S Amer Riazuddin

List of Publications by Year
in descending order

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80 papers	2,552 citations	331670 21 h-index	254184 43 g-index
83 all docs	83 docs citations	83 times ranked	2987 citing authors

#	Article	IF	Citations
1	The role of FYCO1-dependent autophagy in lens fiber cell differentiation. Autophagy, 2022, 18, 2198-2215.	9.1	9
2	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
3	Metabolome profiling of the developing murine lens. Experimental Eye Research, 2021, 202, 108343.	2.6	2
4	Detection and validation of novel mutations in MERTK in a simplex case of retinal degeneration using WGS and hiPSCâ€“RPEs model. Human Mutation, 2021, 42, 189-199.	2.5	5
5	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
6	Cigarette Smoke Triggers Loss of Corneal Endothelial Cells and Disruption of Descemet's Membrane Proteins in Mice. , 2021, 62, 3.		5
7	De novo and bi-allelic variants in AP1G1 cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. American Journal of Human Genetics, 2021, 108, 1330-1341.	6.2	18
8	Pluripotent stem cellâ€“derived corneal endothelial cells as an alternative to donor corneal endothelium in keratoplasty. Stem Cell Reports, 2021, 16, 2320-2335.	4.8	20
9	Examining the effects of cigarette smoke on mouse lens through a multi OMIC approach. Scientific Reports, 2021, 11, 18801.	3.3	1
10	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
11	Aged Nrf2-Null Mice Develop All Major Types of Age-Related Cataracts. , 2021, 62, 10.		13
12	Modulating EGFR-MTORC1-autophagy as a potential therapy for persistent fetal vasculature (PFV) disease. Autophagy, 2020, 16, 1130-1142.	9.1	12
13	Whole genome sequencing data of multiple individuals of Pakistani descent. Scientific Data, 2020, 7, 350.	5.3	1
14	Mutations in CERKL and RP1 cause retinitis pigmentosa in Pakistani families. Human Genome Variation, 2020, 7, 14.	0.7	2
15	Generation and proteome profiling of PBMC-originated, iPSC-derived lentoid bodies. Stem Cell Research, 2020, 46, 101813.	0.7	11
16	Novel mutations in identified in familial cases of primary congenital glaucoma. Molecular Vision, 2020, 26, 14-25.	1.1	4
17	Mutations in identified in families with congenital cataracts. Molecular Vision, 2020, 26, 334-344.	1.1	7
18	Autosomal recessive congenital cataracts linked to HSF4 in a consanguineous Pakistani family. PLoS ONE, 2019, 14, e0225010.	2.5	5

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19	Comparative transcriptome analysis of hESC- and iPSC-derived lentoid bodies. Scientific Reports, 2019, 9, 18552.	3.3	15
20	Identification of a Novel TCF4 Isoform in the Human Corneal Endothelium. Cornea, 2018, 37, 899-903.	1.7	3
21	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
22	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
23	Whole genome sequencing data for two individuals of Pakistani descent. Scientific Data, 2018, 5, 180174.	5.3	2
24	Generation and Proteome Profiling of PBMC-Originated, iPSC-Derived Corneal Endothelial Cells. , 2018, 59, 2437.		24
25	Pilot Study of Audiometric Patterns in Fuchs Corneal Dystrophy. Journal of Speech, Language, and Hearing Research, 2018, 61, 2604-2608.	1.6	1
26	Proteome Profiling of Developing Murine Lens Through Mass Spectrometry. , 2018, 59, 100.		21
27	Comparative transcriptome analysis of hESC- and iPSC-derived corneal endothelial cells. Experimental Eye Research, 2018, 176, 252-257.	2.6	16
28	A novel LRAT mutation affecting splicing in a family with early onset retinitis pigmentosa. Human Genomics, 2018, 12, 35.	2.9	10
29	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	3.5	25
30	IFT88 mutations identified in individuals with non-syndromic recessive retinal degeneration result in abnormal ciliogenesis. Human Genetics, 2018, 137, 447-458.	3.8	11
31	Identification of novel transcripts and peptides in developing murine lens. Scientific Reports, 2018, 8, 11162.	3.3	5
32	Genetic analysis of 10 pedigrees with inherited retinal degeneration by exome sequencing and phenotype-genotype association. Physiological Genomics, 2017, 49, 216-229.	2.3	23
33	Genome-wide association study identifies three novel loci in Fuchs endothelial corneal dystrophy. Nature Communications, 2017, 8, 14898.	12.8	101
34	A mutation in IFT43 causes non-syndromic recessive retinal degeneration. Human Molecular Genetics, 2017, 26, 4741-4751.	2.9	10
35	Molecular Genetic Analysis of Pakistani Families With Autosomal Recessive Congenital Cataracts by Homozygosity Screening. , 2017, 58, 2207.		45
36	Homozygosity Mapping and Genetic Analysis of Autosomal Recessive Retinal Dystrophies in 144 Consanguineous Pakistani Families. , 2017, 58, 2218.		34

