Val C Sheffield

List of Publications by Year
in descending order

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273 papers	36,015 citations	2544 96 h-index	3650 180 g-index
278 all docs	278 docs citations	278 times ranked	26876 citing authors

#	Article	IF	Citations
1	Gene therapy and gene correction: targets, progress, and challenges for treating human diseases. Gene Therapy, 2022, 29, 3-12.	4.5	53
2	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
3	Consensus Recommendation for Mouse Models of Ocular Hypertension to Study Aqueous Humor Outflow and Its Mechanisms. , 2022, 63, 12.		20
4	Retinal ciliopathies through the lens of Bardet-Biedl Syndrome: Past, present and future. Progress in Retinal and Eye Research, 2022, 89, 101035.	15.5	17
5	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
6	Mutation in CATIP (C2orf62) causes oligoteratoasthenozoospermia by affecting actin dynamics. Journal of Medical Genetics, 2021, 58, 106-115.	3.2	8
7	Autophagy stimulation reduces ocular hypertension in a murine glaucoma model via autophagic degradation of mutant myocilin. JCI Insight, 2021, 6, .	5.0	35
8	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
9	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
10	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
11	Exome-based investigation of the genetic basis of human pigmentary glaucoma. BMC Genomics, 2021, 22, 477.	2.8	9
12	Gene therapy rescues olfactory perception in a clinically relevant ciliopathy model of Bardetâ€“Biedl syndrome. FASEB Journal, 2021, 35, e21766.	0.5	8
13	Photoreceptor cilia, in contrast to primary cilia, grant entry to a partially assembled BBSome. Human Molecular Genetics, 2021, 30, 87-102.	2.9	11
14	Exposure to Static Magnetic and Electric Fields Treats Type 2 Diabetes. Cell Metabolism, 2020, 32, 561-574.e7.	16.2	55
15	ATF4 leads to glaucoma by promoting protein synthesis and ER client protein load. Nature Communications, 2020, 11, 5594.	12.8	47
16	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
17	A genome-wide association study implicates the BMP7 locus as a risk factor for nonsyndromic metopic craniosynostosis. Human Genetics, 2020, 139, 1077-1090.	3.8	24
18	The absence of BBSome function decreases synaptogenesis and causes ectopic synapse formation in the retina. Scientific Reports, 2020, 10, 8321.	3.3	15

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19	Topical Ocular Delivery of Nanocarriers: A Feasible Choice for Glaucoma Management. Current Pharmaceutical Design, 2020, 26, 5518-5532.	1.9	2
20	BBS4 is required for IFT coordination and basal body number in mammalian olfactory cilia.. Journal of Cell Science, 2019, 132, .	2.0	27
21	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
22	Development of a Molecularly Stable Gene Therapy Vector for the Treatment of RPGR-Associated X-Linked Retinitis Pigmentosa. Human Gene Therapy, 2019, 30, 967-974.	2.7	16
23	Absence of BBSome function leads to astrocyte reactivity in the brain. Molecular Brain, 2019, 12, 48.	2.6	14
24	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
25	A 30-Mile-per-Hour Headwind. JAMA - Journal of the American Medical Association, 2018, 320, 33.	7.4	0
26	Osteoarthritis-Like Changes in Bardetâ€“Biedl Syndrome Mutant Ciliopathy Mice (Bbs1M390R/M390R): Evidence for a Role of Primary Cilia in Cartilage Homeostasis and Regulation of Inflammation. Frontiers in Physiology, 2018, 9, 708.	2.8	14
27	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
28	Genotypic and phenotypic characterization of the Sdccag8Tn(sb-Tyr)2161B.CA1C2Ove mouse model. PLoS ONE, 2018, 13, e0192755.	2.5	9
29	Bardet-Biedl syndrome 3 regulates the development of cranial base midline structures. Bone, 2017, 101, 179-190.	2.9	10
30	Gene Therapeutic Reversal of Peripheral Olfactory Impairment in Bardet-Biedl Syndrome. Molecular Therapy, 2017, 25, 904-916.	8.2	41
31	Clinically Focused Molecular Investigation of 1000 Consecutive Families with Inherited Retinal Disease. Ophthalmology, 2017, 124, 1314-1331.	5.2	312
32	Mutation in TDRD9 causes non-obstructive azoospermia in infertile men. Journal of Medical Genetics, 2017, 54, 633-639.	3.2	107
33	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
34	The molecular genetics of eye diseases. Human Molecular Genetics, 2017, 26, R1-R1.	2.9	5
35	BBSome function is required for both the morphogenesis and maintenance of the photoreceptor outer segment. PLoS Genetics, 2017, 13, e1007057.	3.5	60
36	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

