Michael E Cheetham

List of Publications by Year
in descending order

156 papers	18,210 citations	28274 55 h-index	14208 128 g-index
164 all docs	164 docs citations	164 times ranked	26094 citing authors

#	Article	IF	Citations
1	Guidelines for the use and interpretation of assays for monitoring autophagy (3rd edition). Autophagy, 2016, 12, 1-222.	9.1	4,701
2	Guidelines for the use and interpretation of assays for monitoring autophagy (4th) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 702 Td (edition	9.1	1,430
3	Guidelines for the nomenclature of the human heat shock proteins. Cell Stress and Chaperones, 2009, 14, 105-111.	2.9	1,105
4	Structure, function and evolution of DnaJ: conservation and adaptation of chaperone function. Cell Stress and Chaperones, 1998, 3, 28.	2.9	528
5	Mutations in MRAP, encoding a new interacting partner of the ACTH receptor, cause familial glucocorticoid deficiency type 2. Nature Genetics, 2005, 37, 166-170.	21.4	388
6	Mechanisms of cell death in rhodopsin retinitis pigmentosa: implications for therapy. Trends in Molecular Medicine, 2005, 11, 177-185.	6.7	339
7	Targeting amyloid-Î² in glaucoma treatment. Proceedings of the National Academy of Sciences of the United States of America, 2007, 104, 13444-13449.	7.1	315
8	In vitro studies show that Hsp70 can be released by glia and that exogenous Hsp70 can enhance neuronal stress tolerance. Brain Research, 2001, 914, 66-73.	2.2	306
9	The cellular fate of mutant rhodopsin: quality control, degradation and aggresome formation. Journal of Cell Science, 2002, 115, 2907-2918.	2.0	295
10	Identification and Correction of Mechanisms Underlying Inherited Blindness in Human iPSC-Derived Optic Cups. Cell Stem Cell, 2016, 18, 769-781.	11.1	279
11	Not all J domains are created equal: Implications for the specificity of Hsp40-Hsp70 interactions. Protein Science, 2005, 14, 1697-1709.	7.6	265
12	The cellular fate of mutant rhodopsin: quality control, degradation and aggresome formation. Journal of Cell Science, 2002, 115, 2907-18.	2.0	262
13	The molecular and cellular basis of rhodopsin retinitis pigmentosa reveals potential strategies for therapy. Progress in Retinal and Eye Research, 2018, 62, 1-23.	15.5	254
14	Oxidative Stress Affects the Junctional Integrity of Retinal Pigment Epithelial Cells. , 2004, 45, 675.		229
15	Cystamine and cysteamine increase brain levels of BDNF in Huntington disease via HSJ1b and transglutaminase. Journal of Clinical Investigation, 2006, 116, 1410-1424.	8.2	211
16	MRAP and MRAP2 are bidirectional regulators of the melanocortin receptor family. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 6146-6151.	7.1	201
17	Mutations in LCA5, encoding the ciliary protein lebercilin, cause Leber congenital amaurosis. Nature Genetics, 2007, 39, 889-895.	21.4	186
18	Effect of an intravitreal antisense oligonucleotide on vision in Leber congenital amaurosis due to a photoreceptor cilium defect. Nature Medicine, 2019, 25, 225-228.	30.7	177

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19	EYS, encoding an ortholog of Drosophila spacemaker, is mutated in autosomal recessive retinitis pigmentosa. Nature Genetics, 2008, 40, 1285-1287.	21.4	175
20	The ataxia protein sacsin is a functional co-chaperone that protects against polyglutamine-expanded ataxin-1. Human Molecular Genetics, 2009, 18, 1556-1565.	2.9	153
21	Late stage treatment with arimoclomol delays disease progression and prevents protein aggregation in the SOD1^{G93A} mouse model of ALS. Journal of Neurochemistry, 2008, 107, 339-350.	3.9	147
