

Zoe Powis

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/3040435/publications.pdf>

Version: 2024-02-01

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	Clinical diagnostic exome sequencing in dystonia: Genetic testing challenges for complex conditions. <i>Clinical Genetics</i> , 2020, 97, 305-311.	2.0	25
2	When moments matter: Finding answers with rapid exome sequencing. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1027.	1.2	12
3	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. <i>European Journal of Human Genetics</i> , 2020, 28, 1422-1431.	2.8	25
4	<i>MAGEL2</i> -related disorders: A study and case series. <i>Clinical Genetics</i> , 2019, 96, 493-505.	2.0	26
5	Spatially clustering de novo variants in <i>CYFIP2</i> , encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. <i>European Journal of Human Genetics</i> , 2019, 27, 747-759.	2.8	47
6	Biallelic disruption of <i>PKDCC</i> is associated with a skeletal disorder characterised by rhizomelic shortening of extremities and dysmorphic features. <i>Journal of Medical Genetics</i> , 2019, 56, 850-854.	3.2	13
7	Clinical germline diagnostic exome sequencing for hereditary cancer: Findings within novel candidate genes are prevalent. <i>Cancer Genetics</i> , 2018, 224-225, 12-20.	0.4	7
8	De novo variants in <i>KLF7</i> are a potential novel cause of developmental delay/intellectual disability, neuromuscular and psychiatric symptoms. <i>Clinical Genetics</i> , 2018, 93, 1030-1038.	2.0	9
9	Exome sequencing in neonates: diagnostic rates, characteristics, and time to diagnosis. <i>Genetics in Medicine</i> , 2018, 20, 1468-1471.	2.4	31
10	Expansion and further delineation of the <i>SETD5</i> phenotype leading to global developmental delay, variable dysmorphic features, and reduced penetrance. <i>Clinical Genetics</i> , 2018, 93, 752-761.	2.0	23
11	De novo mutations in the GTP/GDP-binding region of <i>RALA</i> , a RAS-like small GTPase, cause intellectual disability and developmental delay. <i>PLoS Genetics</i> , 2018, 14, e1007671.	3.5	16
12	Clinical whole-exome sequencing results impact medical management. <i>Molecular Genetics & Genomic Medicine</i> , 2018, 6, 1068-1078.	1.2	33
13	Functional variants in <i>TBX2</i> are associated with a syndromic cardiovascular and skeletal developmental disorder. <i>Human Molecular Genetics</i> , 2018, 27, 2454-2465.	2.9	54
14	The Power Of Exome: Finding Answers And Ending Doubt. , 2018, , .		0
15	Classification of Genes: Standardized Clinical Validity Assessment of Gene-Disease Associations Aids Diagnostic Exome Analysis and Reclassifications. <i>Human Mutation</i> , 2017, 38, 600-608.	2.5	83
16	Outcomes of Diagnostic Exome Sequencing in Patients With Diagnosed or Suspected Autism Spectrum Disorders. <i>Pediatric Neurology</i> , 2017, 70, 34-43.e2.	2.1	82
17	Biallelic mutations in the ferredoxin reductase gene cause novel mitochondriopathy with optic atrophy. <i>Human Molecular Genetics</i> , 2017, 26, 4937-4950.	2.9	32
18	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 <i>RBM10</i> alteration. <i>BMC Medical Genetics</i> , 2017, 18, 60.	2.1	20

#	ARTICLE	IF	CITATIONS
19	Candidate-gene criteria for clinical reporting: diagnostic exome sequencing identifies altered candidate genes among 8% of patients with undiagnosed diseases. <i>Genetics in Medicine</i> , 2017, 19, 224-235.	2.4	47
20	Identification of a syndrome comprising microcephaly and intellectual disability but not white matter disease associated with a homozygous c.676C>T p.R226W <i>DEAF1</i> mutation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1330-1332.	1.2	10
21	Germline activating MTOR mutation arising through gonadal mosaicism in two brothers with megalencephaly and neurodevelopmental abnormalities. <i>BMC Medical Genetics</i> , 2015, 16, 102.	2.1	23
22	Exome sequencing positively identified relevant alterations in more than half of cases with an indication of prenatal ultrasound anomalies. <i>Prenatal Diagnosis</i> , 2015, 35, 1073-1078.	2.3	88
23	Enhanced utility of family-centered diagnostic exome sequencing with inheritance model-based analysis: results from 500 unselected families with undiagnosed genetic conditions. <i>Genetics in Medicine</i> , 2015, 17, 578-586.	2.4	401