Zoe Powis

List of Publications by Year
in descending order

23 papers	1,107 citations	516710 16 h-index	677142 22 g-index
23 all docs	23 docs citations	23 times ranked	3277 citing authors

#	Article	IF	Citations
1	Enhanced utility of family-centered diagnostic exome sequencing with inheritance modelâ€“based analysis: results from 500 unselected families with undiagnosed genetic conditions. Genetics in Medicine, 2015, 17, 578-586.	2.4	401
2	Exome sequencing positively identified relevant alterations in more than half of cases with an indication of prenatal ultrasound anomalies. Prenatal Diagnosis, 2015, 35, 1073-1078.	2.3	88
3	Classification of Genes: Standardized Clinical Validity Assessment of Gene-Disease Associations Aids Diagnostic Exome Analysis and Reclassifications. Human Mutation, 2017, 38, 600-608.	2.5	83
4	Outcomes of Diagnostic Exome Sequencing in Patients With Diagnosed or Suspected Autism Spectrum Disorders. Pediatric Neurology, 2017, 70, 34-43.e2.	2.1	82
5	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. Human Molecular Genetics, 2018, 27, 2454-2465.	2.9	54
6	Candidate-gene criteria for clinical reporting: diagnostic exome sequencing identifies altered candidate genes among 8% of patients with undiagnosed diseases. Genetics in Medicine, 2017, 19, 224-235.	2.4	47
7	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. European Journal of Human Genetics, 2019, 27, 747-759.	2.8	47
8	Clinical wholeâ€exome sequencing results impact medical management. Molecular Genetics & Genomic Medicine, 2018, 6, 1068-1078.	1.2	33
9	Biallelic mutations in the ferredoxin reductase gene cause novel mitochondriopathy with optic atrophy. Human Molecular Genetics, 2017, 26, 4937-4950.	2.9	32
10	Exome sequencing in neonates: diagnostic rates, characteristics, and time to diagnosis. Genetics in Medicine, 2018, 20, 1468-1471.	2.4	31
11	MAGEL2â€related disorders: A study and case series. Clinical Genetics, 2019, 96, 493-505.	2.0	26
12	Clinical diagnostic exome sequencing in dystonia: Genetic testing challenges for complex conditions. Clinical Genetics, 2020, 97, 305-311.	2.0	25
13	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	2.8	25
14	Germline activating MTOR mutation arising through gonadal mosaicism in two brothers with megalencephaly and neurodevelopmental abnormalities. BMC Medical Genetics, 2015, 16, 102.	2.1	23
15	Expansion and further delineation of the SETD5 phenotype leading to global developmental delay, variable dysmorphic features, and reduced penetrance. Clinical Genetics, 2018, 93, 752-761.	2.0	23
16	Clinical diagnostic exome evaluation for an infant with a lethal disorder: genetic diagnosis of TARP syndrome and expansion of the phenotype in a patient with a newly reported RBM10 alteration. BMC Medical Genetics, 2017, 18, 60.	2.1	20
17	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. PLoS Genetics, 2018, 14, e1007671.	3.5	16
18	Biallelic disruption ofPKDCCis associated with a skeletal disorder characterised by rhizomelic shortening of extremities and dysmorphic features. Journal of Medical Genetics, 2019, 56, 850-854.	3.2	13

#	Article	IF	Citations
19	When moments matter: Finding answers with rapid exome sequencing. Molecular Genetics & Genomic Medicine, 2020, 8, e1027.	1.2	12
20	Identification of a syndrome comprising microcephaly and intellectual disability but not white matter disease associated with a homozygous c.676C>T p.R226W DEAF1 mutation. American Journal of Medical Genetics, Part A, 2016, 170, 1330-1332.	1.2	10
21	De novo variants in KLF7 are a potential novel cause of developmental delay/intellectual disability, neuromuscular and psychiatric symptoms. Clinical Genetics, 2018, 93, 1030-1038.	2.0	9
22	Clinical germline diagnostic exome sequencing for hereditary cancer: Findings within novel candidate genes are prevalent. Cancer Genetics, 2018, 224-225, 12-20.	0.4	7
23	The Power Of Exome: Finding Answers And Ending DoubtÂ. , 2018, , .		0