22	Deep intronic mutation in OFD1, identified by targeted genomic next-generation sequencing, causes a severe form of X-linked retinitis pigmentosa (RP23). Human Molecular Genetics, 2012, 21, 3647-3654.	2.9	133
23	The heat shock response plays an important role in TDP-43 clearance: evidence for dysfunction in amyotrophic lateral sclerosis. Brain, 2016, 139, 1417-1432.	7.6	131
24	HSJ1 Is a Neuronal Shuttling Factor for the Sorting of Chaperone Clients to the Proteasome. Current Biology, 2005, 15, 1058-1064.	3.9	130
25	Splice-Modulating Oligonucleotide QR-110 Restores CEP290 mRNA and Function in Human c.2991+1655A>G LCA10 Models. Molecular Therapy - Nucleic Acids, 2018, 12, 730-740.	5.1	130
26	The cell stress machinery and retinal degeneration. FEBS Letters, 2013, 587, 2008-2017.	2.8	126
27	Pharmacological manipulation of gain-of-function and dominant-negative mechanisms in rhodopsin retinitis pigmentosa. Human Molecular Genetics, 2008, 17, 3043-3054.	2.9	125
28	The retinitis pigmentosa protein RP2 links pericentriolar vesicle transport between the Golgi and the primary cilium. Human Molecular Genetics, 2010, 19, 1358-1367.	2.9	124
29	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	6.2	121
30	Localization in the human retina of the X-linked retinitis pigmentosa protein RP2, its homologue cofactor C and the RP2 interacting protein Arl3. Human Molecular Genetics, 2002, 11, 3065-3074.	2.9	119
31	DNAJ Proteins in neurodegeneration: essential and protective factors. Philosophical Transactions of the Royal Society B: Biological Sciences, 2018, 373, 20160534.	4.0	115
32	Modeling and Rescue of RP2 Retinitis Pigmentosa Using iPSC-Derived Retinal Organoids. Stem Cell Reports, 2020, 15, 67-79.	4.8	109
33	Identification of NovelRPGRORF15 Mutations in X-linked Progressive Cone-Rod Dystrophy (XLCORD) Families. , 2005, 46, 1891.		104
34	Suppression of protein aggregation by chaperone modification of high molecular weight complexes. Brain, 2012, 135, 1180-1196.	7.6	103
35	Nuclear translocation of the Hsp70/Hsp90 organizing protein mSTI1 is regulated by cell cycle kinases. Journal of Cell Science, 2004, 117, 701-710.	2.0	100
36	Unfolding retinal dystrophies: a role for molecular chaperones?. Trends in Molecular Medicine, 2001, 7, 414-421.	6.7	99

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37	The Chaperone Environment at the Cytoplasmic Face of the Endoplasmic Reticulum Can Modulate Rhodopsin Processing and Inclusion Formation. Journal of Biological Chemistry, 2003, 278, 19087-19094.	3.4	99
38	Translational read-through of the RP2 Arg120stop mutation in patient iPSC-derived retinal pigment epithelium cells. Human Molecular Genetics, 2015, 24, 972-986.	2.9	97
39	Hsp40 Molecules That Target to the Ubiquitin-proteasome System Decrease Inclusion Formation in Models of Polyglutamine Disease. Molecular Therapy, 2007, 15, 1100-1105.	8.2	96
40	Differential Expression of Two Distinct Functional Isoforms of Melanopsin (Opn4) in the Mammalian Retina. Journal of Neuroscience, 2009, 29, 12332-12342.	3.6	87
41	The role of the ER stress-response protein PERK in rhodopsin retinitis pigmentosa. Human Molecular Genetics, 2017, 26, 4896-4905.	2.9	82
42	Molecular chaperones and neuronal proteostasis. Seminars in Cell and Developmental Biology, 2015, 40, 142-152.	5.0	81
43	The Role of HSP70 and Its Co-chaperones in Protein Misfolding, Aggregation and Disease. Sub-Cellular Biochemistry, 2015, 78, 243-273.	2.4	76