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37	Restoration of Aqueous Humor Outflow Following Transplantation of iPSC-Derived Trabecular Meshwork Cells in a Transgenic Mouse Model of Glaucoma. , 2017, 58, 2054.		76
38	Keeping an Eye on Bardet-Biedl Syndrome: A Comprehensive Review of the Role of Bardet-Biedl Syndrome Genes in the Eye. Medical Research Archives, 2017, 5, .	0.2	45
39	The BBSome Controls Energy Homeostasis by Mediating the Transport of the Leptin Receptor to the Plasma Membrane. PLoS Genetics, 2016, 12, e1005890.	3.5	97
40	Mutations inC8ORF37cause Bardet Biedl syndrome (BBS21). Human Molecular Genetics, 2016, 25, 2283-2294.	2.9	91
41	Transplantation of iPSC-derived TM cells rescues glaucoma phenotypes in vivo. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E3492-500.	7.1	89
42	A HomozygousNme7Mutation Is Associated withSitus Inversus Totalis. Human Mutation, 2016, 37, 727-731.	2.5	22
43	The Bardet-Biedl Syndrome. , 2016, , 237-240.		3
44	De Novo and Rare Variants at Multiple Loci Support the Oligogenic Origins of Atrioventricular Septal Heart Defects. PLoS Genetics, 2016, 12, e1005963.	3.5	92
45	Rat, Mouse, and Primate Models of Chronic Glaucoma Show Sustained Elevation of Extracellular ATP and Altered Purinergic Signaling in the Posterior Eye. , 2015, 56, 3075.		50
46	Regulation of Insulin Receptor Trafficking by Bardet Biedl Syndrome Proteins. PLoS Genetics, 2015, 11, e1005311.	3.5	57
47	CNV-ROC: A cost effective, computer-aided analytical performance evaluator of chromosomal microarrays. Journal of Biomedical Informatics, 2015, 54, 106-113.	4.3	0
48	Accumulation of non-outer segment proteins in the outer segment underlies photoreceptor degeneration in Bardetâ€“Biedl syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E4400-9.	7.1	123
49	PLEKHM2mutation leads to abnormal localization of lysosomes, impaired autophagy flux and associates with recessive dilated cardiomyopathy and left ventricular noncompaction. Human Molecular Genetics, 2015, 24, 7227-7240.	2.9	55
50	Calmodulin Methyltransferase Is Required for Growth, Muscle Strength, Somatosensory Development and Brain Function. PLoS Genetics, 2015, 11, e1005388.	3.5	16
51	Ciliopathy Is Differentially Distributed in the Brain of a Bardet-Biedl Syndrome Mouse Model. PLoS ONE, 2014, 9, e93484.	2.5	25
52	BBS mutations modify phenotypic expression of CEP290-related ciliopathies. Human Molecular Genetics, 2014, 23, 40-51.	2.9	164
53	The Centriolar Satellite Protein AZI1 Interacts with BBS4 and Regulates Ciliary Trafficking of the BBSome. PLoS Genetics, 2014, 10, e1004083.	3.5	33
54	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	9.6	101

