

Richard Caswell

List of Publications by Year in descending order

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34
papers

1,767
citations

471509

17
h-index

434195

31
g-index

36
all docs

36
docs citations

36
times ranked

3778
citing authors

#	ARTICLE	IF	CITATIONS
1	Mitochondrial Retinopathy. <i>Ophthalmology Retina</i> , 2022, 6, 65-79.	2.4	26
2	Mild MDPL in a patient with a novel de novo missense variant in the Cys-B region of POLD1. <i>European Journal of Human Genetics</i> , 2022, 30, 960-966.	2.8	2
3	Robinow syndrome in an extremely preterm infant: Novel homozygous ROR2 variant detected by rapid exome sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2021, , .	1.2	0
4	De Novo Mutations in <i>EIF2B1</i> Affecting eIF2 Signaling Cause Neonatal/Early-Onset Diabetes and Transient Hepatic Dysfunction. <i>Diabetes</i> , 2020, 69, 477-483.	0.6	29
5	Response to Comment on Misra et al. Homozygous Hypomorphic HNF1A Alleles Are a Novel Cause of Young-Onset Diabetes and Result in Sulfonylurea-Sensitive Diabetes. <i>Diabetes Care</i> 2020;43:909-912. <i>Diabetes Care</i> , 2020, 43, e155-e156.	8.6	0
6	Classification and correlation of RYR2 missense variants in individuals with catecholaminergic polymorphic ventricular tachycardia reveals phenotypic relationships. <i>Journal of Human Genetics</i> , 2020, 65, 531-539.	2.3	20
7	Late-onset Pseudoxanthoma Elasticum Associated with a Hypomorphic ABCC6 Variant. <i>American Journal of Ophthalmology</i> , 2020, 218, 255-260.	3.3	11
8	Homozygous Hypomorphic <i>HNF1A</i> Alleles Are a Novel Cause of Young-Onset Diabetes and Result in Sulfonylurea-Sensitive Diabetes. <i>Diabetes Care</i> , 2020, 43, 909-912.	8.6	13
9	Hemizygous UBA5 missense mutation unmasks recessive disorder in a patient with infantile-onset encephalopathy, acquired microcephaly, small cerebellum, movement disorder and severe neurodevelopmental delay. <i>European Journal of Medical Genetics</i> , 2019, 62, 97-102.	1.3	15
10	A hypomorphic allele of SLC35D1 results in Schneckengebeken-like dysplasia. <i>Human Molecular Genetics</i> , 2019, 28, 3543-3551.	2.9	9
11	A Specific CNOT1 Mutation Results in a Novel Syndrome of Pancreatic Agenesis and Holoprosencephaly through Impaired Pancreatic and Neurological Development. <i>American Journal of Human Genetics</i> , 2019, 104, 985-989.	6.2	43
12	Copy number variation of <i>LINGO1</i> in familial dystonic tremor. <i>Neurology: Genetics</i> , 2019, 5, e307.	1.9	8
13	Using Structural Analysis In Silico to Assess the Impact of Missense Variants in MEN1. <i>Journal of the Endocrine Society</i> , 2019, 3, 2258-2275.	0.2	14
14	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. <i>Wellcome Open Research</i> , 2019, 4, 149.	1.8	3
15	Refinement of the critical genomic region for congenital hyperinsulinism in the Chromosome 9p deletion syndrome. <i>Wellcome Open Research</i> , 2019, 4, 149.	1.8	5
16	<i>MAFA</i> missense mutation causes familial insulinomatosis and diabetes mellitus. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2018, 115, 1027-1032.	7.1	88
17	Phenotype of CNTNAP1: a study of patients demonstrating a specific severe congenital hypomyelinating neuropathy with survival beyond infancy. <i>European Journal of Human Genetics</i> , 2018, 26, 796-807.	2.8	13
18	Diagnosis of lethal or prenatal-onset autosomal recessive disorders by parental exome sequencing. <i>Prenatal Diagnosis</i> , 2018, 38, 33-43.	2.3	64

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19	Missense Mutations of the Pro65 Residue of PCGF2 Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. <i>American Journal of Human Genetics</i> , 2018, 103, 786-793.	6.2	17
20	Dominant ER Stressâ€“Inducing <i>WFS1</i> Mutations Underlie a Genetic Syndrome of Neonatal/Infancy-Onset Diabetes, Congenital Sensorineural Deafness, and Congenital Cataracts. <i>Diabetes</i> , 2017, 66, 2044-2053.	0.6	77
21	Polycystic Kidney Disease with Hyperinsulinemic Hypoglycemia Caused by a Promoter Mutation in Phosphomannomutase 2. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 2529-2539.	6.1	99
22	Exome sequencing reveals a de novo POLD1 mutation causing phenotypic variability in mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (MDPL). <i>Metabolism: Clinical and Experimental</i> , 2017, 71, 213-225.	3.4	43
23	A <i>CACNA1D</i> mutation in a patient with persistent hyperinsulinaemic hypoglycaemia, heart defects, and severe hypotonia. <i>Pediatric Diabetes</i> , 2017, 18, 320-323.	2.9	67
24	Analysis of cell-free fetal DNA for non-invasive prenatal diagnosis in a family with neonatal diabetes. <i>Diabetic Medicine</i> , 2017, 34, 582-585.	2.3	27
25	<i>SOS1</i> frameshift mutations cause pure mucosal neuroma syndrome, a clinical phenotype distinct from multiple endocrine neoplasia type 2B. <i>Clinical Endocrinology</i> , 2016, 84, 715-719.	2.4	11
26	Germline or somatic GPR101 duplication leads to X-linked acroigantism: a clinico-pathological and genetic study. <i>Acta Neuropathologica Communications</i> , 2016, 4, 56.	5.2	110
27	Isolated Pancreatic Aplasia Due to a Hypomorphic <i>PTF1A</i> Mutation. <i>Diabetes</i> , 2016, 65, 2810-2815.	0.6	22
28	A comparison of mitochondrial DNA isolation methods in frozen post-mortem human brain tissueâ€“applications for studies of mitochondrial genetics in brain disorders. <i>BioTechniques</i> , 2015, 59, 241-246.	1.8	17
29	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. <i>European Journal of Human Genetics</i> , 2015, 23, 401-404.	2.8	51
30	Biallelic RFX6 mutations can cause childhood as well as neonatal onset diabetes mellitus. <i>European Journal of Human Genetics</i> , 2015, 23, 1744-1748.	2.8	34
31	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. <i>Nature Genetics</i> , 2014, 46, 61-64.	21.4	255
32	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. <i>Nature Genetics</i> , 2014, 46, 812-814.	21.4	411
33	An in-frame deletion at the polymerase active site of POLD1 causes a multisystem disorder with lipodystrophy. <i>Nature Genetics</i> , 2013, 45, 947-950.	21.4	151
34	An enhanced method for targeted next generation sequencing copy number variant detection using ExomeDepth. <i>Wellcome Open Research</i> , 0, 2, 49.	1.8	4