J Fielding Hejtmancik

List of Publications by Year
in descending order

117 papers	5,017 citations	126907 33 h-index	114465 63 g-index
119 all docs	119 docs citations	119 times ranked	3978 citing authors

#	Article	IF	Citations
1	Congenital cataracts and their molecular genetics. Seminars in Cell and Developmental Biology, 2008, 19, 134-149.	5.0	324
2	Bietti Crystalline Corneoretinal Dystrophy Is Caused by Mutations in the Novel Gene CYP4V2. American Journal of Human Genetics, 2004, 74, 817-826.	6.2	237
3	Cat-Map: putting cataract on the map. Molecular Vision, 2010, 16, 2007-15.	1.1	226
4	Localization of two genes for usher syndrome type I to chromosome 11. Genomics, 1992, 14, 995-1002.	2.9	216
5	Mutations and mechanisms in congenital and age-related cataracts. Experimental Eye Research, 2017, 156, 95-102.	2.6	165
6	Genetic Origins of Cataract. JAMA Ophthalmology, 2007, 125, 165.	2.4	154
7	Crystallin gene mutations in Indian families with inherited pediatric cataract. Molecular Vision, 2008, 14, 1157-70.	1.1	134
8	Mutations in FYCO1 Cause Autosomal-Recessive Congenital Cataracts. American Journal of Human Genetics, 2011, 88, 827-838.	6.2	132
9	Genetic Heterogeneity in Microcornea-Cataract: Five Novel Mutations inCRYAA,CRYGD, andGJA8. , 2007, 48, 3937.		131
10	The EPHA2 gene is associated with cataracts linked to chromosome 1p. Molecular Vision, 2008, 14, 2042-55.	1.1	129
11	Molecular genetics of age-related cataract. Experimental Eye Research, 2004, 79, 3-9.	2.6	121
12	Mutations in PIP5K3 Are Associated with FranÃ§ois-Neetens MouchetÃ©e Fleck Corneal Dystrophy. American Journal of Human Genetics, 2005, 77, 54-63.	6.2	115
13	A Homozygous Splice Mutation in theHSF4Gene Is Associated with an Autosomal Recessive Congenital Cataract. , 2004, 45, 2716.		100
14	Autosomal recessive juvenile onset cataract associated with mutation in BFSP1. Human Genetics, 2007, 121, 475-482.	3.8	98
15	Mutations in Î²B3-Crystallin Associated with Autosomal Recessive Cataract in Two Pakistani Families. , 2005, 46, 2100.		97
16	Severe retinitis pigmentosa mapped to 4p15 and associated with a novel mutation in the PROM1 gene. Human Genetics, 2007, 122, 293-299.	3.8	97
17	A 5-base insertion in the Î³C-crystallin gene is associated with autosomal dominant variable zonular pulverulent cataract. Human Genetics, 2000, 106, 531-537.	3.8	93
18	Mutations in KCNJ13 Cause Autosomal-Dominant Snowflake Vitreoretinal Degeneration. American Journal of Human Genetics, 2008, 82, 174-180.	6.2	93