44	Inherited ACTH insensitivity illuminates the mechanisms of ACTH action. Trends in Endocrinology and Metabolism, 2005, 16, 451-457.	7.1	75
45	Structural Variants Create New Topological-Associated Domains and Ectopic Retinal Enhancer-Gene Contact in Dominant Retinitis Pigmentosa. American Journal of Human Genetics, 2020, 107, 802-814.	6.2	75
46	Analysis of the levels of conservation of the J domain among the various types of DnaJ-like proteins. Cell Stress and Chaperones, 2000, 5, 347.	2.9	74
47	Treatment with extracellular HSP70/HSC70 protein can reduce polyglutamine toxicity and aggregation. Journal of Neurochemistry, 2005, 94, 597-606.	3.9	74
48	Downstream caspases are novel targets for the antiapoptotic activity of the molecular chaperone Hsp70. Cell Stress and Chaperones, 2004, 9, 265.	2.9	70
49	Hsp90 inhibition protects against inherited retinal degeneration. Human Molecular Genetics, 2014, 23, 2164-2175.	2.9	70
50	Autosomal-Dominant Corneal Endothelial Dystrophies CHED1 and PPCD1 Are Allelic Disorders Caused by Non-coding Mutations in the Promoter of OVOL2. American Journal of Human Genetics, 2016, 98, 75-89.	6.2	70
51	Regulation of 70-kDa Heat-Shock-Protein ATPase Activity and Substrate Binding by Human DnaJ-Like Proteins, HSJ1a and HSJ1b. FEBS Journal, 1994, 226, 99-107.	0.2	70
52	The Leber congenital amaurosis gene product AIPL1 is localized exclusively in rod photoreceptors of the adult human retina. Human Molecular Genetics, 2002, 11, 823-831.	2.9	67
53	DnaJB6 is present in the core of Lewy bodies and is highly upâ€regulated in parkinsonian astrocytes. Journal of Neuroscience Research, 2009, 87, 238-245.	2.9	67
54	Mutations in ARL2BP, Encoding ADP-Ribosylation-Factor-Like 2 Binding Protein, Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2013, 93, 321-329.	6.2	67

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55	The Melanocortin 2 Receptor Accessory Protein Exists as a Homodimer and Is Essential for the Function of the Melanocortin 2 Receptor in the Mouse Y1 Cell Line. Endocrinology, 2008, 149, 1935-1941.	2.8	65
56	The Nanceâ€“Horan syndrome protein encodes a functional WAVE homology domain (WHD) and is important for co-ordinating actin remodelling and maintaining cell morphology. Human Molecular Genetics, 2010, 19, 2421-2432.	2.9	64
57	The Leber Congenital Amaurosis Protein AIPL1 Functions as Part of a Chaperone Heterocomplex. , 2008, 49, 2878.		63
58	Distinct Melanocortin 2 Receptor Accessory Protein Domains Are Required for Melanocortin 2 Receptor Interaction and Promotion of Receptor Trafficking. Endocrinology, 2009, 150, 720-726.	2.8	63
59	Molecular chaperone-mediated rescue of mitophagy by a Parkin RING1 domain mutant. Human Molecular Genetics, 2011, 20, 16-27.	2.9	63
60	Mutations in the Spliceosome Component CWC27 Cause Retinal Degeneration with or without Additional Developmental Anomalies. American Journal of Human Genetics, 2017, 100, 592-604.	6.2	61
61	Arl3 and RP2 regulate the trafficking of ciliary tip kinesins. Human Molecular Genetics, 2017, 26, 2480-2492.	2.9	60
62	X-Linked Megalocornea Caused by Mutations in CHRDL1 Identifies an Essential Role for Ventroptin in Anterior Segment Development. American Journal of Human Genetics, 2012, 90, 247-259.	6.2	59
63	Antisense Therapy for a Common Corneal Dystrophy Ameliorates TCF4 Repeat Expansion-Mediated Toxicity. American Journal of Human Genetics, 2018, 102, 528-539.	6.2	59
64	PCARE and WASF3 regulate ciliary F-actin assembly that is required for the initiation of photoreceptor outer segment disk formation. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 9922-9931.	7.1	58
