disorder. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 566-574.	7.1	36
8	Newborn screening for cerebrotendinous xanthomatosis is the solution for early identification and treatment. Journal of Lipid Research, 2018, 59, 2214-2222.	4.2	28
9	Cockayne syndrome type II in a Druze isolate in Northern Israel in association with an insertion mutation in ERCC6. American Journal of Medical Genetics, Part A, 2008, 146A, 1423-1429.	1.2	25
10	Clinical utility gene card for: Xeroderma pigmentosum. European Journal of Human Genetics, 2014, 22, 953-953.	2.8	20
11	Sequence variation in <scp>PPP</scp> 1R13L results in a novel form of cardioâ€cutaneous syndrome. EMBO Molecular Medicine, 2017, 9, 319-336.	6.9	18
12	Microarray analysis in pregnancies with isolated echogenic bowel. Early Human Development, 2018, 119, 25-28.	1.8	18
13	Walker-Warburg syndrome and tectocerebellar dysraphia: A novel association caused by a homozygous DAG1 mutation. European Journal of Paediatric Neurology, 2018, 22, 525-531.	1.6	15
14	A clinically validated whole genome pipeline for structural variant detection and analysis. BMC Genomics, 2019, 20, 545.	2.8	15
15	Expert opinion on diagnosing, treating and managing patients with cerebrotendinous xanthomatosis (CTX): a modified Delphi study. Orphanet Journal of Rare Diseases, 2021, 16, 353.	2.7	13
16	A novel XPD mutation in a compound heterozygote; the mutation in the second allele is present in three homozygous patients with mild sun sensitivity. Environmental and Molecular Mutagenesis, 2012, 53, 505-514.	2.2	12
17	Sedaghatian-type spondylometaphyseal dysplasia: Whole exome sequencing in neonatal dry blood spots enabled identification of a novel variant in GPX4. European Journal of Medical Genetics, 2020, 63, 104020.	1.3	12
18	Mammalian Homologue NME3 of DYNAMO1 Regulates Peroxisome Division. International Journal of Molecular Sciences, 2020, 21, 8040.	4.1	11

#	Article	IF	Citations
19	Isolated fetal horseshoe kidney does not seem to increase the risk for abnormal chromosomal microarray results. European Journal of Obstetrics, Gynecology and Reproductive Biology, 2018, 222, 80-83.	1.1	9
20	<scp>COG6â€CDG</scp>: Expanding the phenotype with emphasis on glycosylation defects involved in the causation of male disorders of sex development. Clinical Genetics, 2020, 98, 402-407.	2.0	8
21	De novo <scp>STXBP1</scp> mutation in a child with developmental delay and spasticity reveals a major structural alteration in the interface with syntaxin <scp>1A</scp>. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 412-422.	1.7	7
22	Challenges to effective and autonomous genetic testing and counseling for ethno-cultural minorities: a qualitative study. BMC Medical Ethics, 2020, 21, 98.	2.4	6
23	Congenital Hypotonia: Cracking a SAGA of consanguineous kindred harboring four genetic variants. Molecular Genetics & Genomic Medicine, 2022, 10, e1849.	1.2	6
24	Genetic counseling of highâ€risk isolated populations: A worldwide challenge. Birth Defects Research, 2020, 112, 316-320.	1.5	4
25	The effect of polyhydramnios degree on chromosomal microarray results: a retrospective cohort analysis of 742 singleton pregnancies. Archives of Gynecology and Obstetrics, 2021, 304, 649-656.	1.7	4
26	The versatile DNA nucleotide excision repair (NER) and its medical significance. Pediatric Endocrinology Reviews, 2009, 7, 37-42.	1.2	4
27	NGLY1 Deficiency Zebrafish Model Manifests Abnormalities of the Nervous and Musculoskeletal Systems. Frontiers in Cell and Developmental Biology, 0, 10, .	3.7	3
28	â€˜LEADERSâ€™: A culturally tailored approach to genetic counseling for minority populations. Journal of Genetic Counseling, 2021, 30, 70-74.	1.6	2
29	Concomitant congenital CMV infection and inherited liver diseases. European Journal of Medical Genetics, 2021, 64, 104249.	1.3	2
30	A Puzzling â€œSwitchâ€•in Blood Type Following Blood Transfusion. Annals of Laboratory Medicine, 2017, 37, 293-295.	2.5	1
31	P2â€489: CHARACTERIZING COGNITIVE AND NEURAL ALTERATIONS AMONG HEALTHY PARTICIPANTS WITH A GENETIC PREDISPOSITION FOR ALZHEIMER'S DISEASE. Alzheimer's and Dementia, 2018, 14, P917.	0.8	0