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19	Autosomal Dominant Congenital Cataract. Ophthalmology, 1994, 101, 866-871.	5.2	84
20	INVITED EDITORIAL The Genetics of Cataract: Our Vision Becomes Clearer. American Journal of Human Genetics, 1998, 62, 520-525.	6.2	84
21	Autosomal recessive congenital cataract linked to EPHA2 in a consanguineous Pakistani family. Molecular Vision, 2010, 16, 511-7.	1.1	81
22	A Nonsense Mutation in the Glucosaminyl (N-acetyl) Transferase 2 Gene (GCNT2): Association with Autosomal Recessive Congenital Cataracts. , 2004, 45, 1940.		78
23	CDKN2B Polymorphism Is Associated with Primary Open-Angle Glaucoma (POAG) in the Afro-Caribbean Population of Barbados, West Indies. PLoS ONE, 2012, 7, e39278.	2.5	76
24	Biology of Inherited Cataracts and Opportunities for Treatment. Annual Review of Vision Science, 2019, 5, 123-149.	4.4	76
25	Molecular Genetics of Cataract. Progress in Molecular Biology and Translational Science, 2015, 134, 203-218.	1.7	73
26	Lens Biology and Biochemistry. Progress in Molecular Biology and Translational Science, 2015, 134, 169-201.	1.7	71
27	Genetic Linkage of Bietti Crystallin Corneoretinal Dystrophy to Chromosome 4q35. American Journal of Human Genetics, 2000, 67, 1309-1313.	6.2	69
28	A Mutation in SLC24A1 Implicated in Autosomal-Recessive Congenital Stationary Night Blindness. American Journal of Human Genetics, 2010, 87, 523-531.	6.2	67
29	Molecular Genetics of Cataract. , 2003, 37, 67-82.		65
30	Î²A3/A1-crystallin in astroglial cells regulates retinal vascular remodeling during development. Molecular and Cellular Neurosciences, 2008, 37, 85-95.	2.2	64
31	Allelic and locus heterogeneity in autosomal recessive gelatinous drop-like corneal dystrophy. Human Genetics, 2002, 110, 568-577.	3.8	57
32	The G18V CRYGS Mutation Associated with Human Cataracts Increases Î³S-Crystallin Sensitivity to Thermal and Chemical Stress. Biochemistry, 2009, 48, 7334-7341.	2.5	51
33	Autosomal Recessive Retinitis Pigmentosa Is Associated with Mutations inRP1in Three Consanguineous Pakistani Families. , 2005, 46, 2264.		50
34	Overview of the Lens. Progress in Molecular Biology and Translational Science, 2015, 134, 119-127.	1.7	49
35	Molecular Genetic Analysis of Pakistani Families With Autosomal Recessive Congenital Cataracts by Homozygosity Screening. , 2017, 58, 2207.		45
36	A Mutation in ZNF513, a Putative Regulator of Photoreceptor Development, Causes Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 400-409.	6.2	44

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37	Spatial expression patterns of autophagy genes in the eye lens and induction of autophagy in lens cells. Molecular Vision, 2012, 18, 1773-86.	1.1	43
38	Differentiation State-Specific Mitochondrial Dynamic Regulatory Networks Are Revealed by Global Transcriptional Analysis of the Developing Chicken Lens. G3: Genes, Genomes, Genetics, 2014, 4, 1515-1527.	1.8	39
39	Mutations in RLBP1 associated with fundus albipunctatus in consanguineous Pakistani families. British Journal of Ophthalmology, 2011, 95, 1019-1024.	3.9	35
40	FOXE3 contributes to Peters anomaly through transcriptional regulation of an autophagy-associated protein termed DNAJB1. Nature Communications, 2016, 7, 10953.	12.8	35
41	Homozygosity Mapping and Genetic Analysis of Autosomal Recessive Retinal Dystrophies in 144 Consanguineous Pakistani Families. , 2017, 58, 2218.		34
42	Inherited cataracts: Genetic mechanisms and pathways new and old. Experimental Eye Research, 2021, 209, 108662.	2.6	34
43	A New Locus for Autosomal Recessive Nuclear Cataract Mapped to Chromosome 19q13 in a Pakistani Family. , 2005, 46, 623.		33
44	Investigating the Molecular Basis of Retinal Degeneration in a Familial Cohort of Pakistani Decent by Exome Sequencing. PLoS ONE, 2015, 10, e0136561.	2.5	33
45	The mouse Î²B1-crystallin promoter: strict regulation of lens fiber cell specificity. Biochimica Et Biophysica Acta Gene Regulatory Mechanisms, 2001, 1519, 30-38.	2.4	30
46	Missense Mutations in CRYAB Are Liable for Recessive Congenital Cataracts. PLoS ONE, 2015, 10, e0137973.	2.5	29
47	Clinical description and genome wide linkage study of Y-sutural cataract and myopia in a Chinese family. Molecular Vision, 2004, 10, 890-900.	1.1	29
48	Severe autosomal recessive retinitis pigmentosa maps to chromosome 1p13.3â€“p21.2 between D1S2896 and D1S457 but outside ABCA4. Human Genetics, 2005, 118, 356-365.	3.8	28
49	Thyroid hormone receptor beta mutations alter photoreceptor development and function in Danio rerioÂ(zebrafish). PLoS Genetics, 2020, 16, e1008869.	3.5	28
50	Polymorphism rs7278468 is associated with Age-related cataract through decreasing transcriptional activity of the CRYAA promoter. Scientific Reports, 2016, 6, 23206.	3.3	25
51	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	3.5	25
52	Novel truncation mutations in MYRF cause autosomal dominant high hyperopia mapped to 11p12â€“q13.3. Human Genetics, 2019, 138, 1077-1090.	3.8	25
53	A new locus for autosomal recessive congenital cataract identified in a Pakistani family. Molecular Vision, 2010, 16, 240-5.	1.1	25
54	Genetic Linkage of Snowflake Vitreoretinal Degeneration to Chromosome 2q36. , 2004, 45, 4498.		24

