Madhuri Hegde

List of Publications by Year
in descending order

37 papers	22,069 citations	331670 21 h-index	361022 35 g-index
37 all docs	37 docs citations	37 times ranked	34768 citing authors

#	Article	IF	Citations
1	Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine, 2015, 17, 405-424.	2.4	20,455
2	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758.	2.4	191
3	Navigating highly homologous genes in a molecular diagnostic setting: a resource for clinical next-generation sequencing. Genetics in Medicine, 2016, 18, 1282-1289.	2.4	170
4	ACMG technical standards and guidelines for genetic testing for inherited colorectal cancer (Lynch) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5 16, 101-116.	2.4	148
5	ClinGen Variant Curation Expert Panel experiences and standardized processes for disease and geneâ€level specification of the ACMG/AMP guidelines for sequence variant interpretation. Human Mutation, 2018, 39, 1614-1622.	2.5	132
6	Genetic landscape and novel disease mechanisms from a large <scp>LGMD</scp> cohort of 4656 patients. Annals of Clinical and Translational Neurology, 2018, 5, 1574-1587.	3.7	129
7	Geneâ€specific criteria for PTEN variant curation: Recommendations from the ClinGen PTEN Expert Panel. Human Mutation, 2018, 39, 1581-1592.	2.5	123
8	Diagnostic Yield From 339 Epilepsy Patients Screened on a Clinical Gene Panel. Pediatric Neurology, 2017, 77, 61-66.	2.1	83
9	A survey of current practices for genomic sequencing test interpretation and reporting processes in US laboratories. Genetics in Medicine, 2017, 19, 575-582.	2.4	68
10	Reporting Incidental Findings in Genomic Scale Clinical Sequencingâ€”A Clinical Laboratory Perspective. Journal of Molecular Diagnostics, 2015, 17, 107-117.	2.8	55
11	Reassessment of Genomic Sequence Variation to Harmonize Interpretation for Personalized Medicine. American Journal of Human Genetics, 2016, 99, 1140-1149.	6.2	53
12	Mutations in TBCK, Encoding TBC1-Domain-Containing Kinase, Lead to a Recognizable Syndrome of Intellectual Disability and Hypotonia. American Journal of Human Genetics, 2016, 98, 782-788.	6.2	50
13	Peripheral motor neuropathy is associated with defective kinase regulation of the KCC3 cotransporter. Science Signaling, 2016, 9, ra77.	3.6	46
14	Gene and Variant Annotation for Mendelian Disorders in the Era of Advanced Sequencing Technologies. Annual Review of Genomics and Human Genetics, 2017, 18, 229-256.	6.2	37
15	De novo and inherited SCN8A epilepsy mutations detected by gene panel analysis. Epilepsy Research, 2017, 129, 17-25.	1.6	34
16	Variants of uncertain significance in newborn screening disorders: implications for large-scale genomic sequencing. Genetics in Medicine, 2017, 19, 77-82.	2.4	29
17	Why West? Comparisons of clinical, genetic and molecular features of infants with and without spasms. PLoS ONE, 2018, 13, e0193599.	2.5	28
18	A single NGSâ€based assayÂcovering the entire genomic sequence of the DMD gene facilitates diagnostic and newborn screening confirmatory testing. Human Mutation, 2021, 42, 626-638.	2.5	25

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19	The Case for Laboratory Developed Procedures. Academic Pathology, 2017, 4, 2374289517708309.	1.1	24
20	Inferring the effect of genomic variation in the new era of genomics. Human Mutation, 2018, 39, 756-773.	2.5	24
21	Evidence-Based Consensus and Systematic Review on Reducing the Time to Diagnosis of Duchenne Muscular Dystrophy. Journal of Pediatrics, 2019, 204, 305-313.e14.	1.8	24
22	Development and Performance of a Comprehensive Targeted Sequencing Assay for Pan-Ethnic Screening of Carrier Status. Journal of Molecular Diagnostics, 2014, 16, 350-360.	2.8	21
23	A report on GNE myopathy: Individuals of Rajasthan ancestry share the Roma gene. Journal of the Neurological Sciences, 2017, 375, 239-240.	0.6	18
24	Detection limit of intragenic deletions with targeted array comparative genomic hybridization. BMC Genetics, 2013, 14, 116.	2.7	15
25	Detecting APC Gene Mutations in Familial Adenomatous Polyposis (FAP). Current Protocols in Human Genetics, 2017, 92, 10.8.1-10.8.16.	3.5	15
26	Clinical utility of RNA sequencing to resolve unusual GNE myopathy with a novel promoter deletion. Muscle and Nerve, 2019, 60, 98-103.	2.2	15
27	Genomic Technologies and the New Era of Genomic Medicine. Journal of Molecular Diagnostics, 2014, 16, 7-10.	2.8	11
28	Detection of dysferlin gene pathogenic variants in the Indian Population in patients predicted to have a dysferlinopathy using a blood-based monocyte assay and clinical algorithm: A model for accurate and cost-effective diagnosis. Annals of Indian Academy of Neurology, 2017, 20, 302.	0.5	10
29	Genetic Testing for Hereditary Nonpolyposis Colorectal Cancer (HNPCC). Current Protocols in Human Genetics, 2017, 94, 10.12.1-10.12.23.	3.5	8
30	Results from an external proficiency testing program: 11 years of molecular genetics testing for myotonic dystrophy type 1. Genetics in Medicine, 2016, 18, 1290-1294.	2.4	6
31	Caution in interpretation of disease causality for heterozygous loss-of-function variants in the MYH8 gene associated with autosomal dominant disorder. European Journal of Medical Genetics, 2017, 60, 312-316.	1.3	6
32	RNA-seq in DMD urinary stem cells recognized muscle-related transcription signatures and addressed the identification of atypical mutations by whole-genome sequencing. Human Genetics and Genomics Advances, 2022, 3, 100054.	1.7	6
33	Clinical Utility of Transcriptome Sequencing: Toward a Better Diagnosis for Mendelian Disorders. Clinical Chemistry, 2018, 64, 882-884.	3.2	5
34	An Educational Assessment of Evidence Used for Variant Classification. Journal of Molecular Diagnostics, 2022, 24, 555-565.	2.8	3
35	Reply: Autosomal dominant segregation of CAPN3 c.598_612del15 associated with a mild form of calpainopathy. Annals of Clinical and Translational Neurology, 2020, 7, 2541-2541.	3.7	2
36	Reply to Clarity and claims in variation/mutation databasing. Nature Biotechnology, 2011, 29, 792-794.	17.5	0

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37	High-Throughput Sequencing in the Context of Human Genetic Diseases: Now and Tomorrow. Human Mutation, 2016, 37, 1247-1247.	2.5	0