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37	CTG18.1 Expansion in TCF4 Among African Americans With Fuchs' Corneal Dystrophy. , 2017, 58, 6046.		9
38	A Common Ancestral Mutation in CRYBB3 Identified in Multiple Consanguineous Families with Congenital Cataracts. PLoS ONE, 2016, 11, e0157005.	2.5	9
39	Mutation in LIM2 Is Responsible for Autosomal Recessive Congenital Cataracts. PLoS ONE, 2016, 11, e0162620.	2.5	17
40	A spectrum of CYP1B1 mutations associated with primary congenital glaucoma in families of Pakistani descent. Human Genome Variation, 2016, 3, 16021.	0.7	20
41	Mutations in phosphodiesterase 6 identified in familial cases of retinitis pigmentosa. Human Genome Variation, 2016, 3, 16036.	0.7	8
42	A missense mutation inASRGL1is involved in causing autosomal recessive retinal degeneration. Human Molecular Genetics, 2016, 25, ddw113.	2.9	16
43	FOXE3 contributes to Peters anomaly through transcriptional regulation of an autophagy-associated protein termed DNAJB1. Nature Communications, 2016, 7, 10953.	12.8	35
44	Non-coding RNA profiling of the developing murine lens. Experimental Eye Research, 2016, 145, 347-351.	2.6	21
45	Deletion at the GCNT2 Locus Causes Autosomal Recessive Congenital Cataracts. PLoS ONE, 2016, 11, e0167562.	2.5	9
46	Loss of function mutations in RP1 are responsible for retinitis pigmentosa in consanguineous familial cases. Molecular Vision, 2016, 22, 610-25.	1.1	11
47	Pathogenic mutations in TULP1 responsible for retinitis pigmentosa identified in consanguineous familial cases. Molecular Vision, 2016, 22, 797-815.	1.1	12
48	Transcriptome Profiling of Developing Murine Lens Through RNA Sequencing. , 2015, 56, 4919.		44
49	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
50	Missense Mutations in CRYAB Are Liable for Recessive Congenital Cataracts. PLoS ONE, 2015, 10, e0137973.	2.5	29
51	Phenotypic Variability Associated with the D226N Allele of IMPDH1. Ophthalmology, 2015, 122, 429-431.	5.2	8
52	Fuchs Corneal Dystrophy. Progress in Molecular Biology and Translational Science, 2015, 134, 79-97.	1.7	56
53	Lens Biology and Biochemistry. Progress in Molecular Biology and Translational Science, 2015, 134, 169-201.	1.7	71
54	Overview of the Cornea. Progress in Molecular Biology and Translational Science, 2015, 134, 7-23.	1.7	200