65	An Assessment of the Apex Microarray Technology in Genotyping Patients with Leber Congenital Amaurosis and Early-Onset Severe Retinal Dystrophy. , 2007, 48, 5684.		56
66	Rescue of the MERTK phagocytic defect in a human iPSC disease model using translational read-through inducing drugs. Scientific Reports, 2017, 7, 51.	3.3	55
67	Detailed Phenotyping and Therapeutic Strategies for Intronic ABCA4 Variants in Stargardt Disease. Molecular Therapy - Nucleic Acids, 2020, 21, 412-427.	5.1	55
68	The heat-shock response co-inducer arimoclomol protects against retinal degeneration in rhodopsin retinitis pigmentosa. Cell Death and Disease, 2014, 5, e1236-e1236.	6.3	54
69	Three Different Cone Opsin Gene Array Mutational Mechanisms; Genotype-Phenotype Correlation and Functional Investigation of Cone Opsin Variants. Human Mutation, 2014, 35, n/a-n/a.	2.5	53
70	Molecular chaperones and photoreceptor function. Progress in Retinal and Eye Research, 2008, 27, 434-449.	15.5	52
71	A dual role for EDEM1 in the processing of rod opsin. Journal of Cell Science, 2009, 122, 4465-4472.	2.0	52
72	Apolipoprotein E-∈A allele and Alzheimer's disease. Lancet, The, 1993, 342, 1308-1309.	13.7	51

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73	Arl3 and RP2 mediated assembly and traffic of membrane associated cilia proteins. Vision Research, 2012, 75, 2-4.	1.4	51
74	Molecular Chaperone Mediated Late-Stage Neuroprotection in the SOD1G93A Mouse Model of Amyotrophic Lateral Sclerosis. PLoS ONE, 2013, 8, e73944.	2.5	51
75	TOPORS, implicated in retinal degeneration, is a cilia-centrosomal protein. Human Molecular Genetics, 2011, 20, 975-987.	2.9	49
76	Mutations in theCACNA1F andNYX genes in British CSNBX families. Human Mutation, 2003, 21, 169-169.	2.5	48
77	Intravitreal antisense oligonucleotide sepofarsen in Leber congenital amaurosis type 10: a phase 1b/2 trial. Nature Medicine, 2022, 28, 1014-1021.	30.7	46
78	The Expression of the Leber Congenital Amaurosis Protein AIPL1 Coincides with Rod and Cone Photoreceptor Development. , 2003, 44, 5396.		45
79	Isolation and characterization of murine Cds (CDP-diacylglycerol synthase) 1 and 2. Gene, 2005, 356, 19-31.	2.2	45
80	X-Linked Cone Dystrophy Caused by Mutation of the Red and Green Cone Opsins. American Journal of Human Genetics, 2010, 87, 26-39.	6.2	45
81	Ectopic GRHL2 Expression Due to Non-coding Mutations Promotes Cell State Transition and Causes Posterior Polymorphous Corneal Dystrophy 4. American Journal of Human Genetics, 2018, 102, 447-459.	6.2	45
82	BiP prevents rod opsin aggregation. Molecular Biology of the Cell, 2012, 23, 3522-3531.	2.1	44
83	Domain Requirements of DnaJ-like (Hsp40) Molecular Chaperones in the Activation of a Steroid Hormone Receptor. Journal of Biological Chemistry, 1999, 274, 34045-34052.	3.4	43
84	Lack of Interphotoreceptor Retinoid Binding Protein Caused by Homozygous Mutation ofRBP3Is Associated With High Myopia and Retinal Dystrophy. , 2015, 56, 2358.		42
85	Biallelic Mutations in the Autophagy Regulator DRAM2 Cause Retinal Dystrophy with Early Macular Involvement. American Journal of Human Genetics, 2015, 96, 948-954.	6.2	42
86	Delineation of the plasma membrane targeting domain of the X-linked retinitis pigmentosa protein RP2. Investigative Ophthalmology and Visual Science, 2002, 43, 2015-20.	3.3	41
87	Rescue of mutant rhodopsin traffic by metformin-induced AMPK activation accelerates photoreceptor degeneration. Human Molecular Genetics, 2017, 26, ddw387.	2.9	39
