Heather Mason-Suares

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/2464627/publications.pdf

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	Polygenic Prediction of Weight and Obesity Trajectories from Birth to Adulthood. Cell, 2019, 177, 587-596.e9.	28.9	516
2	Exome Sequencingâ€“Based Screening for BRCA1/2 Expected Pathogenic Variants Among Adult Biobank Participants. JAMA Network Open, 2018, 1, e182140.	5.9	163
3	A Model for Genome-First Care: Returning Secondary Genomic Findings to Participants and Their Healthcare Providers in a Large Research Cohort. American Journal of Human Genetics, 2018, 103, 328-337.	6.2	130
4	ClinGenâ€™s RASopathy Expert Panel consensus methods for variant interpretation. Genetics in Medicine, 2018, 20, 1334-1345.	2.4	126
5	The current state of clinical interpretation of sequence variants. Current Opinion in Genetics and Development, 2017, 42, 33-39.	3.3	77
6	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. JAMA Network Open, 2020, 3, e203959.	5.9	75
7	Elucidation of MRAS-mediated Noonan syndrome with cardiac hypertrophy. JCI Insight, 2017, 2, e91225.	5.0	66
8	Assessing the geneâ€“disease association of 19 genes with the RASopathies using the ClinGen gene curation framework. Human Mutation, 2018, 39, 1485-1493.	2.5	66
9	Early cancer diagnoses through BRCA1/2 screening of unselected adult biobank participants. Genetics in Medicine, 2018, 20, 554-558.	2.4	46
10	Electronic health record phenotype in subjects with genetic variants associated with arrhythmogenic right ventricular cardiomyopathy: a study of 30,716 subjects with exome sequencing. Genetics in Medicine, 2017, 19, 1245-1252.	2.4	43
11	Targeted Droplet-Digital PCR as a Tool for Novel Deletion Discovery at the DFNB1 Locus. Human Mutation, 2016, 37, 119-126.	2.5	37
12	Rare Genetic Variants Associated With Sudden Cardiac Death in Adults. Journal of the American College of Cardiology, 2019, 74, 2623-2634.	2.8	27
13	Retrospective study of prenatal ultrasound findings in newborns with a Noonan spectrum disorder. Prenatal Diagnosis, 2016, 36, 418-423.	2.3	25
14	Density matters: comparison of array platforms for detection of copy-number variation and copy-neutral abnormalities. Genetics in Medicine, 2013, 15, 706-712.	2.4	24
15	Training the Future Leaders in Personalized Medicine. Journal of Personalized Medicine, 2016, 6, 1.	2.5	21
16	Detecting Copy Number Variation via Next Generation Technology. Current Genetic Medicine Reports, 2016, 4, 74-85.	1.9	20
17	Considerations for clinical curation, classification, and reporting of low-penetrance and low effect size variants associated with disease risk. Genetics in Medicine, 2019, 21, 2765-2773.	2.4	20
18	Juvenile myelomonocytic leukemia-associated variants are associated with neo-natal lethal Noonan syndrome. European Journal of Human Genetics, 2017, 25, 509-511.	2.8	18

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19	Considerations for whole exome sequencing unique to prenatal care. Human Genetics, 2020, 139, 1149-1159.	3.8	18
20	When ultrasound anomalies are present: An estimation of the frequency of chromosome abnormalities not detected by cellâ€free DNA aneuploidy screens. Prenatal Diagnosis, 2018, 38, 250-257.	2.3	13
21	NGS testing for cardiomyopathy: Utility of adding RASopathy-associated genes. Human Mutation, 2018, 39, 954-958.	2.5	11
22	An assessment of the role of vinculin loss of function variants in inherited cardiomyopathy. Human Mutation, 2020, 41, 1577-1587.	2.5	10
23	Analysis of intragenic USH2A copy number variation unveils broad spectrum of unique and recurrent variants. European Journal of Medical Genetics, 2018, 61, 621-626.	1.3	9
24	Expanding the Noonan spectrum/RASopathy NGS panel: Benefits of adding NF1 and SPRED1. Molecular Genetics & Genomic Medicine, 2020, 8, e1180.	1.2	9
25	A Role for Chromosomal Microarray Testing in the Workup of Male Infertility. Journal of Molecular Diagnostics, 2020, 22, 1189-1198.	2.8	4
26	Polycomb silencing of the Drosophila 4E-BP gene regulates imaginal disc cell growth. Developmental Biology, 2013, 380, 111-124.	2.0	3
27	RASopathies. , 2021, , 389-398.		0
28	Combined X-linked familial exudative vitreoretinopathy and retinopathy of prematurity phenotype in an infant with mosaic turner syndrome with ring X chromosome. Ophthalmic Genetics, 2023, 44, 198-203.	1.2	0