

Richard Caswell

List of Publications by Year in descending order

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34
papers

1,767
citations

471061

17
h-index

433756

31
g-index

36
all docs

36
docs citations

36
times ranked

3778
citing authors

#	ARTICLE	IF	CITATIONS
1	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. <i>Nature Genetics</i> , 2014, 46, 812-814.	9.4	411
2	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. <i>Nature Genetics</i> , 2014, 46, 61-64.	9.4	255
3	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.	9.4	151
4	Germline or somatic GPR101 duplication leads to X-linked acrogermism: a clinico-pathological and genetic study. <i>Acta Neuropathologica Communications</i> , 2016, 4, 56.	2.4	110
5	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.	3.0	99
6	<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.	3.3	88
7	Dominant ER Stress-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.	0.3	77
8	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.	1.2	67
9	Diagnosis of lethal or prenatal-onset autosomal recessive disorders by parental exome sequencing. <i>Prenatal Diagnosis</i> , 2018, 38, 33-43.	1.1	64
10	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. <i>European Journal of Human Genetics</i> , 2015, 23, 401-404.	1.4	51
11	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.	1.5	43
12	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.	2.6	43
13	Biallelic RFX6 mutations can cause childhood as well as neonatal onset diabetes mellitus. <i>European Journal of Human Genetics</i> , 2015, 23, 1744-1748.	1.4	34
14	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.	0.3	29
15	Analysis of cell-free fetal <i>scp</i> DNA for non-invasive prenatal diagnosis in a family with neonatal diabetes. <i>Diabetic Medicine</i> , 2017, 34, 582-585.	1.2	27
16	Mitochondrial Retinopathy. <i>Ophthalmology Retina</i> , 2022, 6, 65-79.	1.2	26
17	Isolated Pancreatic Aplasia Due to a Hypomorphic <i>PTF1A</i> Mutation. <i>Diabetes</i> , 2016, 65, 2810-2815.	0.3	22
18	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.	1.1	20

#	ARTICLE	IF	CITATIONS
19	A comparison of mitochondrial DNA isolation methods in frozen post-mortem human brain tissue – applications for studies of mitochondrial genetics in brain disorders. <i>BioTechniques</i> , 2015, 59, 241-246.	0.8	17
20	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.	2.6	17
21	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.	0.7	15
22	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.	0.1	14
23	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.	1.4	13
24	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.	4.3	13
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.	1.2	11
26	Late-onset Pseudoxanthoma Elasticum Associated with a Hypomorphic <i>ABCC6</i> Variant. <i>American Journal of Ophthalmology</i> , 2020, 218, 255-260.	1.7	11
27	A hypomorphic allele of <i>SLC35D1</i> results in Schneckenbecken-like dysplasia. <i>Human Molecular Genetics</i> , 2019, 28, 3543-3551.	1.4	9
28	Copy number variation of <i>LINGO1</i> in familial dystonic tremor. <i>Neurology: Genetics</i> , 2019, 5, e307.	0.9	8
29	Refinement of the critical genomic region for congenital hyperinsulinism in the Chromosome 9p deletion syndrome. <i>Wellcome Open Research</i> , 2019, 4, 149.	0.9	5
30	An enhanced method for targeted next generation sequencing copy number variant detection using ExomeDepth. <i>Wellcome Open Research</i> , 0, 2, 49.	0.9	4
31	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. <i>Wellcome Open Research</i> , 2019, 4, 149.	0.9	3
32	Mild MDPL in a patient with a novel de novo missense variant in the Cys-B region of <i>POLD1</i> . <i>European Journal of Human Genetics</i> , 2022, 30, 960-966.	1.4	2
33	Response to Comment on Misra et al. 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. <i>Diabetes Care</i> , 2020, 43, e155-e156.	4.3	0
34	Robinow syndrome in an extremely preterm infant: Novel homozygous <i>ROR2</i> variant detected by rapid exome sequencing. <i>American Journal of Medical Genetics, Part A</i> , 2021, , .	0.7	0