88	REEP6 deficiency leads to retinal degeneration through disruption of ER homeostasis and protein trafficking. Human Molecular Genetics, 2017, 26, 2667-2677.	2.9	39
89	DYNC2H1 hypomorphic or retina-predominant variants cause nonsyndromic retinal degeneration. Genetics in Medicine, 2020, 22, 2041-2051.	2.4	38
90	Molecular Mechanisms of Disease for Mutations at Gly-90 in Rhodopsin. Journal of Biological Chemistry, 2011, 286, 39993-40001.	3.4	37

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91	Neuronal DnaJ proteins HSJ1a and HSJ1b: a role in linking the Hsp70 chaperone machine to the ubiquitinâ€“proteasome system?. Biochemical Society Transactions, 2004, 32, 640-642.	3.4	36
92	A look into retinal organoids: methods, analytical techniques, and applications. Cellular and Molecular Life Sciences, 2021, 78, 6505-6532.	5.4	36
93	Identification of novel mutations in the carbohydrate sulfotransferase gene (CHST6) causing macular corneal dystrophy. Investigative Ophthalmology and Visual Science, 2002, 43, 377-82.	3.3	36
94	The X-linked retinitis pigmentosa protein RP2 facilitates G protein traffic. Human Molecular Genetics, 2012, 21, 863-873.	2.9	34
95	Inhibition of hsc70-catalysed clathrin uncoating by HSJ1 proteins. Biochemical Journal, 1996, 319, 103-108.	3.7	33
96	Brittle Cornea Syndrome ZNF469 Mutation Carrier Phenotype and Segregation Analysis of Rare ZNF469 Variants in Familial Keratoconus. Investigative Ophthalmology and Visual Science, 2015, 56, 578-586.	3.3	33
97	Cytosolic and ER J-domains of mammalian and parasitic origin can functionally interact with DnaK. International Journal of Biochemistry and Cell Biology, 2007, 39, 736-751.	2.8	32
98	Knockdown of the Zebrafish Ortholog of the Retinitis Pigmentosa 2 (RP2) Gene Results in Retinal Degeneration. , 2011, 52, 2960.		32
99	Differential Light-induced Responses in Sectorial Inherited Retinal Degeneration. Journal of Biological Chemistry, 2014, 289, 35918-35928.	3.4	32
100	Allele-specific editing ameliorates dominant retinitis pigmentosa in a transgenic mouse model. American Journal of Human Genetics, 2021, 108, 295-308.	6.2	31
101	The Leber Congenital Amaurosis Protein AIPL1 Modulates the Nuclear Translocation of NUB1 and Suppresses Inclusion Formation by NUB1 Fragments. Journal of Biological Chemistry, 2004, 279, 48038-48047.	3.4	30
102	NUB1 modulation of GSK3Î² reduces tau aggregation. Human Molecular Genetics, 2012, 21, 5254-5267.	2.9	29
103	Novel CHST6 nonsense and missense mutations responsible for macular corneal dystrophy. American Journal of Ophthalmology, 2005, 139, 192-193.	3.3	28
104	Leber Congenital Amaurosis Associated with AIPL1: Challenges in Ascribing Disease Causation, Clinical Findings, and Implications for Gene Therapy. PLoS ONE, 2012, 7, e32330.	2.5	28
105	Novel Retinal and Cone Photoreceptor Transcripts Revealed by Human Macular Expression Profiling. , 2007, 48, 5388.		26
106	Biallelic Mutation of ARHGEF18, Involved in the Determination of Epithelial Apicobasal Polarity, Causes Adult-Onset Retinal Degeneration. American Journal of Human Genetics, 2017, 100, 334-342.	6.2	26
107	Symmetric arrangement of mitochondria:plasma membrane contacts between adjacent photoreceptor cells regulated by Opa1. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 15684-15693.	7.1	26
108	Recessive Retinopathy Consequent on Mutant G-Protein Î² Subunit 3 (GNB3). JAMA Ophthalmology, 2016, 134, 924.	2.5	25

