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	Functional impact of global rare copy number variation in autism spectrum disorders. Nature, 2010, 466, 368-372.	27.8	1,803
2	Identification of a Gene That Causes Primary Open Angle Glaucoma. Science, 1997, 275, 668-670.	12.6	1,274
3	Mapping autism risk loci using genetic linkage and chromosomal rearrangements. Nature Genetics, 2007, 39, 319-328.	21.4	1,272
4	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
5	Pendred syndrome is caused by mutations in a putative sulphate transporter gene (PDS). Nature Genetics, 1997, 17, 411-422.	21.4	1,081
6	Comprehensive Human Genetic Maps: Individual and Sex-Specific Variation in Recombination. American Journal of Human Genetics, 1998, 63, 861-869.	6.2	1,042
7	The Sensitivity of Single-Strand Conformation Polymorphism Analysis for the Detection of Single Base Substitutions. Genomics, 1993, 16, 325-332.	2.9	653
8	A genome-wide linkage and association scan reveals novel loci for autism. Nature, 2009, 461, 802-808.	27.8	570
9	A genome-wide scan for common alleles affecting risk for autism. Human Molecular Genetics, 2010, 19, 4072-4082.	2.9	538
10	Mapping the NPHP-JBTS-MKS Protein Network Reveals Ciliopathy Disease Genes and Pathways. Cell, 2011, 145, 513-528.	28.9	531
11	The Pendred syndrome gene encodes a chloride-iodide transport protein. Nature Genetics, 1999, 21, 440-443.	21.4	524
12	Mutation of TRPM6 causes familial hypomagnesemia with secondary hypocalcemia. Nature Genetics, 2002, 31, 171-174.	21.4	506
13	Analysis of Myocilin Mutations in 1703 Glaucoma Patients From Five Different Populations. Human Molecular Genetics, 1999, 8, 899-905.	2.9	496
14	A single EFEMP1 mutation associated with both Malattia Leventinese and Doyne honeycomb retinal dystrophy. Nature Genetics, 1999, 22, 199-202.	21.4	453
15	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
16	Clinical Features Associated with Mutations in the Chromosome 1 Open-Angle Glaucoma Gene (GLC1A). New England Journal of Medicine, 1998, 338, 1022-1027.	27.0	423
17	The forkhead transcription factor gene FKHL7 is responsible for glaucoma phenotypes which map to 6p25. Nature Genetics, 1998, 19, 140-147.	21.4	416
18	Genetic linkage of familial open angle glaucoma to chromosome 1q21â€“q31. Nature Genetics, 1993, 4, 47-50.	21.4	410

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19	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
20	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
21	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
22	Individual common variants exert weak effects on the risk for autism spectrum disorders. Human Molecular Genetics, 2012, 21, 4781-4792.	2.9	334
23	Carrier Rates in the Midwestern United States for <EMPH TYPE="ITAL">GJB2</EMPH> Mutations Causing Inherited Deafness. JAMA - Journal of the American Medical Association, 1999, 281, 2211.	7.4	331
24	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
25	Clinically Focused Molecular Investigation of 1000 Consecutive Families with Inherited Retinal Disease. Ophthalmology, 2017, 124, 1314-1331.	5.2	312
26	Bardetâ€“Biedl syndrome type 4 (BBS4)-null mice implicate Bbs4 in flagella formation but not global cilia assembly. Proceedings of the National Academy of Sciences of the United States of America, 2004, 101, 8664-8669.	7.1	309
27	Missense Variations in the Fibulin 5 Gene and Age-Related Macular Degeneration. New England Journal of Medicine, 2004, 351, 346-353.	27.0	298
28	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
29	De novo mutations in the CRX homeobox gene associated with Leber congenital amaurosis. Nature Genetics, 1998, 18, 311-312.	21.4	276
30	Butterflyâ€“shaped pigment dystrophy of the fovea caused by a point mutation in codon 167 of the RDS gene. Nature Genetics, 1993, 3, 202-207.	21.4	272
31	A BBSome Subunit Links Ciliogenesis, Microtubule Stability, and Acetylation. Developmental Cell, 2008, 15, 854-865.	7.0	272
32	Requirement of Bardet-Biedl syndrome proteins for leptin receptor signaling. Human Molecular Genetics, 2009, 18, 1323-1331.	2.9	272
33	Pendred syndrome, DFNB4, andPDS/SLC26A4 identification of eight novel mutations and possible genotype-phenotype correlations. Human Mutation, 2001, 17, 403-411.	2.5	267
34	Mutations in MKKS cause Bardet-Biedl syndrome. Nature Genetics, 2000, 26, 15-16.	21.4	256
35	Identification of the gene that, when mutated, causes the human obesity syndrome BBS4. Nature Genetics, 2001, 28, 188-191.	21.4	254
36	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

