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	Activating germline mutations in STAT3 cause early-onset multi-organ autoimmune disease. Nature Genetics, 2014, 46, 812-814.	21.4	411
2	Recessive mutations in a distal PTF1A enhancer cause isolated pancreatic agenesis. Nature Genetics, 2014, 46, 61-64.	21.4	255
3	An in-frame deletion at the polymerase active site of POLD1 causes a multisystem disorder with lipodystrophy. Nature Genetics, 2013, 45, 947-950.	21.4	151
4	Germline or somatic GPR101 duplication leads to X-linked acrogigantism: a clinico-pathological and genetic study. Acta Neuropathologica Communications, 2016, 4, 56.	5.2	110
5	Polycystic Kidney Disease with Hyperinsulinemic Hypoglycemia Caused by a Promoter Mutation in Phosphomannomutase 2. Journal of the American Society of Nephrology: JASN, 2017, 28, 2529-2539.	6.1	99
6	MAFA missense mutation causes familial insulinomatosis and diabetes mellitus. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 1027-1032.	7.1	88
7	Dominant ER Stressâ€“Inducing WFS1 Mutations Underlie a Genetic Syndrome of Neonatal/Infancy-Onset Diabetes, Congenital Sensorineural Deafness, and Congenital Cataracts. Diabetes, 2017, 66, 2044-2053.	0.6	77
8	A CACNA1D mutation in a patient with persistent hyperinsulinaemic hypoglycaemia, heart defects, and severe hypotonia. Pediatric Diabetes, 2017, 18, 320-323.	2.9	67
9	Diagnosis of lethal or prenatalâ€onset autosomal recessive disorders by parental exome sequencing. Prenatal Diagnosis, 2018, 38, 33-43.	2.3	64
10	An exome sequencing strategy to diagnose lethal autosomal recessive disorders. European Journal of Human Genetics, 2015, 23, 401-404.	2.8	51
11	Exome sequencing reveals a de novo POLD1 mutation causing phenotypic variability in mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome (MDPL). Metabolism: Clinical and Experimental, 2017, 71, 213-225.	3.4	43
12	A Specific CNOT1 Mutation Results in a Novel Syndrome of Pancreatic Agenesis and Holoprosencephaly through Impaired Pancreatic and Neurological Development. American Journal of Human Genetics, 2019, 104, 985-989.	6.2	43
13	Biallelic RFX6 mutations can cause childhood as well as neonatal onset diabetes mellitus. European Journal of Human Genetics, 2015, 23, 1744-1748.	2.8	34
14	De Novo Mutations in EIF2B1 Affecting eIF2 Signaling Cause Neonatal/Early-Onset Diabetes and Transient Hepatic Dysfunction. Diabetes, 2020, 69, 477-483.	0.6	29
15	Analysis of cellâ€free fetal <scp>DNA</scp> for nonâ€invasive prenatal diagnosis in a family with neonatal diabetes. Diabetic Medicine, 2017, 34, 582-585.	2.3	27
16	Mitochondrial Retinopathy. Ophthalmology Retina, 2022, 6, 65-79.	2.4	26
17	Isolated Pancreatic Aplasia Due to a Hypomorphic PTF1A Mutation. Diabetes, 2016, 65, 2810-2815.	0.6	22
18	Classification and correlation of RYR2 missense variants in individuals with catecholaminergic polymorphic ventricular tachycardia reveals phenotypic relationships. Journal of Human Genetics, 2020, 65, 531-539.	2.3	20

#	Article	IF	Citations
19	A comparison of mitochondrial DNA isolation methods in frozen post-mortem human brain tissueâ€"applications for studies of mitochondrial genetics in brain disorders. BioTechniques, 2015, 59, 241-246.	1.8	17
20	Missense Mutations of the Pro65 Residue of PCGF2 Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. American Journal of Human Genetics, 2018, 103, 786-793.	6.2	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. European Journal of Medical Genetics, 2019, 62, 97-102.	1.3	15
22	Using Structural Analysis In Silico to Assess the Impact of Missense Variants in MEN1. Journal of the Endocrine Society, 2019, 3, 2258-2275.	0.2	14
23	Phenotype of CNTNAP1: a study of patients demonstrating a specific severe congenital hypomyelinating neuropathy with survival beyond infancy. European Journal of Human Genetics, 2018, 26, 796-807.	2.8	13
24	Homozygous Hypomorphic HNF1A Alleles Are a Novel Cause of Young-Onset Diabetes and Result in Sulfonylurea-Sensitive Diabetes. Diabetes Care, 2020, 43, 909-912.	8.6	13
25	<scp>SOS</scp>1 frameshift mutations cause pure mucosal neuroma syndrome, a clinical phenotype distinct from multiple endocrine neoplasia type 2B. Clinical Endocrinology, 2016, 84, 715-719.	2.4	11
26	Late-onset Pseudoxanthoma Elasticum Associated with a Hypomorphic ABCC6 Variant. American Journal of Ophthalmology, 2020, 218, 255-260.	3.3	11
27	A hypomorphic allele of SLC35D1 results in Schneckenbecken-like dysplasia. Human Molecular Genetics, 2019, 28, 3543-3551.	2.9	9
28	Copy number variation of LINGO1 in familial dystonic tremor. Neurology: Genetics, 2019, 5, e307.	1.9	8
29	Refinement of the critical genomic region for congenital hyperinsulinismÂin the Chromosome 9p deletion syndrome. Wellcome Open Research, 2019, 4, 149.	1.8	5
30	An enhanced method for targeted next generation sequencing copy number variant detection using ExomeDepth. Wellcome Open Research, 0, 2, 49.	1.8	4
31	Refinement of the critical genomic region for hypoglycaemia in the Chromosome 9p deletion syndrome. Wellcome Open Research, 2019, 4, 149.	1.8	3
32	Mild MDPL in a patient with a novel de novo missense variant in the Cys-B region of POLD1. European Journal of Human Genetics, 2022, 30, 960-966.	2.8	2
33	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. Diabetes Care 2020;43:909â€"912. Diabetes Care, 2020, 43, e155-e156.	8.6	0
34	Robinow syndrome in an extremely preterm infant: Novel homozygous ROR2 variant detected by rapid exome sequencing. American Journal of Medical Genetics, Part A, 2021, , .	1.2	0