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109	Loss-of-Function Mutations in the CFH Gene Affecting Alternatively Encoded Factor H-like 1 Protein Cause Dominant Early-Onset Macular Drusen. Ophthalmology, 2019, 126, 1410-1421.	5.2	25
110	Identification and characterization of a human mitochondrial homologue of the bacterial co-chaperone GrpE. Gene, 2001, 267, 125-134.	2.2	24
111	Regulation of 70â€kDa Heatâ€Shockâ€Protein ATPase Activity and Substrate Binding by Human DnaJâ€Like Proteins, HSJ1a and HSJ1b. FEBS Journal, 1994, 226, 99-107.	0.2	24
112	Organization on the plasma membrane of the retinitis pigmentosa protein RP2: investigation of association with detergent-resistant membranes and polarized sorting. Biochemical Journal, 2003, 372, 427-433.	3.7	23
113	The retinitis pigmentosa-mutated RP2 protein exhibits exonuclease activity and translocates to the nucleus in response to DNA damage. Experimental Cell Research, 2006, 312, 1323-1334.	2.6	23
114	The co-chaperone and reductase ERdj5 facilitates rod opsin biogenesis and quality control. Human Molecular Genetics, 2014, 23, 6594-6606.	2.9	23
115	Missense variants in the X-linked genePRPS1cause retinal degeneration in females. Human Mutation, 2018, 39, 80-91.	2.5	23
116	Pharmacological Manipulation of Rhodopsin Retinitis Pigmentosa. Advances in Experimental Medicine and Biology, 2010, 664, 317-323.	1.6	23
117	Modulation of Sub-RPE Deposits In Vitro: A Potential Model for Age-Related Macular Degeneration. Investigative Ophthalmology and Visual Science, 2004, 45, 1281-1288.	3.3	22
118	CRISPR-Cas9 correction of OPA1 c.1334G>A: p.R445H restores mitochondrial homeostasis in dominant optic atrophy patient-derived iPSCs. Molecular Therapy - Nucleic Acids, 2021, 26, 432-443.	5.1	21
119	Calnexin is not essential for mammalian rod opsin biogenesis. Molecular Vision, 2008, 14, 2466-74.	1.1	21
120	Focus on Molecules: Xâ€linked Retinitis Pigmentosa 2 protein, RP2. Experimental Eye Research, 2006, 82, 543-544.	2.6	20
121	Hsp90 as a Potential Therapeutic Target in Retinal Disease. Advances in Experimental Medicine and Biology, 2016, 854, 161-167.	1.6	19
122	Using induced pluripotent stem cells to understand retinal ciliopathy disease mechanisms and develop therapies. Biochemical Society Transactions, 2016, 44, 1245-1251.	3.4	18
123	The Inherited Blindness Protein AIPL1 Regulates the Ubiquitin-Like FAT10 Pathway. PLoS ONE, 2012, 7, e30866.	2.5	17
124	Interaction of the human DnaJ homologue, HSJ1b with the 90 kDa heat shock protein, Hsp90. Life Sciences, 2000, 67, 1455-1465.	4.3	16
125	Focus on Molecules: Centrosomal protein 290 (CEP290). Experimental Eye Research, 2011, 92, 316-317.	2.6	15
126	The Cochaperone Murine Stress-Inducible Protein 1: Overexpression, Purification, and Characterization. Protein Expression and Purification, 2001, 21, 462-469.	1.3	14

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127	A recurrent splice-site mutation in EPHA2 causing congenital posterior nuclear cataract. Ophthalmic Genetics, 2018, 39, 236-241.	1.2	13
128	Negative Regulator of Ubiquitin-Like Protein 1 modulates the autophagyâ€“lysosomal pathway via p62 to facilitate the extracellular release of tau following proteasome impairment. Human Molecular Genetics, 2020, 29, 80-96.	2.9	12
129	Correlative light and immuno-electron microscopy of retinal tissue cryostat sections. PLoS ONE, 2018, 13, e0191048.	2.5	12
130	Induced Pluripotent Stem Cells for Inherited Optic Neuropathiesâ€”Disease Modeling and Therapeutic Development. Journal of Neuro-Ophthalmology, 2022, 42, 35-44.	0.8	12
131	Predominant rod photoreceptor degeneration in Leber congenital amaurosis. Molecular Vision, 2005, 11, 542-53.	1.1	12
