

Charles C Searby

List of Publications by Year in descending order

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70
papers

6,719
citations

76326

40
h-index

118850

62
g-index

73
all docs

73
docs citations

73
times ranked

5944
citing authors

#	ARTICLE	IF	CITATIONS
1	Mutation of a nuclear receptor gene, NR2E3, causes enhanced S cone syndrome, a disorder of retinal cell fate. <i>Nature Genetics</i> , 2000, 24, 127-131.	21.4	439
2	The forkhead transcription factor gene FKHL7 is responsible for glaucoma phenotypes which map to 6p25. <i>Nature Genetics</i> , 1998, 19, 140-147.	21.4	416
3	<i>Bbs2</i> -null mice have neurosensory deficits, a defect in social dominance, and retinopathy associated with mislocalization of rhodopsin. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2004, 101, 16588-16593.	7.1	345
4	Identification of the gene (BBS1) most commonly involved in Bardet-Biedl syndrome, a complex human obesity syndrome. <i>Nature Genetics</i> , 2002, 31, 435-438.	21.4	327
5	A BBSome Subunit Links Ciliogenesis, Microtubule Stability, and Acetylation. <i>Developmental Cell</i> , 2008, 15, 854-865.	7.0	272
6	Identification of the gene that, when mutated, causes the human obesity syndrome BBS4. <i>Nature Genetics</i> , 2001, 28, 188-191.	21.4	254
7	Reduction of ER stress via a chemical chaperone prevents disease phenotypes in a mouse model of primary open angle glaucoma. <i>Journal of Clinical Investigation</i> , 2011, 121, 3542-3553.	8.2	249
8	A knockin mouse model of the Bardet-Biedl syndrome 1 M390R mutation has cilia defects, ventriculomegaly, retinopathy, and obesity. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2007, 104, 19422-19427.	7.1	237
9	Comparative Genomic Analysis Identifies an ADP-Ribosylation Factor-like Gene as the Cause of Bardet-Biedl Syndrome (BBS3). <i>American Journal of Human Genetics</i> , 2004, 75, 475-484.	6.2	220
10	ARL13B, PDE6D, and CEP164 form a functional network for INPP5E ciliary targeting. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2012, 109, 19691-19696.	7.1	213
11	Comparative Genomics and Gene Expression Analysis Identifies BBS9, a New Bardet-Biedl Syndrome Gene. <i>American Journal of Human Genetics</i> , 2005, 77, 1021-1033.	6.2	194
12	A Spectrum of FOXC1 Mutations Suggests Gene Dosage as a Mechanism for Developmental Defects of the Anterior Chamber of the Eye. <i>American Journal of Human Genetics</i> , 2001, 68, 364-372.	6.2	185
13	A Novel Protein LZTFL1 Regulates Ciliary Trafficking of the BBSome and Smoothens. <i>PLoS Genetics</i> , 2011, 7, e1002358.	3.5	182
14	<i>Mkks</i> -null mice have a phenotype resembling Bardet-Biedl syndrome. <i>Human Molecular Genetics</i> , 2005, 14, 1109-1118.	2.9	181
15	Compound Heterozygosity for Loss-of-Function Lysyl-tRNA Synthetase Mutations in a Patient with Peripheral Neuropathy. <i>American Journal of Human Genetics</i> , 2010, 87, 560-566.	6.2	169
16	BBS mutations modify phenotypic expression of CEP290-related ciliopathies. <i>Human Molecular Genetics</i> , 2014, 23, 40-51.	2.9	164
17	CRISPR-Cas9-based treatment of myocilin-associated glaucoma. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2017, 114, 11199-11204.	7.1	137
18	Bardet-Biedl syndrome 3 (<i>Bbs3</i>) knockout mouse model reveals common BBS-associated phenotypes and <i>Bbs3</i> unique phenotypes. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2011, 108, 20678-20683.	7.1	135

