

Heather Mason-Suares

List of Publications by Year in descending order

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Version: 2024-02-01

28
papers

1,579
citations

516710

16
h-index

552781

26
g-index

31
all docs

31
docs citations

31
times ranked

4231
citing authors

#	ARTICLE	IF	CITATIONS
1	Combined X-linked familial exudative vitreoretinopathy and retinopathy of prematurity phenotype in an infant with mosaic turner syndrome with ring X chromosome. <i>Ophthalmic Genetics</i> , 2023, 44, 198-203.	1.2	0
2	RASopathies. , 2021, , 389-398.		0
3	Considerations for whole exome sequencing unique to prenatal care. <i>Human Genetics</i> , 2020, 139, 1149-1159.	3.8	18
4	Association of Rare Pathogenic DNA Variants for Familial Hypercholesterolemia, Hereditary Breast and Ovarian Cancer Syndrome, and Lynch Syndrome With Disease Risk in Adults According to Family History. <i>JAMA Network Open</i> , 2020, 3, e203959.	5.9	75
5	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. <i>Human Mutation</i> , 2020, 41, 1577-1587.	2.5	10
6	A Role for Chromosomal Microarray Testing in the Workup of Male Infertility. <i>Journal of Molecular Diagnostics</i> , 2020, 22, 1189-1198.	2.8	4
7	Expanding the Noonan spectrum/RASopathy NGS panel: Benefits of adding <i>NF1</i> and <i>SPRED1</i> . <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1180.	1.2	9
8	Rare Genetic Variants Associated With Sudden Cardiac Death in Adults. <i>Journal of the American College of Cardiology</i> , 2019, 74, 2623-2634.	2.8	27
9	Considerations for clinical curation, classification, and reporting of low-penetrance and low effect size variants associated with disease risk. <i>Genetics in Medicine</i> , 2019, 21, 2765-2773.	2.4	20
10	Polygenic Prediction of Weight and Obesity Trajectories from Birth to Adulthood. <i>Cell</i> , 2019, 177, 587-596.e9.	28.9	516
11	ClinGen™s RASopathy Expert Panel consensus methods for variant interpretation. <i>Genetics in Medicine</i> , 2018, 20, 1334-1345.	2.4	126
12	Analysis of intragenic USH2A copy number variation unveils broad spectrum of unique and recurrent variants. <i>European Journal of Medical Genetics</i> , 2018, 61, 621-626.	1.3	9
13	When ultrasound anomalies are present: An estimation of the frequency of chromosome abnormalities not detected by cell-free DNA aneuploidy screens. <i>Prenatal Diagnosis</i> , 2018, 38, 250-257.	2.3	13
14	Early cancer diagnoses through BRCA1/2 screening of unselected adult biobank participants. <i>Genetics in Medicine</i> , 2018, 20, 554-558.	2.4	46
15	NGS testing for cardiomyopathy: Utility of adding RASopathy-associated genes. <i>Human Mutation</i> , 2018, 39, 954-958.	2.5	11
16	Exome Sequencing-Based Screening for BRCA1/2 Expected Pathogenic Variants Among Adult Biobank Participants. <i>JAMA Network Open</i> , 2018, 1, e182140.	5.9	163
17	Assessing the gene-disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. <i>Human Mutation</i> , 2018, 39, 1485-1493.	2.5	66
18	A Model for Genome-First Care: Returning Secondary Genomic Findings to Participants and Their Healthcare Providers in a Large Research Cohort. <i>American Journal of Human Genetics</i> , 2018, 103, 328-337.	6.2	130

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19	Juvenile myelomonocytic leukemia-associated variants are associated with neo-natal lethal Noonan syndrome. <i>European Journal of Human Genetics</i> , 2017, 25, 509-511.	2.8	18
20	The current state of clinical interpretation of sequence variants. <i>Current Opinion in Genetics and Development</i> , 2017, 42, 33-39.	3.3	77
21	Electronic health record phenotype in subjects with genetic variants associated with arrhythmogenic right ventricular cardiomyopathy: a study of 30,716 subjects with exome sequencing. <i>Genetics in Medicine</i> , 2017, 19, 1245-1252.	2.4	43
22	Elucidation of MRAS-mediated Noonan syndrome with cardiac hypertrophy. <i>JCI Insight</i> , 2017, 2, e91225.	5.0	66
23	Training the Future Leaders in Personalized Medicine. <i>Journal of Personalized Medicine</i> , 2016, 6, 1.	2.5	21
24	Detecting Copy Number Variation via Next Generation Technology. <i>Current Genetic Medicine Reports</i> , 2016, 4, 74-85.	1.9	20
25	Retrospective study of prenatal ultrasound findings in newborns with a Noonan spectrum disorder. <i>Prenatal Diagnosis</i> , 2016, 36, 418-423.	2.3	25
26	Targeted Droplet-Digital PCR as a Tool for Novel Deletion Discovery at the DFNB1 Locus. <i>Human Mutation</i> , 2016, 37, 119-126.	2.5	37
27	Polycomb silencing of the <i>Drosophila</i> 4E-BP gene regulates imaginal disc cell growth. <i>Developmental Biology</i> , 2013, 380, 111-124.	2.0	3
28	Density matters: comparison of array platforms for detection of copy-number variation and copy-neutral abnormalities. <i>Genetics in Medicine</i> , 2013, 15, 706-712.	2.4	24