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55	Mechanosensitive release of adenosine 5â€²â€triphosphate through pannexin channels and mechanosensitive upregulation of pannexin channels in optic nerve head astrocytes: A mechanism for purinergic involvement in chronic strain. Glia, 2014, 62, 1486-1501.	4.9	140
56	Functional characterization of Prickle2 and BBS7 identify overlapping phenotypes yet distinct mechanisms. Developmental Biology, 2014, 392, 245-255.	2.0	13
57	Ocular-specific ER stress reduction rescues glaucoma in murine glucocorticoid-induced glaucoma. Journal of Clinical Investigation, 2014, 124, 1956-1965.	8.2	133
58	Author reply. Ophthalmology, 2013, 120, e73.	5.2	0
59	Varied Clinical Presentations of Seven Patients With Mutations inCYP11A1Encoding the Cholesterol Side-Chain Cleavage Enzyme, P450scc. Journal of Clinical Endocrinology and Metabolism, 2013, 98, 713-720.	3.6	59
60	Non-exomic and synonymous variants in ABCA4 are an important cause of Stargardt disease. Human Molecular Genetics, 2013, 22, 5136-5145.	2.9	159
61	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
62	Congenital myopathy is caused by mutation of HACD1. Human Molecular Genetics, 2013, 22, 5229-5236.	2.9	48
63	Subretinal Gene Therapy of Mice With Bardet-Biedl Syndrome Type 1. , 2013, 54, 6118.		79
64	Ectopic Expression of Human BBS4 Can Rescue Bardet-Biedl Syndrome Phenotypes in Bbs4 Null Mice. PLoS ONE, 2013, 8, e59101.	2.5	23
65	A Genome-Wide Association Study for Primary Open Angle Glaucoma and Macular Degeneration Reveals Novel Loci. PLoS ONE, 2013, 8, e58657.	2.5	35
66	Calpain-5 Mutations Cause Autoimmune Uveitis, Retinal Neovascularization, and Photoreceptor Degeneration. PLoS Genetics, 2012, 8, e1003001.	3.5	76
67	BBS proteins interact genetically with the IFT pathway to influence SHH-related phenotypes. Human Molecular Genetics, 2012, 21, 1945-1953.	2.9	123
68	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	2.9	334
69	Topical Ocular Sodium 4-Phenylbutyrate Rescues Glaucoma in a Myocilin Mouse Model of Primary Open-Angle Glaucoma. , 2012, 53, 1557.		100
70	Intrinsic Protein-Protein Interaction-mediated and Chaperonin-assisted Sequential Assembly of Stable Bardet-Biedl Syndrome Protein Complex, the BBSome. Journal of Biological Chemistry, 2012, 287, 20625-20635.	3.4	142
71	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
72	Germline mosaic transmission of a novel duplication of PXDN and MYT1L to two male half-siblings with autism. Psychiatric Genetics, 2012, 22, 137-140.	1.1	27

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73	2q24 deletions: Further characterization of clinical findings and their relation to the SCN cluster. American Journal of Medical Genetics, Part A, 2012, 158A, 2767-2774.	1.2	4
74	Phenotypic expression of Bardetâ€“Biedl syndrome in patients homozygous for the common M390R mutation in the BBS1 gene. Vision Research, 2012, 75, 77-87.	1.4	34
75	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
76	Recommendations for Genetic Testing of Inherited Eye Diseases. Ophthalmology, 2012, 119, 2408-2410.	5.2	157
77	TUDCA Slows Retinal Degeneration in Two Different Mouse Models of Retinitis Pigmentosa and Prevents Obesity in Bardet-Biedl Syndrome Type 1 Mice. , 2012, 53, 100.		84
78	Bardet Biedl syndrome genes are required for autonomic control of the circulation. FASEB Journal, 2012, 26, 891.17.	0.5	0
79	Sequencing and disease variation detection tools and techniques. , 2011, , .		1
80	Genomics and the Eye. New England Journal of Medicine, 2011, 364, 1932-1942.	27.0	81
81	Mapping the NPHP-JBTS-MKS Protein Network Reveals Ciliopathy Disease Genes and Pathways. Cell, 2011, 145, 513-528.	28.9	531
82	Variations in NPHP5 in Patients With Nonsyndromic Leber Congenital Amaurosis and Senior-Loken Syndrome. JAMA Ophthalmology, 2011, 129, 81.	2.4	62
83	Primary Ciliary Dyskinesia Caused by Homozygous Mutation in DNAL1, Encoding Dynein Light Chain 1. American Journal of Human Genetics, 2011, 88, 599-607.	6.2	116
84	Genome-wide analysis of copy number variants in age-related macular degeneration. Human Genetics, 2011, 129, 91-100.	3.8	36
85	Autosomal recessive hyponatremia due to isolated salt wasting in sweat associated with a mutation in the active site of Carbonic Anhydrase 12. Human Genetics, 2011, 129, 397-405.	3.8	35
86	Microdeletion of 17q22q23.2 encompassing TBX2 and TBX4 in a patient with congenital microcephaly, thyroid duct cyst, sensorineural hearing loss, and pulmonary hypertension. American Journal of Medical Genetics, Part A, 2011, 155, 418-423.	1.2	53
87	Inactivation of Bardet-Biedl syndrome genes causes kidney defects. American Journal of Physiology - Renal Physiology, 2011, 300, F574-F580.	2.7	26
88	Copy Number Variations and Primary Open-Angle Glaucoma. , 2011, 52, 7122.		31
89	Functional analysis of BBS3 A89V that results in non-syndromic retinal degeneration. Human Molecular Genetics, 2011, 20, 1625-1632.	2.9	38
90	The N-terminal region of centrosomal protein 290 (CEP290) restores vision in a zebrafish model of human blindness. Human Molecular Genetics, 2011, 20, 1467-1477.	2.9	56

