Sarah Bowdin

List of Publications by Year
in descending order

21 papers	2,226 citations	516710 16 h-index	752698 20 g-index
21 all docs	21 docs citations	21 times ranked	4994 citing authors

#	Article	IF	Citations
1	Loeysâ€“Dietz syndrome: a primer for diagnosis and management. Genetics in Medicine, 2014, 16, 576-587.	2.4	435
2	Improved diagnostic yield compared with targeted gene sequencing panels suggests a role for whole-genome sequencing as a first-tier genetic test. Genetics in Medicine, 2018, 20, 435-443.	2.4	404
3	Whole-genome sequencing expands diagnostic utility and improves clinical management in paediatric medicine. Npj Genomic Medicine, 2016, 1, .	3.8	295
4	Reappraisal of Reported Genes for Sudden Arrhythmic Death. Circulation, 2018, 138, 1195-1205.	1.6	271
5	PhenoTips: Patient Phenotyping Software for Clinical and Research Use. Human Mutation, 2013, 34, 1057-1065.	2.5	207
6	Recommendations for the integration of genomics into clinical practice. Genetics in Medicine, 2016, 18, 1075-1084.	2.4	125
7	Periodic reanalysis of whole-genome sequencing data enhances the diagnostic advantage over standard clinical genetic testing. European Journal of Human Genetics, 2018, 26, 740-744.	2.8	88
8	The Personal Genome Project Canada: findings from whole genome sequences of the inaugural 56 participants. Cmaj, 2018, 190, E126-E136.	2.0	57
9	The Cardiac Genome Clinic: implementing genome sequencing in pediatric heart disease. Genetics in Medicine, 2020, 22, 1015-1024.	2.4	51
10	MKS1 regulates ciliary INPP5E levels in Joubert syndrome. Journal of Medical Genetics, 2016, 53, 62-72.	3.2	48
11	Novel MASP1 mutations are associated with an expanded phenotype in 3MC1 syndrome. Orphanet Journal of Rare Diseases, 2015, 10, 128.	2.7	46
12	Genome sequencing as a platform for pharmacogenetic genotyping: a pediatric cohort study. Npj Genomic Medicine, 2017, 2, 19.	3.8	41
13	Exome sequencing identifies rare variants in multiple genes in atrioventricular septal defect. Genetics in Medicine, 2016, 18, 189-198.	2.4	39
14	Care and cost consequences of pediatric whole genome sequencing compared to chromosome microarray. European Journal of Human Genetics, 2017, 25, 1303-1312.	2.8	32
15	The Genome Clinic: A Multidisciplinary Approach to Assessing the Opportunities and Challenges of Integrating Genomic Analysis into Clinical Care. Human Mutation, 2014, 35, 513-519.	2.5	31
16	Heterozygous mutations in ERF cause syndromic craniosynostosis with multiple suture involvement. American Journal of Medical Genetics, Part A, 2015, 167, 2544-2547.	1.2	21
17	FGFRâ€associated craniosynostosis syndromes and gastrointestinal defects. American Journal of Medical Genetics, Part A, 2016, 170, 3215-3221.	1.2	13
18	Use of Clinical Exome Sequencing in Isolated Congenital Heart Disease. Circulation: Cardiovascular Genetics, 2017, 10, .	5.1	13

#	Article	IF	Citations
19	Genome-wide sequencing expands the phenotypic spectrum of EP300 variants. European Journal of Medical Genetics, 2018, 61, 125-129.	1.3	8
20	MG-132â€...Diagnostic utility of whole genome sequencing in paediatric medicine. Journal of Medical Genetics, 2015, 52, A12.1-A12.	3.2	1
21	MG-130â€...Utilising whole exome sequencing to identify causative variants in genetically heterogeneous disorders. Journal of Medical Genetics, 2015, 52, A11.2-A11.	3.2	0