132	Isolation of cDNAs coding for epitopes shared by microtubule-associated proteins and neurofibrillary tangles in Alzheimer's disease. FEBS Letters, 1987, 226, 28-32.	2.8	11
133	Delineating the expanding phenotype associated with SCAPER gene mutation. American Journal of Medical Genetics, Part A, 2019, 179, 1665-1671.	1.2	10
134	AAV-mediated ERdj5 overexpression protects against P23H rhodopsin toxicity. Human Molecular Genetics, 2020, 29, 1310-1318.	2.9	10
135	Focus on Molecules: Nyctalopin. Experimental Eye Research, 2005, 81, 627-628.	2.6	9
136	Pheno4J: a gene to phenotype graph database. Bioinformatics, 2017, 33, 3317-3319.	4.1	9
137	Modelling autosomal dominant optic atrophy associated with OPA1 variants in iPSC-derived retinal ganglion cells. Human Molecular Genetics, 2022, 31, 3478-3493.	2.9	9
138	The binding of the molecular chaperone Hsc70 to the prion protein PrP is modulated by pH and copper. International Journal of Biochemistry and Cell Biology, 2010, 42, 1226-1232.	2.8	8
139	Protein kinase CK2 modulates HSJ1 function through phosphorylation of the UIM2 domain. Human Molecular Genetics, 2017, 26, ddw420.	2.9	8
140	The Leber Congenital Amaurosis Protein AIPL1 and EB Proteins Co-Localize at the Photoreceptor Cilium. PLoS ONE, 2015, 10, e0121440.	2.5	8
141	Targeting the Proteostasis Network in Rhodopsin Retinitis Pigmentosa. Advances in Experimental Medicine and Biology, 2016, 854, 479-484.	1.6	7
142	Expression of alternatively-spliced glutamate receptors in human hippocampus. European Journal of Pharmacology, 1993, 244, 89-92.	2.6	6
143	Assay and Functional Analysis of the ARL3 Effector RP2 Involved in Xâ€Linked Retinitis Pigmentosa. Methods in Enzymology, 2005, 404, 468-480.	1.0	6
144	Whole-genome sequencing reveals a recurrent missense mutation in the Connexin 46 (GJA3) gene causing autosomal-dominant lamellar cataract. Eye, 2018, 32, 1661-1668.	2.1	6

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145	The Role of the X-linked Retinitis Pigmentosa Protein RP2 in Vesicle Traffic and Cilia Function. Advances in Experimental Medicine and Biology, 2012, 723, 527-532.	1.6	6
146	Novel homozygous splicing mutations in cause autosomal recessive retinitis pigmentosa. Molecular Vision, 2018, 24, 603-612.	1.1	6
147	A simple cell based assay to measure Parkin activity. Journal of Neurochemistry, 2011, 116, 342-349.	3.9	5
148	Eye on a Dish Models to Evaluate Splicing Modulation. Methods in Molecular Biology, 2022, 2434, 245-255.	0.9	5
149	The Chaperone Function of the LCA Protein AIPL1. Advances in Experimental Medicine and Biology, 2006, 572, 471-476.	1.6	4
150	A Novel Missense Mutation in Both OPN1LW and OPN1MW Cone Opsin Genes Causes X-Linked Cone Dystrophy (XLCOD5). Advances in Experimental Medicine and Biology, 2012, 723, 595-601.	1.6	2
151	Functional characterization of a novel c.614-622del rhodopsin mutation in a French pedigree with retinitis pigmentosa. Molecular Vision, 2012, 18, 581-7.	1.1	2
152	COMMENTARY: Chaperoning against neuronal vulnerability (Commentary on Zijlstra et al.). European Journal of Neuroscience, 2010, 32, 759-759.	2.6	0
153	Unpicking the UPR. , 2012, 53, 7167.		0
154	The Role of Hsp70 and Its Co-Chaperones in Protein Misfolding, Aggregation and Disease. , 2007, , 122-136.		0
155	Intravitreal Sepofarsen for Leber Congenital Amaurosis Type 10 (LCA10). SSRN Electronic Journal, 0, , .	0.4	0
156	The Role of Chaperones and Co-Chaperones in Retinal Degenerative Diseases. , 2008, , 109-123.		0