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91	Copy number variations on chromosome 12q14 in patients with normal tension glaucoma. Human Molecular Genetics, 2011, 20, 2482-2494.	2.9	189
92	Exome sequencing and analysis of induced pluripotent stem cells identify the cilia-related gene male germ cell-associated kinase (MAK) as a cause of retinitis pigmentosa. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, E569-76.	7.1	186
93	Primary cilia membrane assembly is initiated by Rab11 and transport protein particle II (TRAPPII) complex-dependent trafficking of Rabin8 to the centrosome. Proceedings of the National Academy of Sciences of the United States of America, 2011, 108, 2759-2764.	7.1	376
94	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
95	An ARL3â€“UNC119â€“RP2 GTPase cycle targets myristoylated NPHP3 to the primary cilium. Genes and Development, 2011, 25, 2347-2360.	5.9	202
96	A Novel Protein LZTFL1 Regulates Ciliary Trafficking of the BBSome and Smoothened. PLoS Genetics, 2011, 7, e1002358.	3.5	182
97	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
98	Evaluation of Embryonic and Perinatal Myosin Gene Mutations and the Etiology of Congenital Idiopathic Clubfoot. Journal of Pediatric Orthopaedics, 2010, 30, 231-234.	1.2	22
99	Evaluation of GPR50, hMel-1B, and ROR-Î± Melatonin-related Receptors and the Etiology of Adolescent Idiopathic Scoliosis. Journal of Pediatric Orthopaedics, 2010, 30, 539-543.	1.2	22
100	Light aversion in mice depends on nonimage-forming irradiance detection.. Behavioral Neuroscience, 2010, 124, 821-827.	1.2	33
101	Discovery and Functional Analysis of a Retinitis Pigmentosa Gene, C2ORF71. American Journal of Human Genetics, 2010, 86, 686-695.	6.2	70
102	Bardet-Biedl syndrome in Denmark-report of 13 novel sequence variations in six genes. Human Mutation, 2010, 31, 429-436.	2.5	72
103	Familial neonatal isolated cardiomyopathy caused by a mutation in the flavoprotein subunit of succinate dehydrogenase. European Journal of Human Genetics, 2010, 18, 1160-1165.	2.8	100
104	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	27.8	1,803
105	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	2.9	538
106	BBS6, BBS10, and BBS12 form a complex with CCT/TRiC family chaperonins and mediate BBSome assembly. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 1488-1493.	7.1	279
107	Deducing the pathogenic contribution of recessive ABCA4 alleles in an outbred population. Human Molecular Genetics, 2010, 19, 3693-3701.	2.9	53
108	A mouse model of osteochondromagenesis from clonal inactivation of Ext1 in chondrocytes. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 2054-2059.	7.1	109