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19	Ocular-specific ER stress reduction rescues glaucoma in murine glucocorticoid-induced glaucoma. <i>Journal of Clinical Investigation</i> , 2014, 124, 1956-1965.	8.2	133
20	Point mutations of the connexin32 (GJB1) gene in X-linked dominant Charcot-Marie-Tooth neuropathy. <i>Human Molecular Genetics</i> , 1994, 3, 355-358.	2.9	128
21	Autosomal dominant Charcot-Marie-Tooth axonal neuropathy mapped on chromosome 7p (CMT2D). <i>Human Molecular Genetics</i> , 1996, 5, 1373-1375.	2.9	118
22	Evaluation of Complex Inheritance Involving the Most Common Bardet-Biedl Syndrome Locus (BBS1). <i>American Journal of Human Genetics</i> , 2003, 72, 429-437.	6.2	117
23	BBS7 is required for BBSome formation and its absence in mice results in Bardet-Biedl syndrome phenotypes and selective abnormalities in membrane protein trafficking. <i>Journal of Cell Science</i> , 2013, 126, 2372-80.	2.0	115
24	Abnormal development of NG2+PDGFR- β neural progenitor cells leads to neonatal hydrocephalus in a ciliopathy mouse model. <i>Nature Medicine</i> , 2012, 18, 1797-1804.	30.7	106
25	Loss of Bardet-Biedl syndrome proteins alters the morphology and function of motile cilia in airway epithelia. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008, 105, 3380-3385.	7.1	105
26	Glaucoma-causing myocilin mutants require the Peroxisomal targeting signal-1 receptor (PTS1R) to elevate intraocular pressure. <i>Human Molecular Genetics</i> , 2007, 16, 609-617.	2.9	101
27	Correlation between connexin 32 gene mutations and clinical phenotype in X-linked dominant Charcot-Marie-tooth neuropathy. , 1996, 63, 486-491.		84
28	Identification and Functional Analysis of the Vision-Specific BBS3 (ARL6) Long Isoform. <i>PLoS Genetics</i> , 2010, 6, e1000884.	3.5	75
29	Genetic interaction between Bardet-Biedl syndrome genes and implications for limb patterning. <i>Human Molecular Genetics</i> , 2008, 17, 1956-1967.	2.9	74
30	Discovery and Functional Analysis of a Retinitis Pigmentosa Gene, C2ORF71. <i>American Journal of Human Genetics</i> , 2010, 86, 686-695.	6.2	70
31	Transforming growth factor β 2 (TGF β 2) signaling plays a key role in glucocorticoid-induced ocular hypertension. <i>Journal of Biological Chemistry</i> , 2018, 293, 9854-9868.	3.4	68
32	Dejerine-Sottas neuropathy in mother and son with same point mutation of PMP22 gene. <i>Muscle and Nerve</i> , 1997, 20, 97-99.	2.2	67
33	Clinical evidence of decreased olfaction in Bardet-Biedl syndrome caused by a deletion in theBBS4Gene. <i>American Journal of Medical Genetics, Part A</i> , 2005, 132A, 343-346.	1.2	66
34	Screening of dominantly inherited Charcot-Marie-Tooth neuropathies. <i>Muscle and Nerve</i> , 1993, 16, 1232-1238.	2.2	62
35	BBSome function is required for both the morphogenesis and maintenance of the photoreceptor outer segment. <i>PLoS Genetics</i> , 2017, 13, e1007057.	3.5	60
36	Exposure to Static Magnetic and Electric Fields Treats Type 2 Diabetes. <i>Cell Metabolism</i> , 2020, 32, 561-574.e7.	16.2	55

