Janson J White

List of Publications by Year
in descending order

21	1,115	567281 15	713466 21
papers	citations	h-index	g-index

21	21	21	2875
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Novel pathogenic variants and quantitative phenotypic analyses of Robinow syndrome: WNT signaling perturbation and phenotypic variability. Human Genetics and Genomics Advances, 2022, 3, 100074.	1.7	14
2	Phenotypic and mutational spectrum of ROR2 â€related Robinow syndrome. Human Mutation, 2022, 43, 900-918.	2.5	8
3	Novel pathogenic genomic variants leading to autosomal dominant and recessive Robinow syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 3593-3600.	1.2	16
4	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	2.4	24
5	Exome sequencing of family trios from the National Birth Defects Prevention Study: Tapping into a rich resource of genetic and environmental data. Birth Defects Research, 2019, 111, 1618-1632.	1.5	9
6	Paralog Studies Augment Gene Discovery: DDX and DHX Genes. American Journal of Human Genetics, 2019, 105, 302-316.	6.2	56
7	Exome Sequencing of a Primary Ovarian Insufficiency Cohort Reveals Common Molecular Etiologies for a Spectrum of Disease. Journal of Clinical Endocrinology and Metabolism, 2019, 104, 3049-3067.	3.6	53
8	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	2.4	161
9	WNT Signaling Perturbations Underlie the Genetic Heterogeneity of Robinow Syndrome. American Journal of Human Genetics, 2018, 102, 27-43.	6.2	88
10	Identifying Genes Whose Mutant Transcripts Cause Dominant Disease Traits by Potential Gain-of-Function Alleles. American Journal of Human Genetics, 2018, 103, 171-187.	6.2	160
11	CHRNA7 copy number gains are enriched in adolescents with major depressive and anxiety disorders. Journal of Affective Disorders, 2018, 239, 247-252.	4.1	12
12	Dual molecular diagnosis contributes to atypical Praderâ€“Willi phenotype in monozygotic twins. American Journal of Medical Genetics, Part A, 2017, 173, 2451-2455.	1.2	26
13	CHRNA7 Deletions are Enriched in Risperidone-Treated Children and Adolescents. Journal of Child and Adolescent Psychopharmacology, 2017, 27, 908-915.	1.3	9
14	REST Final-Exon-Truncating Mutations Cause Hereditary Gingival Fibromatosis. American Journal of Human Genetics, 2017, 101, 149-156.	6.2	44
15	De novo missense variants in PPP1CB are associated with intellectual disability and congenital heart disease. Human Genetics, 2016, 135, 1399-1409.	3.8	40
16	Hutteriteâ€type cataract maps to chromosome 6p21.32â€p21.31, cosegregates with a homozygous mutation in <scp>LEMD</scp>2, and is associated with sudden cardiac death. Molecular Genetics & Genomic Medicine, 2016, 4, 77-94.	1.2	28
17	Identification of a RAI1-associated disease network through integration of exome sequencing, transcriptomics, and 3D genomics. Genome Medicine, 2016, 8, 105.	8.2	20
18	POGZ truncating alleles cause syndromic intellectual disability. Genome Medicine, 2016, 8, 3.	8.2	78

#	Article	IF	Citations
19	DVL3 Alleles Resulting in a â^'1 Frameshift of the Last Exon Mediate Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2016, 98, 553-561.	6.2	88
20	Whole-Exome Sequencing in Familial Parkinson Disease. JAMA Neurology, 2016, 73, 68.	9.0	71
21	DVL1 Frameshift Mutations Clustering in the Penultimate Exon Cause Autosomal-Dominant Robinow Syndrome. American Journal of Human Genetics, 2015, 96, 612-622.	6.2	110