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37	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
38	Genetic linkage of vitelliform macular degeneration (Best's disease) to chromosome 11q13. Nature Genetics, 1992, 1, 246-250.	21.4	234
39	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
40	The nuclear receptor NR2E3 plays a role in human retinal photoreceptor differentiation and degeneration. Proceedings of the National Academy of Sciences of the United States of America, 2002, 99, 473-478.	7.1	218
41	Identification of a Bardet-Biedl syndrome locus on chromosome 3 and evaluation of an efficient approach to homozygosity mapping. Human Molecular Genetics, 1994, 3, 1331-1335.	2.9	216
42	Integrated human genomeâ€“wide maps constructed using the CEPH reference panel. Nature Genetics, 1994, 6, 391-393.	21.4	216
43	Identification of mutations in the connexin 26 gene that cause autosomal recessive nonsyndromic hearing loss. Human Mutation, 1998, 11, 387-394.	2.5	216
44	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
45	Crumbs homolog 1 (CRB1) mutations result in a thick human retina with abnormal lamination. Human Molecular Genetics, 2003, 12, 1073-1078.	2.9	205
46	An ARL3â€“UNC119â€“RP2 GTPase cycle targets myristoylated NPHP3 to the primary cilium. Genes and Development, 2011, 25, 2347-2360.	5.9	202
47	Myocilin Glaucoma. Survey of Ophthalmology, 2002, 47, 547-561.	4.0	201
48	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
49	Night blindness in Sorsby's fundus dystrophy reversed by vitamin A. Nature Genetics, 1995, 11, 27-32.	21.4	197
50	Allelic variation in ABCR associated with Stargardt disease but not age-related macular degeneration. Nature Genetics, 1998, 20, 328-329.	21.4	194
51	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
52	Incorporating language phenotypes strengthens evidence of linkage to autism. American Journal of Medical Genetics Part A, 2001, 105, 539-547.	2.4	192
53	Examination of AVPR1a as an autism susceptibility gene. Molecular Psychiatry, 2004, 9, 968-972.	7.9	190
54	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

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55	Copy number variations on chromosome 12q14 in patients with normal tension glaucoma. Human Molecular Genetics, 2011, 20, 2482-2494.	2.9	189
56	Evidence supporting WNT2 as an autism susceptibility gene. American Journal of Medical Genetics Part A, 2001, 105, 406-413.	2.4	188
57	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
58	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
59	A Novel Protein LZTFL1 Regulates Ciliary Trafficking of the BBSome and Smoothened. PLoS Genetics, 2011, 7, e1002358.	3.5	182
60	Mkks-null mice have a phenotype resembling Bardetâ€“Biedl syndrome. Human Molecular Genetics, 2005, 14, 1109-1118.	2.9	181
61	A collection of tri- and tetranucleotide repeat markers used to generate high quality, high resolution human genome-wide linkage maps. Human Molecular Genetics, 1995, 4, 1837-1844.	2.9	180
62	Linkage of Bardetâ€“Biedl syndrome to chromosome 16q and evidence for nonâ€“allelic genetic heterogeneity. Nature Genetics, 1993, 5, 392-396.	21.4	176
63	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
64	Use of a DNA pooling strategy to identify a human obesity syndrome locus on chromosome 15. Human Molecular Genetics, 1995, 4, 9-13.	2.9	171
65	Evaluation of optineurin sequence variations in 1,048 patients with open-angle glaucoma. American Journal of Ophthalmology, 2003, 136, 904-910.	3.3	164
66	BBS mutations modify phenotypic expression of CEP290-related ciliopathies. Human Molecular Genetics, 2014, 23, 40-51.	2.9	164
67	Three autosomal dominant corneal dystrophies map to chromosome 5q. Nature Genetics, 1994, 6, 47-51.	21.4	159
68	Pendred syndrome maps to chromosome 7q21-34 and is caused by an intrinsic defect in thyroid iodine organification. Nature Genetics, 1996, 12, 424-426.	21.4	159
69	Non-exomic and synonymous variants in ABCA4 are an important cause of Stargardt disease. Human Molecular Genetics, 2013, 22, 5136-5145.	2.9	159
70	Recommendations for Genetic Testing of Inherited Eye Diseases. Ophthalmology, 2012, 119, 2408-2410.	5.2	157
71	Linkage Disequilibrium at the Angelman Syndrome Gene UBE3A in Autism Families. Genomics, 2001, 77, 105-113.	2.9	154
72	Increased expression of the WNT antagonist sFRP-1 in glaucoma elevates intraocular pressure. Journal of Clinical Investigation, 2008, 118, 1056-64.	8.2	143