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55	Mutations in the gene encoding the alpha-subunit of rod phosphodiesterase in consanguineous Pakistani families. Molecular Vision, 2006, 12, 1283-91.	1.1	24
56	Genetic linkage of Francois-Neetens fleck (mouchetÃ©e) corneal dystrophy to chromosome 2q35. Human Genetics, 2003, 112, 593-599.	3.8	23
57	Identification and population history of CYP4V2 mutations in patients with Bietti crystalline corneoretinal dystrophy. European Journal of Human Genetics, 2017, 25, 461-471.	2.8	23
58	MITF protects against oxidative damage-induced retinal degeneration by regulating the NRF2 pathway in the retinal pigment epithelium. Redox Biology, 2020, 34, 101537.	9.0	22
59	A 5-base insertion in the Î³C-crystallin gene is associated with autosomal dominant variable zonular pulverulent cataract. Human Genetics, 2000, 106, 531-537.	3.8	21
60	Lysyl Oxidaseâ€“Like 1 Gene in the Reversal of Promoter Risk Allele in Pseudoexfoliation Syndrome. JAMA Ophthalmology, 2014, 132, 949.	2.5	21
61	Splice-site mutations identified in PDE6A responsible for retinitis pigmentosa in consanguineous Pakistani families. Molecular Vision, 2015, 21, 871-82.	1.1	20
62	Mutations in the Î²-subunit of rod phosphodiesterase identified in consanguineous Pakistani families with autosomal recessive retinitis pigmentosa. Molecular Vision, 2011, 17, 1373-80.	1.1	19
63	Autosomal recessive retinitis pigmentosa withRP1mutations is associated with myopia. British Journal of Ophthalmology, 2015, 99, 1360-1365.	3.9	18
64	Human Î²A3/A1-crystallin splicing mutation causes cataracts by activating the unfolded protein response and inducing apoptosis in differentiating lens fiber cells. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 1214-1227.	3.8	18
65	Increased Sensitivity of Amino-Arm Truncated Î²A3-Crystallin to UV-Lightâ€“Induced Photoaggregation. , 2005, 46, 3263.		17
66	Structural variations in a non-coding region at 1q32.1 are responsible for the NYS7 locus in two large families. Human Genetics, 2020, 139, 1057-1064.	3.8	17
67	Autosomal recessive retinitis pigmentosa in a Pakistani family mapped to CNGA1 with identification of a novel mutation. Molecular Vision, 2004, 10, 884-9.	1.1	17
68	A missense mutation inASRGL1is involved in causing autosomal recessive retinal degeneration. Human Molecular Genetics, 2016, 25, ddw113.	2.9	16
69	A Spontaneous Missense Mutation in Branched Chain Keto Acid Dehydrogenase Kinase in the Rat Affects Both the Central and Peripheral Nervous Systems. PLoS ONE, 2016, 11, e0160447.	2.5	16
70	Autosomal recessive congenital cataract in consanguineous Pakistani families is associated with mutations in GALK1. Molecular Vision, 2010, 16, 682-8.	1.1	16
71	Cataracts dissolved. Nature, 2015, 523, 540-541.	27.8	15
72	Overexpression of Human Î³C-crystallin 5 bp Duplication Disrupts Lens Morphology in Transgenic Mice. , 2011, 52, 5369.		13