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109	Identification and Functional Analysis of the Vision-Specific BBS3 (ARL6) Long Isoform. PLoS Genetics, 2010, 6, e1000884.	3.5	75
110	Contrasting vascular effects caused by loss of Bardet-Biedl syndrome genes. American Journal of Physiology - Heart and Circulatory Physiology, 2010, 299, H1902-H1907.	3.2	14
111	Recurrence risks for Bardet-Biedl syndrome: Implications of locus heterogeneity. Genetics in Medicine, 2010, 12, 623-627.	2.4	18
112	The blind leading the obese: the molecular pathophysiology of a human obesity syndrome. Transactions of the American Clinical and Climatological Association, 2010, 121, 172-81; discussion 181-2.	0.5	40
113	Requirement of Bardet-Biedl syndrome proteins for leptin receptor signaling. Human Molecular Genetics, 2009, 18, 1323-1331.	2.9	272
114	Mice defective in Trpm6 show embryonic mortality and neural tube defects. Human Molecular Genetics, 2009, 18, 4367-4375.	2.9	97
115	Cartilage abnormalities associated with defects of chondrocytic primary cilia in Bardetâ€Biedl syndrome mutant mice. Journal of Orthopaedic Research, 2009, 27, 1093-1099.	2.3	41
116	Evaluation of CAND2 and WNT7a as Candidate Genes for Congenital Idiopathic Clubfoot. Clinical Orthopaedics and Related Research, 2009, 467, 1201-1205.	1.5	15
117	A genome-wide linkage and association scan reveals novel loci for autism. Nature, 2009, 461, 802-808.	27.8	570
118	Novel mutations in BBS5 highlight the importance of this gene in nonâ€Caucasian Bardetâ€“Biedl syndrome patients. American Journal of Medical Genetics, Part A, 2008, 146A, 517-520.	1.2	18
119	Cortical enlargement in autism is associated with a functional VNTR in the monoamine oxidase A gene. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2008, 147B, 1145-1151.	1.7	55
120	Classical and melanopsin photoreception in irradiance detection: negative masking of locomotor activity by light. European Journal of Neuroscience, 2008, 27, 1973-1979.	2.6	39
121	A BBSome Subunit Links Ciliogenesis, Microtubule Stability, and Acetylation. Developmental Cell, 2008, 15, 854-865.	7.0	272
122	Evidence for a Novel X-Linked Modifier Locus for Leber Hereditary Optic Neuropathy. Ophthalmic Genetics, 2008, 29, 17-24.	1.2	105
123	Mutation in theSLC4A11Gene Associated with Autosomal Recessive Congenital Hereditary Endothelial Dystrophy in a Large Saudi Family. Ophthalmic Genetics, 2008, 29, 41-45.	1.2	34
124	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
125	Genetic interaction between Bardet-Biedl syndrome genes and implications for limb patterning. Human Molecular Genetics, 2008, 17, 1956-1967.	2.9	74
126	Association of a Novel Mutation in the Retinol Dehydrogenase 12 (RDH12) Gene With Autosomal Dominant Retinitis Pigmentosa. JAMA Ophthalmology, 2008, 126, 1301.	2.4	47

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127	Posterior probability of linkage analysis of autism dataset identifies linkage to chromosome 16. Psychiatric Genetics, 2008, 18, 85-91.	1.1	9
128	Leptin resistance contributes to obesity and hypertension in mouse models of Bardet-Biedl syndrome. Journal of Clinical Investigation, 2008, 118, 1458-1467.	8.2	201
129	Increased expression of the WNT antagonist sFRP-1 in glaucoma elevates intraocular pressure. Journal of Clinical Investigation, 2008, 118, 1056-64.	8.2	143
130	TRANSCRIPT ANNOTATION PRIORITIZATION AND SCREENING SYSTEM (TrAPSS) FOR MUTATION SCREENING. Journal of Bioinformatics and Computational Biology, 2007, 05, 1155-1172.	0.8	1
131	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
132	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
133	Complement Factor H Polymorphism p.Tyr402His and Cuticular Drusen. JAMA Ophthalmology, 2007, 125, 93.	2.4	48
134	No Association Between Variations in the WDR36 Gene and Primary Open-Angle Glaucoma. JAMA Ophthalmology, 2007, 125, 434.	2.4	58
135	Familial Cavitary Optic Disk Anomalies: Identification of a Novel Genetic Locus. American Journal of Ophthalmology, 2007, 143, 795-800.e1.	3.3	23
136	LOXL1 Mutations Are Associated with Exfoliation Syndrome in Patients from the Midwestern United States. American Journal of Ophthalmology, 2007, 144, 974-975.e1.	3.3	111
137	A Core Complex of BBS Proteins Cooperates with the GTPase Rab8 to Promote Ciliary Membrane Biogenesis. Cell, 2007, 129, 1201-1213.	28.9	1,248
138	Mitochondrial Variant G4132A is Associated with Familial Non-Arteritic Anterior Ischemic Optic Neuropathy in One Large Pedigree. Ophthalmic Genetics, 2007, 28, 1-7.	1.2	14
139	CHD7 Gene Polymorphisms Are Associated with Susceptibility to Idiopathic Scoliosis. American Journal of Human Genetics, 2007, 80, 957-965.	6.2	142
140	Gene Expression Analysis of Photoreceptor Cell Loss inBbs4-Knockout Mice Reveals an Early Stress Gene Response and Photoreceptor Cell Damage. , 2007, 48, 3329.		57
141	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	21.4	1,272
142	Autozygosity mapping of Bardetâ€“Biedl syndrome to 12q21.2 and confirmation of FLJ23560 as BBS10. European Journal of Human Genetics, 2007, 15, 173-178.	2.8	28
143	Systematic Screening for Subtelomeric Anomalies in a Clinical Sample of Autism. Journal of Autism and Developmental Disorders, 2007, 37, 703-708.	2.7	10
144	Case of Stargardt Disease Caused by Uniparental Isodisomy. JAMA Ophthalmology, 2006, 124, 744.	2.4	27