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73	CHD7 Gene Polymorphisms Are Associated with Susceptibility to Idiopathic Scoliosis. American Journal of Human Genetics, 2007, 80, 957-965.	6.2	142
74	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
75	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
76	Functional differences of the PDS gene product are associated with phenotypic variation in patients with Pendred syndrome and non-syndromic hearing loss (DFNB4). Human Molecular Genetics, 2000, 9, 1709-1715.	2.9	139
77	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
78	Linkage of posterior polymorphous corneal dystrophy to 20q11. Human Molecular Genetics, 1995, 4, 485-488.	2.9	135
79	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
80	Mutations in a novel gene encoding a CRAL-TRIO domain cause human Cayman ataxia and ataxia/dystonia in the jittery mouse. Nature Genetics, 2003, 35, 264-269.	21.4	134
81	Ocular-specific ER stress reduction rescues glaucoma in murine glucocorticoid-induced glaucoma. Journal of Clinical Investigation, 2014, 124, 1956-1965.	8.2	133
82	A family with Axenfeldâ€“Rieger syndrome and Peters Anomaly caused by a point mutation (Phe112Ser) in the FOXC1 gene. American Journal of Ophthalmology, 2003, 135, 368-375.	3.3	128
83	BBS proteins interact genetically with the IFT pathway to influence SHH-related phenotypes. Human Molecular Genetics, 2012, 21, 1945-1953.	2.9	123
84	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
85	Use of isolated inbred human populations for identification of disease genes. Trends in Genetics, 1998, 14, 391-396.	6.7	121
86	cDNA expressed sequence tags of Trypanosoma brucei rhodesiense provide new insights into the biology of the parasite. Molecular and Biochemical Parasitology, 1995, 73, 75-90.	1.1	118
87	Autosomal dominant Charcot-Marie-Tooth axonal neuropathy mapped on chromosome 7p (CMT2D). Human Molecular Genetics, 1996, 5, 1373-1375.	2.9	118
88	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
89	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
90	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

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91	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
92	Complement Factor H Gene Mutation Associated with Autosomal Recessive Atypical Hemolytic Uremic Syndrome. American Journal of Human Genetics, 1999, 65, 1538-1546.	6.2	110
93	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
94	Mutation in TDRD9 causes non-obstructive azoospermia in infertile men. Journal of Medical Genetics, 2017, 54, 633-639.	3.2	107
95	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
96	Evidence for a Novel X-Linked Modifier Locus for Leber Hereditary Optic Neuropathy. Ophthalmic Genetics, 2008, 29, 17-24.	1.2	105
97	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
98	Characterization and Comparison of the Human and MouseGLC1A Glaucomaâ€‰Genes. Genome Research, 1998, 8, 377-384.	5.5	103
99	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
100	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
101	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
102	Topical Ocular Sodium 4-Phenylbutyrate Rescues Glaucoma in a Myocilin Mouse Model of Primary Open-Angle Glaucoma. , 2012, 53, 1557.		100
103	Connexin mutations and hearing loss. Nature, 1998, 391, 32-32.	27.8	98
104	Mice defective in Trpm6 show embryonic mortality and neural tube defects. Human Molecular Genetics, 2009, 18, 4367-4375.	2.9	97
105	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
106	Phenotypic differences among patients with Bardetâ€Biedl syndrome linked to three different chromosome loci. American Journal of Medical Genetics Part A, 1995, 59, 199-203.	2.4	96
107	Variations in the Myocilin Gene in Patients With Open-Angle Glaucoma. JAMA Ophthalmology, 2002, 120, 1189.	2.4	96
108	Nuclear receptor NR2E3 gene mutations distort human retinal laminar architecture and cause an unusual degeneration. Human Molecular Genetics, 2004, 13, 1893-1902.	2.9	94