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73	A Novel MIP Gene Mutation Analysis in a Chinese Family Affected with Congenital Progressive Punctate Cataract. PLoS ONE, 2014, 9, e102733.	2.5	13
74	Deciphering the genetic architecture and ethnographic distribution of IRD in three ethnic populations by whole genome sequence analysis. PLoS Genetics, 2021, 17, e1009848.	3.5	13
75	Novel SIL1 mutations in consanguineous Pakistani families mapping to chromosomes 5q31. Molecular Vision, 2009, 15, 1050-6.	1.1	13
76	LncRNA NEAT1 Recruits SFPQ to Regulate MITF Splicing and Control RPE Cell Proliferation. , 2021, 62, 18.		13
77	Aged Nrf2-Null Mice Develop All Major Types of Age-Related Cataracts. , 2021, 62, 10.		13
78	Analysis of RP2 and RPGR Mutations in Five X-Linked Chinese Families with Retinitis Pigmentosa. Scientific Reports, 2017, 7, 44465.	3.3	12
79	Pathogenic mutations in TULP1 responsible for retinitis pigmentosa identified in consanguineous familial cases. Molecular Vision, 2016, 22, 797-815.	1.1	12
80	Loss of function mutations in RP1 are responsible for retinitis pigmentosa in consanguineous familial cases. Molecular Vision, 2016, 22, 610-25.	1.1	11
81	A mutation in IFT43 causes non-syndromic recessive retinal degeneration. Human Molecular Genetics, 2017, 26, 4741-4751.	2.9	10
82	Functional non-coding polymorphism in an EPHA2 promoter PAX2 binding site modifies expression and alters the MAPK and AKT pathways. Scientific Reports, 2017, 7, 9992.	3.3	10
83	Mapping of a novel locus associated with autosomal recessive congenital cataract to chromosome 8p. Molecular Vision, 2010, 16, 2911-5.	1.1	10
84	Noncoding variation of the gene for ferritin light chain in hereditary and age-related cataract. Molecular Vision, 2013, 19, 835-44.	1.1	10
85	A Common Ancestral Mutation in CRYBB3 Identified in Multiple Consanguineous Families with Congenital Cataracts. PLoS ONE, 2016, 11, e0157005.	2.5	9
86	Mapping of a new locus associated with autosomal recessive congenital cataract to chromosome 3q. Molecular Vision, 2010, 16, 2634-8.	1.1	9
87	The role of FYCO1-dependent autophagy in lens fiber cell differentiation. Autophagy, 2022, 18, 2198-2215.	9.1	9
88	Novel FOXL2 mutations in two Chinese families with blepharophimosis-ptosis-epicanthus inversus syndrome. BMC Medical Genetics, 2015, 16, 73.	2.1	8
89	Phenotypic Variability Associated with the D226N Allele of IMPDH1. Ophthalmology, 2015, 122, 429-431.	5.2	8
90	Germ-line and somatic EPHA2 coding variants in lens aging and cataract. PLoS ONE, 2017, 12, e0189881.	2.5	8