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37	Impaired Function is a Common Feature of Neuropathy-Associated Glycyl-tRNA Synthetase Mutations. <i>Human Mutation</i> , 2014, 35, n/a-n/a.	2.5	51
38	The Cloning and Developmental Expression of Unconventional Myosin IXA (MYO9A) a Gene in the Bardet-Biedl Syndrome (BBS4) Region at Chromosome 15q22-q23. <i>Genomics</i> , 1999, 59, 150-160.	2.9	49
39	Charcot-Marie-Tooth neuropathy type 1A with both duplication and non-duplication. <i>Human Molecular Genetics</i> , 1993, 2, 405-410.	2.9	48
40	ATF4 leads to glaucoma by promoting protein synthesis and ER client protein load. <i>Nature Communications</i> , 2020, 11, 5594.	12.8	47
41	X-linked recessive Charcot-Marie-Tooth neuropathy: Clinical and genetic study. <i>Muscle and Nerve</i> , 1992, 15, 368-373.	2.2	43
42	Clinical presentation of a variant of Axenfeld-Rieger syndrome associated with subtelomeric 6p deletion. <i>European Journal of Medical Genetics</i> , 2007, 50, 120-127.	1.3	35
43	Autophagy stimulation reduces ocular hypertension in a murine glaucoma model via autophagic degradation of mutant myocilin. <i>JCI Insight</i> , 2021, 6, .	5.0	35
44	Disruption of RPGR protein interaction network is the common feature of RPGR missense variations that cause XLRP. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2019, 116, 1353-1360.	7.1	34
45	Simultaneous detection of the enzyme activities of GPx1 and GPx4 guide optimization of selenium in cell biological experiments. <i>Redox Biology</i> , 2020, 32, 101518.	9.0	34
46	The Centriolar Satellite Protein AZI1 Interacts with BBS4 and Regulates Ciliary Trafficking of the BBSome. <i>PLoS Genetics</i> , 2014, 10, e1004083.	3.5	33
47	The BBSome in POMC and AgRP Neurons Is Necessary for Body Weight Regulation and Sorting of Metabolic Receptors. <i>Diabetes</i> , 2019, 68, 1591-1603.	0.6	32
48	New mutations in BBS genes in small consanguineous families with Bardet-Biedl syndrome: detection of candidate regions by homozygosity mapping. <i>Molecular Vision</i> , 2010, 16, 137-43.	1.1	31
49	X-linked dominant charcot-marie-tooth neuropathy with 15 cases in a family genetic linkage study. <i>Muscle and Nerve</i> , 1988, 11, 1154-1156.	2.2	28
50	Ectopic Expression of Human BBS4 Can Rescue Bardet-Biedl Syndrome Phenotypes in Bbs4 Null Mice. <i>PLoS ONE</i> , 2013, 8, e59101.	2.5	23
51	Nuclear/cytoplasmic transport defects in BBS6 underlie congenital heart disease through perturbation of a chromatin remodeling protein. <i>PLoS Genetics</i> , 2017, 13, e1006936.	3.5	23
52	Disulfiram causes selective hypoxic cancer cell toxicity and radio-chemo-sensitization via redox cycling of copper. <i>Free Radical Biology and Medicine</i> , 2020, 150, 1-11.	2.9	22
53	Severe Charcot-Marie-Tooth neuropathy type 1A with 1-base pair deletion and frameshift mutation in the peripheral myelin protein 22 gene. , 1997, 20, 1308-1310.		20
54	A Dejerine-Sottas neuropathy family with a gene mapped on chromosome 8. , 1996, 19, 319-323.		19

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55	Linkage analysis of Charcot-Marie-Tooth neuropathy (HMSN type I). Journal of the Neurological Sciences, 1987, 80, 73-78.	0.6	10
56	Charcot-marie-tooth neuropathy related to chromosome 1. American Journal of Medical Genetics Part A, 1992, 42, 728-732.	2.4	10
57	Mutation in <i>CATIP</i> (C2orf62) causes oligoteratoasthenozoospermia by affecting actin dynamics. Journal of Medical Genetics, 2021, 58, 106-115.	3.2	8
58	A mouse model of Bardet-Biedl Syndrome has impaired fear memory, which is rescued by lithium treatment. PLoS Genetics, 2021, 17, e1009484.	3.5	8
59	Duchenne muscular dystrophy in monozygotic twins: Deletion of 5â€² fragments of the gene. American Journal of Medical Genetics Part A, 1989, 33, 113-116.	2.4	6
60	Ectopic expression of BBS1 rescues male infertility, but not retinal degeneration, in a BBS1 mouse model. Gene Therapy, 2022, 29, 227-235.	4.5	4
61	Counterpoint: An alternative hypothesis for why exposure to static magnetic and electric fields treats type 2 diabetes. American Journal of Physiology - Endocrinology and Metabolism, 2021, 320, E1001-E1002.	3.5	4
62	OMIGA 2.0. Biotech Software & Internet Report, 2000, 1, 198-207.	0.0	3
63	Becker muscular dystrophy recombinant DNA studies in identical twins. Muscle and Nerve, 1988, 11, 287-290.	2.2	2
64	Plasmid 1.1. Biotech Software & Internet Report, 2000, 1, 31-35.	0.0	1
65	Inherited metabolic myopathy with storage of glycoproteins and glycosaminoglycans. American Journal of Medical Genetics Part A, 1984, 18, 333-343.	2.4	0
66	Cyrillic 3 Standard Edition. Biotech Software & Internet Report, 2000, 1, 71-77.	0.0	0
67	Plasmid Premier 2.02. Biotech Software & Internet Report, 2001, 2, 64-70.	0.0	0
68	GenTerpret 1.26. Biotech Software & Internet Report, 2001, 2, 233-238.	0.0	0
69	Reply to Petersen et al.: An alternative hypothesis for why exposure to static magnetic and electric fields treats type 2 diabetes. American Journal of Physiology - Endocrinology and Metabolism, 2021, 320, E1004-E1005.	3.5	0
70	An open source and convenient method for the wide-spread testing of COVID-19 using deep throat sputum samples. PeerJ, 2022, 10, e13277.	2.0	0