Farah R Zahir

List of Publications by Year
in descending order

28 papers	1,519 citations	567281 15 h-index	580821 25 g-index
29 all docs	29 docs citations	29 times ranked	3286 citing authors

#	Article	IF	Citations
1	Utility of wholeâ€exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clinical Genetics, 2016, 89, 275-284.	2.0	323
2	Oligonucleotide Microarray Analysis of Genomic Imbalance in Children with Mental Retardation. American Journal of Human Genetics, 2006, 79, 500-513.	6.2	261
3	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758.	2.4	191
4	A patient with vertebral, cognitive and behavioural abnormalities and a de novo deletion of NRXN1Â. Journal of Medical Genetics, 2007, 45, 239-243.	3.2	123
5	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	6.2	83
6	Novel deletions of 14q11.2 associated with developmental delay, cognitive impairment and similar minor anomalies in three children. Journal of Medical Genetics, 2007, 44, 556-561.	3.2	68
7	Epigenetic Impacts on Neurodevelopment: Pathophysiological Mechanisms and Genetic Modes of Action. Pediatric Research, 2011, 69, 92R-100R.	2.3	62
8	Osteopoikilosis, short stature and mental retardation as key features of a new microdeletion syndrome on 12q14. Journal of Medical Genetics, 2007, 44, 264-268.	3.2	58
9	Duplications of the critical Rubinstein-Taybi deletion region on chromosome 16p13.3 cause a novel recognisable syndrome. Journal of Medical Genetics, 2010, 47, 155-161.	3.2	47
10	The impact of array genomic hybridization on mental retardation research: a review of current technologies and their clinical utility. Clinical Genetics, 2007, 72, 271-287.	2.0	43
11	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
12	Single exon-resolution targeted chromosomal microarray analysis of known and candidate intellectual disability genes. European Journal of Human Genetics, 2014, 22, 792-800.	2.8	35
13	A distinct neurodevelopmental syndrome with intellectual disability, autism spectrum disorder, characteristic facies, and macrocephaly is caused by defects in CHD8. Journal of Human Genetics, 2019, 64, 271-280.	2.3	35
14	Detection of pathogenic copy number variants in children with idiopathic intellectual disability using 500 K SNP array genomic hybridization. BMC Genomics, 2009, 10, 526.	2.8	30
15	A characteristic syndrome associated with microduplication of 8q12, inclusive of CHD7. European Journal of Medical Genetics, 2009, 52, 436-439.	1.3	21
16	Use of Affymetrix Arrays in the Diagnosis of Gene Copyâ€Number Variation. Current Protocols in Human Genetics, 2015, 85, 8.13.1-8.13.13.	3.5	15
17	Comprehensive whole genome sequence analyses yields novel genetic and structural insights for Intellectual Disability. BMC Genomics, 2017, 18, 403.	2.8	15
18	A novel de novo 1.1 Mb duplication of 17q21.33 associated with cognitive impairment and other anomalies. American Journal of Medical Genetics, Part A, 2009, 149A, 1257-1262.	1.2	14

#	Article	IF	Citations
19	Intragenic CNVs for epigenetic regulatory genes in intellectual disability: Survey identifies pathogenic and benign single exon changes. American Journal of Medical Genetics, Part A, 2016, 170, 2916-2926.	1.2	14
20	De novo pathogenic DNM1L variant in a patient diagnosed with atypical hereditary sensory and autonomic neuropathy. Molecular Genetics & Genomic Medicine, 2019, 7, e00961.	1.2	12
21	Maternal Prenatal Exposures in Pregnancy and Autism Spectrum Disorder: An Insight into the Epigenetics of Drugs and Diet as Key Environmental Influences. Advances in Neurobiology, 2020, 24, 143-162.	1.8	10
22	Discovering a new part of the phenotypic spectrum of Coffin-Siris syndrome in a fetal cohort. Genetics in Medicine, 2022, 24, 1753-1760.	2.4	6
23	Lifeâ€history chronicle for a patient with the recently described chromosome 4q21 microdeletion syndrome. American Journal of Medical Genetics, Part A, 2012, 158A, 2606-2609.	1.2	5
24	Chromodomain helicase DNA-binding proteins and neurodevelopmental disorders. , 0, , .		5
25	Recent Major Transcriptomics and Epitranscriptomics Contributions toward Personalized and Precision Medicine. Journal of Personalized Medicine, 2022, 12, 199.	2.5	3
26	First Whole Transcriptome RNAseq on CHD8 Haploinsufficient Patient and Meta-Analyses Across Cellular Models Uncovers Likely Key Pathophysiological Target Genes. Cureus, 2020, 12, e11571.	0.5	2
27	Understanding environmental epigenomics in autism spectrum disorder: an interview with Farah RÂZahir. Epigenomics, 2021, 13, 1341-1345.	2.1	0
28	The Need for Precision Therapies as Determined by Genetic Signature for Cystic Fibrosis. Journal of Personalized Medicine, 2021, 11, 1353.	2.5	0