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91	Changes in DNA methylation hallmark alterations in chromatin accessibility and gene expression for eye lens differentiation. Epigenetics and Chromatin, 2022, 15, 8.	3.9	8
92	Understanding the genetic architecture of human retinal degenerations. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 3904-3906.	7.1	7
93	Mutations in GRM6 identified in consanguineous Pakistani families with congenital stationary night blindness. Molecular Vision, 2015, 21, 1261-71.	1.1	7
94	Mutations in identified in families with congenital cataracts. Molecular Vision, 2020, 26, 334-344.	1.1	7
95	A missense allele of PEX5 is responsible for the defective import of PTS2 cargo proteins into peroxisomes. Human Genetics, 2021, 140, 649-666.	3.8	6
96	Pathogenicity evaluation and the genotypeâ€“phenotype analysis of OPA1 variants. Molecular Genetics and Genomics, 2021, 296, 845-862.	2.1	6
97	AIPL1 implicated in the pathogenesis of two cases of autosomal recessive retinal degeneration. Molecular Vision, 2014, 20, 1-14.	1.1	6
98	Autosomal recessive congenital cataracts linked to HSF4 in a consanguineous Pakistani family. PLoS ONE, 2019, 14, e0225010.	2.5	5
99	A novel mutation of the RPGR gene in a Chinese X-linked retinitis pigmentosa family and possible involvement of X-chromosome inactivation. Eye, 2021, 35, 1688-1696.	2.1	5
100	Novel mutations in RPE65 identified in consanguineous Pakistani families with retinal dystrophy. Molecular Vision, 2013, 19, 1554-64.	1.1	5
101	Cataracts, ataxia, short stature, and mental retardation in a Chinese family mapped to Xpter-q13.1. Journal of Human Genetics, 2006, 51, 695-700.	2.3	4
102	Overview of the Visual System. Progress in Molecular Biology and Translational Science, 2015, 134, 1-4.	1.7	4
103	Congenital Cataracts: Classification and Association With Anterior Segment Abnormalities. , 2016, 57, 6396.		3
104	Identification of Novel Deletions as the Underlying Cause of Retinal Degeneration in Two Pedigrees. Advances in Experimental Medicine and Biology, 2018, 1074, 229-236.	1.6	2
105	Whole genome sequencing data for two individuals of Pakistani descent. Scientific Data, 2018, 5, 180174.	5.3	2
106	Mutations in CERKL and RP1 cause retinitis pigmentosa in Pakistani families. Human Genome Variation, 2020, 7, 14.	0.7	2
107	Association and interaction of myopia with SNP markers rs13382811 and rs6469937 at and in Han Chinese and European populations. Molecular Vision, 2017, 23, 588-604.	1.1	2
108	Retinal Development and Pathophysiology in Kcnj13 Knockout Mice. Frontiers in Cell and Developmental Biology, 2021, 9, 810020.	3.7	2

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109	Patterns of Crystallin Gene Expression in Differentiation State Specific Regions of the Embryonic Chicken Lens. , 2022, 63, 8.		2
110	Whole-Exome Sequencing Identifies Novel Variants that Co-segregates with Autosomal Recessive Retinal Degeneration in a Pakistani Pedigree. Advances in Experimental Medicine and Biology, 2018, 1074, 219-228.	1.6	1
111	Whole genome sequencing data of multiple individuals of Pakistani descent. Scientific Data, 2020, 7, 350.	5.3	1
112	Lens Proteins and Their Molecular Biology. , 2008, , 1341-1364.		1
113	Apoferritin is maintaining the native conformation of citrate synthase. Journal of Analytical & Pharmaceutical Research, 2018, 7, 680-684.	1.0	1
114	CLCC1 c. 75C>A Mutation in Pakistani Derived Retinitis Pigmentosa Families Likely Originated With a Single Founder Mutation 2,000â€“5,000 Years Ago. Frontiers in Genetics, 2022, 13, 804924.	2.3	1
115	Size matters, but how much?. Translational Research, 2011, 158, 82-84.	5.0	0
116	Hexokinase 1 and Retinitis Pigmentosa: Insights Into the Retina and the Molecule. , 2014, 55, 7165.		0
117	Genetic Epidemiology of Congenital Cataracts and Autosomal Recessive Retinal Degenerations in Pakistan. Essentials in Ophthalmology, 2017, , 41-53.	0.1	0