Charles C Searby

List of Publications by Year
in descending order

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	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
2	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
3	Mutation in CATIP (C2orf62) causes oligoteratoasthenozoospermia by affecting actin dynamics. Journal of Medical Genetics, 2021, 58, 106-115.	3.2	8
4	Autophagy stimulation reduces ocular hypertension in a murine glaucoma model via autophagic degradation of mutant myocilin. JCI Insight, 2021, 6, .	5.0	35
5	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
6	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
7	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
8	Exposure to Static Magnetic and Electric Fields Treats Type 2 Diabetes. Cell Metabolism, 2020, 32, 561-574.e7.	16.2	55
9	ATF4 leads to glaucoma by promoting protein synthesis and ER client protein load. Nature Communications, 2020, 11, 5594.	12.8	47
10	Disulfiram causes selective hypoxic cancer cell toxicity and radio-chemo-sensitization via redox cycling of copper. Free Radical Biology and Medicine, 2020, 150, 1-11.	2.9	22
11	Simultaneous detection of the enzyme activities of GPx1 and GPx4 guide optimization of selenium in cell biological experiments. Redox Biology, 2020, 32, 101518.	9.0	34
12	The BBSome in POMC and AgRP Neurons Is Necessary for Body Weight Regulation and Sorting of Metabolic Receptors. Diabetes, 2019, 68, 1591-1603.	0.6	32
13	Disruption of RPGR protein interaction network is the common feature of RPGR missense variations that cause XLRP. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 1353-1360.	7.1	34
14	Transforming growth factor Î²2 (TGFÎ²2) signaling plays a key role in glucocorticoid-induced ocular hypertension. Journal of Biological Chemistry, 2018, 293, 9854-9868.	3.4	68
15	CRISPR-Cas9â€“based treatment of myocilin-associated glaucoma. Proceedings of the National Academy of Sciences of the United States of America, 2017, 114, 11199-11204.	7.1	137
16	BBSome function is required for both the morphogenesis and maintenance of the photoreceptor outer segment. PLoS Genetics, 2017, 13, e1007057.	3.5	60
17	Nuclear/cytoplasmic transport defects in BBS6 underlie congenital heart disease through perturbation of a chromatin remodeling protein. PLoS Genetics, 2017, 13, e1006936.	3.5	23
18	BBS mutations modify phenotypic expression of CEP290-related ciliopathies. Human Molecular Genetics, 2014, 23, 40-51.	2.9	164

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19	Impaired Function is a Common Feature of Neuropathy-Associated Glycyl-tRNA Synthetase Mutations. Human Mutation, 2014, 35, n/a-n/a.	2.5	51
20	The Centriolar Satellite Protein AZI1 Interacts with BBS4 and Regulates Ciliary Trafficking of the BBSome. PLoS Genetics, 2014, 10, e1004083.	3.5	33
21	Ocular-specific ER stress reduction rescues glaucoma in murine glucocorticoid-induced glaucoma. Journal of Clinical Investigation, 2014, 124, 1956-1965.	8.2	133
22	BBS7 is required for BBSome formation and its absence in mice results in Bardet-Biedl syndrome phenotypes and selective abnormalities in membrane protein trafficking. Journal of Cell Science, 2013, 126, 2372-80.	2.0	115
23	Ectopic Expression of Human BBS4 Can Rescue Bardet-Biedl Syndrome Phenotypes in Bbs4 Null Mice. PLoS ONE, 2013, 8, e59101.	2.5	23
24	ARL13B, PDE6D, and CEP164 form a functional network for INPP5E ciliary targeting. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 19691-19696.	7.1	213
25	Abnormal development of NG2+PDGFR-Î±+ neural progenitor cells leads to neonatal hydrocephalus in a ciliopathy mouse model. Nature Medicine, 2012, 18, 1797-1804.	30.7	106
26	Bardet-Biedl syndrome 3 (Bbs3) knockout mouse model reveals common BBS-associated phenotypes and Bbs3 unique phenotypes. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 20678-20683.	7.1	135
27	A Novel Protein LZTFL1 Regulates Ciliary Trafficking of the BBSome and Smoothened. PLoS Genetics, 2011, 7, e1002358.	3.5	182
28	Reduction of ER stress via a chemical chaperone prevents disease phenotypes in a mouse model of primary open angle glaucoma. Journal of Clinical Investigation, 2011, 121, 3542-3553.	8.2	249
29	Discovery and Functional Analysis of a Retinitis Pigmentosa Gene, C2ORF71. American Journal of Human Genetics, 2010, 86, 686-695.	6.2	70
30	Compound Heterozygosity for Loss-of-Function Lysyl-tRNA Synthetase Mutations in a Patient with Peripheral Neuropathy. American Journal of Human Genetics, 2010, 87, 560-566.	6.2	169
31	Identification and Functional Analysis of the Vision-Specific BBS3 (ARL6) Long Isoform. PLoS Genetics, 2010, 6, e1000884.	3.5	75
32	New mutations in BBS genes in small consanguineous families with Bardet-Biedl syndrome: detection of candidate regions by homozygosity mapping. Molecular Vision, 2010, 16, 137-43.	1.1	31
33	A BBSome Subunit Links Ciliogenesis, Microtubule Stability, and Acetylation. Developmental Cell, 2008, 15, 854-865.	7.0	272
34	Loss of Bardetâ€“Biedl syndrome proteins alters the morphology and function of motile cilia in airway epithelia. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 3380-3385.	7.1	105
35	Genetic interaction between Bardet-Biedl syndrome genes and implications for limb patterning. Human Molecular Genetics, 2008, 17, 1956-1967.	2.9	74
36	A knockin mouse model of the Bardetâ€“Biedl syndrome 1 M390R mutation has cilia defects, ventriculomegaly, retinopathy, and obesity. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 19422-19427.	7.1	237

