Himanshu Goel

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/692998/publications.pdf

Version: 2024-02-01

18	517	1040056 9	839539 18
papers	citations	h-index	g-index

19	19	19	1315
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. Genetics in Medicine, 2022, 24, 130-145.	2.4	45
2	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	1.2	15
3	A case of Whiteâ€“Sutton syndrome with previously described lossâ€ofâ€function variant in <scp>DDE</scp> domain of <scp>POGZ</scp> (p.Arg1211*) and Kartagener syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1006-1007.	1.2	5
4	Pseudoxanthoma elasticum and retinitis pigmentosa: dual diagnosis of recessive conditions with ophthalmological consequences. Ophthalmic Genetics, 2020, 41, 470-473.	1.2	2
5	Pathogenic nonsense variant in NFIB in another patient with dysmorphism, Autism Spectrum Disorder, agenesis of the corpus callosum, and intellectual disability. European Journal of Medical Genetics, 2020, 63, 104092.	1.3	6
6	Novel de novo <scp>TRIP12</scp> mutation reveals variable phenotypic presentation while emphasizing core features of <scp>TRIP12</scp> variations. American Journal of Medical Genetics, Part A, 2020, 182, 1801-1806.	1.2	6
7	De Novo Variants in CNOT1, a Central Component of the CCR4-NOT Complex Involved in Gene Expression and RNA and Protein Stability, Cause Neurodevelopmental Delay. American Journal of Human Genetics, 2020, 107, 164-172.	6.2	37
8	Two novel B9D1 variants causing Joubert syndrome: Utility of mRNA and splicing studies. European Journal of Medical Genetics, 2020, 63, 104000.	1.3	1
9	Clinical spectrum, prognosis and estimated prevalence of DNAJB11-kidney disease. Kidney International, 2020, 98, 476-487.	5.2	38
10	Expanding the phenotype of intellectual disability caused by HIVEP2 variants. American Journal of Medical Genetics, Part A, 2019, 179, 1872-1877.	1.2	7
11	DNA Polymerase Epsilon Deficiency Causes IMAGe Syndrome with Variable Immunodeficiency. American Journal of Human Genetics, 2018, 103, 1038-1044.	6.2	71
12	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. American Journal of Human Genetics, 2017, 101, 23-36.	6.2	74
13	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	2.5	40
14	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114
15	Heterozygous Deletion of KLHL1/ATX8OS at the SCA8 Locus Is Unlikely Associated With Cerebellar Impairment in Humans. Cerebellum, 2016, 15, 208-212.	2.5	3
16	Narrowing the critical region for overgrowth within 13q14.2-q14.3 microdeletions. European Journal of Medical Genetics, 2015, 58, 629-633.	1.3	9
17	An intragenic deletion of the NFIA gene in a patient with a hypoplastic corpus callosum, craniofacial abnormalities and urinary tract defects. European Journal of Medical Genetics, 2014, 57, 65-70.	1.3	36
18	Carbimazole/methimazole embryopathy in siblings: A possible genetic susceptibility. Birth Defects Research Part A: Clinical and Molecular Teratology, 2013, 97, 755-758.	1.6	8