

# Himanshu Goel

## List of Publications by Year in descending order

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Version: 2024-02-01

18  
papers

517  
citations

1040056

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839539

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19  
docs citations

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times ranked

1315  
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. <i>Nature Genetics</i> , 2016, 48, 1185-1192.	21.4	114
2	Mutations in ARMC9, which Encodes a Basal Body Protein, Cause Joubert Syndrome in Humans and Ciliopathy Phenotypes in Zebrafish. <i>American Journal of Human Genetics</i> , 2017, 101, 23-36.	6.2	74
3	DNA Polymerase Epsilon Deficiency Causes IMAGE Syndrome with Variable Immunodeficiency. <i>American Journal of Human Genetics</i> , 2018, 103, 1038-1044.	6.2	71
4	Standardized practices for RNA diagnostics using clinically accessible specimens reclassifies 75% of putative splicing variants. <i>Genetics in Medicine</i> , 2022, 24, 130-145.	2.4	45
5	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. <i>Human Mutation</i> , 2016, 37, 653-660.	2.5	40
6	Clinical spectrum, prognosis and estimated prevalence of DNAJB11-kidney disease. <i>Kidney International</i> , 2020, 98, 476-487.	5.2	38
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. <i>American Journal of Human Genetics</i> , 2020, 107, 164-172.	6.2	37
8	An intragenic deletion of the NFIA gene in a patient with a hypoplastic corpus callosum, craniofacial abnormalities and urinary tract defects. <i>European Journal of Medical Genetics</i> , 2014, 57, 65-70.	1.3	36
9	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 15-25.	1.2	15
10	Narrowing the critical region for overgrowth within 13q14.2-q14.3 microdeletions. <i>European Journal of Medical Genetics</i> , 2015, 58, 629-633.	1.3	9
11	Carbimazole/methimazole embryopathy in siblings: A possible genetic susceptibility. <i>Birth Defects Research Part A: Clinical and Molecular Teratology</i> , 2013, 97, 755-758.	1.6	8
12	Expanding the phenotype of intellectual disability caused by <i>HIVEP2</i> variants. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1872-1877.	1.2	7
13	Pathogenic nonsense variant in NFIB in another patient with dysmorphism, Autism Spectrum Disorder, agenesis of the corpus callosum, and intellectual disability. <i>European Journal of Medical Genetics</i> , 2020, 63, 104092.	1.3	6
14	Novel de novo <i>TRIP12</i> mutation reveals variable phenotypic presentation while emphasizing core features of <i>TRIP12</i> variations. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1801-1806.	1.2	6
15	A case of Whiteâ€Sutton syndrome with previously described loss-of-function variant in <i>DDE</i> domain of <i>POGZ</i> (p.Arg1211*) and Kartagener syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1006-1007.	1.2	5
16	Heterozygous Deletion of KLHL1/ATX8OS at the SCA8 Locus Is Unlikely Associated With Cerebellar Impairment in Humans. <i>Cerebellum</i> , 2016, 15, 208-212.	2.5	3
17	Pseudoxanthoma elasticum and retinitis pigmentosa: dual diagnosis of recessive conditions with ophthalmological consequences. <i>Ophthalmic Genetics</i> , 2020, 41, 470-473.	1.2	2
18	Two novel B9D1 variants causing Joubert syndrome: Utility of mRNA and splicing studies. <i>European Journal of Medical Genetics</i> , 2020, 63, 104000.	1.3	1