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109	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
110	Mutations inC8ORF37cause Bardet Biedl syndrome (BBS21). Human Molecular Genetics, 2016, 25, 2283-2294.	2.9	91
111	Establishing a connection between cilia and Bardetâ€“Biedl Syndrome. Trends in Molecular Medicine, 2004, 10, 106-109.	6.7	89
112	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
113	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
114	A Bedouin Kindred with Infantile Nephronophthisis Demonstrates Linkage to Chromosome 9 by Homozygosity Mapping. American Journal of Human Genetics, 1998, 63, 1404-1410.	6.2	83
115	Retinal Disease Expression in Bardet-Biedl Syndrome-1 (BBS1) Is a Spectrum from Maculopathy to Retina-Wide Degeneration. , 2006, 47, 5004.		83
116	Genome-wide identification of pseudogenes capable of disease-causing gene conversion. Human Mutation, 2006, 27, 545-552.	2.5	82
117	Genomics and the Eye. New England Journal of Medicine, 2011, 364, 1932-1942.	27.0	81
118	Sets of short tandem repeat polymorphisms for efficient linkage screening of the human genome. Human Molecular Genetics, 1995, 4, 449-452.	2.9	80
119	Subretinal Gene Therapy of Mice With Bardet-Biedl Syndrome Type 1. , 2013, 54, 6118.		79
120	Survey of trinucleotide repeats in the human genome: assessment of their utility as genetic markers. Human Molecular Genetics, 1995, 4, 1829-1836.	2.9	78
121	Performance of cochlear implant recipients with GJB2â€related deafness. American Journal of Medical Genetics Part A, 2002, 109, 167-170.	2.4	78
122	Calpain-5 Mutations Cause Autoimmune Uveitis, Retinal Neovascularization, and Photoreceptor Degeneration. PLoS Genetics, 2012, 8, e1003001.	3.5	76
123	Restoration of Aqueous Humor Outflow Following Transplantation of iPSC-Derived Trabecular Meshwork Cells in a Transgenic Mouse Model of Glaucoma. , 2017, 58, 2054.		76
124	Identification and Functional Analysis of the Vision-Specific BBS3 (ARL6) Long Isoform. PLoS Genetics, 2010, 6, e1000884.	3.5	75
125	Genetic interaction between Bardet-Biedl syndrome genes and implications for limb patterning. Human Molecular Genetics, 2008, 17, 1956-1967.	2.9	74
126	Pitfalls in Homozygosity Mapping. American Journal of Human Genetics, 2000, 67, 1348-1351.	6.2	72