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145	Prioritizing regions of candidate genes for efficient mutation screening. Human Mutation, 2006, 27, 195-200.	2.5	10
146	Genome-wide identification of pseudogenes capable of disease-causing gene conversion. Human Mutation, 2006, 27, 545-552.	2.5	82
147	The C677T Variant in the Methylenetetrahydrofolate Reductase Gene Is Not Associated with Disease in Cohorts of Pseudoexfoliation Glaucoma and Primary Open-Angle Glaucoma Patients from Iowa. Ophthalmic Genetics, 2006, 27, 39-41.	1.2	22
148	Retinal Disease Expression in Bardet-Biedl Syndrome-1 (BBS1) Is a Spectrum from Maculopathy to Retina-Wide Degeneration. , 2006, 47, 5004.		83
149	Regulation of gene expression in the mammalian eye and its relevance to eye disease. Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 14429-14434.	7.1	190
150	Bardetâ€“Biedl syndrome genes are important in retrograde intracellular trafficking and Kupffer's vesicle cilia function. Human Molecular Genetics, 2006, 15, 667-677.	2.9	176
151	Homozygosity mapping with SNP arrays identifiesTRIM32, an E3 ubiquitin ligase, as a Bardetâ€“Biedl syndrome gene (BBS11). Proceedings of the National Academy of Sciences of the United States of America, 2006, 103, 6287-6292.	7.1	378
152	Molecular Genetics of Retinal Disease. , 2006, , 373-394.		5
153	Pathophysiological mechanisms of obesity and hypertension in mouse models of Bardetâ€Biedl syndrome. FASEB Journal, 2006, 20, A1207.	0.5	0
154	Ocular phenotypes of three genetic variants of Bardet-Biedl syndrome. , 2005, 132A, 283-287.		66
155	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
156	Evaluation of the chromosome 2q37.3 geneCENTG2as an autism susceptibility gene. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2005, 136B, 36-44.	1.7	60
157	A case of autism and uniparental disomy of chromosome 1. Human Genetics, 2005, 117, 200-206.	3.8	14
158	Mkks-null mice have a phenotype resembling Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2005, 14, 1109-1118.	2.9	181
159	Generation, characterization, and molecular cloning of theNoerg-1mutation of rhodopsin in the mouse. Visual Neuroscience, 2005, 22, 619-629.	1.0	21
160	A Constrained-Likelihood Approach to Marker-Trait Association Studies. American Journal of Human Genetics, 2005, 77, 768-780.	6.2	51
161	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
162	A comprehensive nonredundant expressed sequence tag collection for the developing Rattus norvegicus heart. Physiological Genomics, 2004, 17, 245-252.	2.3	7

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163	Use of Isolated Populations in the Study of a Human Obesity Syndrome, the Bardet-Biedl Syndrome. Pediatric Research, 2004, 55, 908-911.	2.3	23
164	Missense Variations in the Fibulin 5 Gene and Age-Related Macular Degeneration. New England Journal of Medicine, 2004, 351, 346-353.	27.0	298
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