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55	Autosomal recessive retinitis pigmentosa withRP1mutations is associated with myopia. British Journal of Ophthalmology, 2015, 99, 1360-1365.	3.9	18
56	Splice-site mutations identified in PDE6A responsible for retinitis pigmentosa in consanguineous Pakistani families. Molecular Vision, 2015, 21, 871-82.	1.1	20
57	Mutations in GRM6 identified in consanguineous Pakistani families with congenital stationary night blindness. Molecular Vision, 2015, 21, 1261-71.	1.1	7
58	AIPL1 implicated in the pathogenesis of two cases of autosomal recessive retinal degeneration. Molecular Vision, 2014, 20, 1-14.	1.1	6
59	Mutations in AGBL1 Cause Dominant Late-Onset Fuchs Corneal Dystrophy and Alter Protein-Protein Interaction with TCF4. American Journal of Human Genetics, 2013, 93, 758-764.	6.2	86
60	Novel mutations in RPE65 identified in consanguineous Pakistani families with retinal dystrophy. Molecular Vision, 2013, 19, 1554-64.	1.1	5
61	Mutations in LOXHD1, a Recessive-Deafness Locus, Cause Dominant Late-Onset Fuchs Corneal Dystrophy. American Journal of Human Genetics, 2012, 90, 533-539.	6.2	141
62	Replication of theTCF4Intronic Variant in Late-Onset Fuchs Corneal Dystrophy and Evidence of Independence from theFCD2Locus. , 2011, 52, 2825.		61
63	Mutations in FYCO1 Cause Autosomal-Recessive Congenital Cataracts. American Journal of Human Genetics, 2011, 88, 827-838.	6.2	132
64	Mutations in RLBP1 associated with fundus albipunctatus in consanguineous Pakistani families. British Journal of Ophthalmology, 2011, 95, 1019-1024.	3.9	35
65	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
66	Missense Mutations in TCF8 Cause Late-Onset Fuchs Corneal Dystrophy and Interact with FCD4 on Chromosome 9p. American Journal of Human Genetics, 2010, 86, 45-53.	6.2	167
67	A Splice-Site Mutation in a Retina-Specific Exon of BBS8 Causes Nonsyndromic Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 86, 805-812.	6.2	109
68	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
69	A Mutation in SLC24A1 Implicated in Autosomal-Recessive Congenital Stationary Night Blindness. American Journal of Human Genetics, 2010, 87, 523-531.	6.2	67
70	Missense mutations in the sodium borate cotransporter SLC4A11 cause late-onset Fuchs corneal dystrophya. Human Mutation, 2010, 31, 1261-1268.	2.5	117
71	A new locus for autosomal recessive congenital cataract identified in a Pakistani family. Molecular Vision, 2010, 16, 240-5.	1.1	25
72	Autosomal recessive congenital cataract linked to EPHA2 in a consanguineous Pakistani family. Molecular Vision, 2010, 16, 511-7.	1.1	81

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73	Mapping of a new locus associated with autosomal recessive congenital cataract to chromosome 3q. Molecular Vision, 2010, 16, 2634-8.	1.1	9
74	Mapping of a novel locus associated with autosomal recessive congenital cataract to chromosome 8p. Molecular Vision, 2010, 16, 2911-5.	1.1	10
75	Linkage of a Mild Late-Onset Phenotype of Fuchs Corneal Dystrophy to a Novel Locus at 5q33.1-q35.2. , 2009, 50, 5667.		80
76	Mutations in the gene encoding the alpha-subunit of rod phosphodiesterase in consanguineous Pakistani families. Molecular Vision, 2006, 12, 1283-91.	1.1	24
77	Autosomal Recessive Retinitis Pigmentosa Is Associated with Mutations inRP1in Three Consanguineous Pakistani Families. , 2005, 46, 2264.		50
78	Mutations in Î²B3-Crystallin Associated with Autosomal Recessive Cataract in Two Pakistani Families. , 2005, 46, 2100.		97
79	A New Locus for Autosomal Recessive Nuclear Cataract Mapped to Chromosome 19q13 in a Pakistani Family. , 2005, 46, 623.		33
80	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