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	Recent Major Transcriptomics and Epitranscriptomics Contributions toward Personalized and Precision Medicine. Journal of Personalized Medicine, 2022, 12, 199.	2.5	3
2	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
3	Understanding environmental epigenomics in autism spectrum disorder: an interview with Farah RÂZahir. Epigenomics, 2021, 13, 1341-1345.	2.1	0
4	The Need for Precision Therapies as Determined by Genetic Signature for Cystic Fibrosis. Journal of Personalized Medicine, 2021, 11, 1353.	2.5	0
5	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
6	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
7	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
8	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
9	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
10	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	6.2	83
11	Comprehensive whole genome sequence analyses yields novel genetic and structural insights for Intellectual Disability. BMC Genomics, 2017, 18, 403.	2.8	15
12	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
13	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
14	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
15	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
16	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758.	2.4	191
17	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
18	Epigenetic Impacts on Neurodevelopment: Pathophysiological Mechanisms and Genetic Modes of Action. Pediatric Research, 2011, 69, 92R-100R.	2.3	62

#	Article	IF	Citations
19	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
20	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
21	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
22	A characteristic syndrome associated with microduplication of 8q12, inclusive of CHD7. European Journal of Medical Genetics, 2009, 52, 436-439.	1.3	21
23	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
24	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
25	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
26	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
27	Oligonucleotide Microarray Analysis of Genomic Imbalance in Children with Mental Retardation. American Journal of Human Genetics, 2006, 79, 500-513.	6.2	261
28	Chromodomain helicase DNA-binding proteins and neurodevelopmental disorders. , 0, , .		5