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127	Bardet-Biedl syndrome in Denmark-report of 13 novel sequence variations in six genes. Human Mutation, 2010, 31, 429-436.	2.5	72
128	Evaluation ofFOXP2 as an autism susceptibility gene. American Journal of Medical Genetics Part A, 2002, 114, 566-569.	2.4	71
129	Homozygosity and Linkage-Disequilibrium Mapping of the Syndrome of Congenital Hypoparathyroidism, Growth and Mental Retardation, and Dysmorphism to a 1-cM Interval on Chromosome 1q42-43. American Journal of Human Genetics, 1998, 63, 163-169.	6.2	70
130	Discovery and Functional Analysis of a Retinitis Pigmentosa Gene, C2ORF71. American Journal of Human Genetics, 2010, 86, 686-695.	6.2	70
131	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
132	Ocular phenotypes of three genetic variants of Bardet-Biedl syndrome. , 2005, 132A, 283-287.		66
133	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
134	Linkage of Infantile Bartter Syndrome with Sensorineural Deafness to Chromosome 1p. American Journal of Human Genetics, 1998, 62, 355-361.	6.2	64
135	Two Different Mutations in the Thyroid Peroxidase Gene of a Large Inbred Amish Kindred: Power and Limits of Homozygosity Mapping1. Journal of Clinical Endocrinology and Metabolism, 1999, 84, 1061-1071.	3.6	64
136	Variations in NPHP5 in Patients With Nonsyndromic Leber Congenital Amaurosis and Senior-Loken Syndrome. JAMA Ophthalmology, 2011, 129, 81.	2.4	62
137	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
138	BBSome function is required for both the morphogenesis and maintenance of the photoreceptor outer segment. PLoS Genetics, 2017, 13, e1007057.	3.5	60
139	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
140	No Association Between Variations in the WDR36 Gene and Primary Open-Angle Glaucoma. JAMA Ophthalmology, 2007, 125, 434.	2.4	58
141	Gene Expression Analysis of Photoreceptor Cell Loss inBbs4-Knockout Mice Reveals an Early Stress Gene Response and Photoreceptor Cell Damage. , 2007, 48, 3329.		57
142	Regulation of Insulin Receptor Trafficking by Bardet Biedl Syndrome Proteins. PLoS Genetics, 2015, 11, e1005311.	3.5	57
143	A high-density microsatellite map of the ataxia-telangiectasia locus. Human Genetics, 1995, 95, 451-454.	3.8	56
144	Novel approaches to linkage mapping. Current Opinion in Genetics and Development, 1995, 5, 335-341.	3.3	56

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145	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
146	Full characterization of the maculopathy associated with an Arg-172-Trp mutation in the RDS/peripherin gene. Ophthalmic Genetics, 1996, 17, 175-186.	1.2	55
147	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
148	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
149	Exposure to Static Magnetic and Electric Fields Treats Type 2 Diabetes. Cell Metabolism, 2020, 32, 561-574.e7.	16.2	55
150	Expression of theMf1 gene in developing mouse hearts: Implication in the development of human congenital heart defects. , 1999, 216, 16-27.		54
151	Allelic Variants of Human Melatonin 1A Receptor in Patients with Familial Adolescent Idiopathic Scoliosis. Spine, 2003, 28, 2025-2028.	2.0	54
152	Deducing the pathogenic contribution of recessive ABCA4 alleles in an outbred population. Human Molecular Genetics, 2010, 19, 3693-3701.	2.9	53
153	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
154	Gene therapy and gene correction: targets, progress, and challenges for treating human diseases. Gene Therapy, 2022, 29, 3-12.	4.5	53
155	A Constrained-Likelihood Approach to Marker-Trait Association Studies. American Journal of Human Genetics, 2005, 77, 768-780.	6.2	51
156	New gene for autosomal recessive non-syndromic hearing loss maps to either chromosome 3q or 19p. American Journal of Medical Genetics Part A, 1997, 71, 467-471.	2.4	50
157	Clinicopathologic effects of mutant GUCY2D in Leber congenital amaurosis. Ophthalmology, 2003, 110, 549-558.	5.2	50
158	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
159	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
160	Complement Factor H Polymorphism p.Tyr402His and Cuticular Drusen. JAMA Ophthalmology, 2007, 125, 93.	2.4	48
161	Congenital myopathy is caused by mutation of HACD1. Human Molecular Genetics, 2013, 22, 5229-5236.	2.9	48
162	Novel Mutation in the TIMP3 Gene Causes Sorsby Fundus Dystrophy. JAMA Ophthalmology, 2002, 120, 376.	2.4	47

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163	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
164	ATF4 leads to glaucoma by promoting protein synthesis and ER client protein load. Nature Communications, 2020, 11, 5594.	12.8	47
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