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37	Glaucoma-causing myocilin mutants require the Peroxisomal targeting signal-1 receptor (PTS1R) to elevate intraocular pressure. Human Molecular Genetics, 2007, 16, 609-617.	2.9	101
38	Clinical presentation of a variant of Axenfeldâ€“Rieger syndrome associated with subtelomeric 6p deletion. European Journal of Medical Genetics, 2007, 50, 120-127.	1.3	35
39	Clinical evidence of decreased olfaction in Bardet-Biedl syndrome caused by a deletion in theBBS4Gene. American Journal of Medical Genetics, Part A, 2005, 132A, 343-346.	1.2	66
40	Mkks-null mice have a phenotype resembling Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2005, 14, 1109-1118.	2.9	181
41	Comparative Genomics and Gene Expression Analysis Identifies BBS9, a New Bardet-Biedl Syndrome Gene. American Journal of Human Genetics, 2005, 77, 1021-1033.	6.2	194
42	Bbs2 -null mice have neurosensory deficits, a defect in social dominance, and retinopathy associated with mislocalization of rhodopsin. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 16588-16593.	7.1	345
43	Comparative Genomic Analysis Identifies an ADP-Ribosylation Factorâ€“like Gene as the Cause of Bardet-Biedl Syndrome (BBS3). American Journal of Human Genetics, 2004, 75, 475-484.	6.2	220
44	Evaluation of Complex Inheritance Involving the Most Common Bardet-Biedl Syndrome Locus (BBS1). American Journal of Human Genetics, 2003, 72, 429-437.	6.2	117
45	Identification of the gene (BBS1) most commonly involved in Bardet-Biedl syndrome, a complex human obesity syndrome. Nature Genetics, 2002, 31, 435-438.	21.4	327
46	A Spectrum of FOXC1 Mutations Suggests Gene Dosage as a Mechanism for Developmental Defects of the Anterior Chamber of the Eye. American Journal of Human Genetics, 2001, 68, 364-372.	6.2	185
47	Plasmid Premier 2.02. Biotech Software & Internet Report, 2001, 2, 64-70.	0.0	0
48	GenTerpret 1.26. Biotech Software & Internet Report, 2001, 2, 233-238.	0.0	0
49	Identification of the gene that, when mutated, causes the human obesity syndrome BBS4. Nature Genetics, 2001, 28, 188-191.	21.4	254
50	Cyrillic 3 Standard Edition. Biotech Software & Internet Report, 2000, 1, 71-77.	0.0	0
51	Mutation of a nuclear receptor gene, NR2E3, causes enhanced S cone syndrome, a disorder of retinal cell fate. Nature Genetics, 2000, 24, 127-131.	21.4	439
52	Plasmid 1.1. Biotech Software & Internet Report, 2000, 1, 31-35.	0.0	1
53	OMIGA 2.0. Biotech Software & Internet Report, 2000, 1, 198-207.	0.0	3
54	The Cloning and Developmental Expression of Unconventional Myosin IXA (MYO9A) a Gene in the Bardetâ€“Biedl Syndrome (BBS4) Region at Chromosome 15q22â€“q23. Genomics, 1999, 59, 150-160.	2.9	49

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55	The forkhead transcription factor gene FKHL7 is responsible for glaucoma phenotypes which map to 6p25. Nature Genetics, 1998, 19, 140-147.	21.4	416
56	Dejerine-Sottas neuropathy in mother and son with same point mutation of PMP22 gene. Muscle and Nerve, 1997, 20, 97-99.	2.2	67
57	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
58	Correlation between connexin 32 gene mutations and clinical phenotype in X-linked dominant Charcot-Marie-tooth neuropathy. , 1996, 63, 486-491.		84
59	A Dejerine-Sottas neuropathy family with a gene mapped on chromosome 8. , 1996, 19, 319-323.		19
60	Autosomal dominant Charcot-Marie-Tooth axonal neuropathy mapped on chromosome 7p (CMT2D). Human Molecular Genetics, 1996, 5, 1373-1375.	2.9	118
61	Point mutations of the connexin32 (GJB1) gene in X-linked dominant Charcot â€" Marie â€" Tooth neuropathy. Human Molecular Genetics, 1994, 3, 355-358.	2.9	128
62	Screening of dominantly inherited Charcot-Marie-Tooth neuropathies. Muscle and Nerve, 1993, 16, 1232-1238.	2.2	62
63	Charcot â€" Marie â€" Tooth neuropathy type 1A with both duplication and non-duplication. Human Molecular Genetics, 1993, 2, 405-410.	2.9	48
64	X-linked recessive Charcot-Marie-Tooth neuropathy: Clinical and genetic study. Muscle and Nerve, 1992, 15, 368-373.	2.2	43
65	Charcot-marie-tooth neuropathy related to chromosome 1. American Journal of Medical Genetics Part A, 1992, 42, 728-732.	2.4	10
66	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
67	Becker muscular dystrophy recombinant DNA studies in identical twins. Muscle and Nerve, 1988, 11, 287-290.	2.2	2
68	X-linked dominant charcot-marie-tooth neuropathy with 15 cases in a family genetic linkage study. Muscle and Nerve, 1988, 11, 1154-1156.	2.2	28
69	Linkage analysis of Charcot-Marie-Tooth neuropathy (HMSN type I). Journal of the Neurological Sciences, 1987, 80, 73-78.	0.6	10
70	Inherited metabolic myopathy with storage of glycoproteins and glycosaminoglycans. American Journal of Medical Genetics Part A, 1984, 18, 333-343.	2.4	0