Manir Ali

List of Publications by Year in descending order

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66 papers	4,299 citations	201575 27 h-index	161767 54 g-index
70 all docs	70 docs citations	70 times ranked	6229 citing authors

#	Article	IF	Citations
1	Mutations in the gene encoding the 3â€²-5â€² DNA exonuclease TREX1 cause Aicardi-GoutiÃ¨res syndrome at the AGS1 locus. Nature Genetics, 2006, 38, 917-920.	9.4	752
2	Mutations involved in Aicardi-GoutiÃ¨res syndrome implicate SAMHD1 as regulator of the innate immune response. Nature Genetics, 2009, 41, 829-832.	9.4	610
3	Mutations in genes encoding ribonuclease H2 subunits cause Aicardi-GoutiÃ¨res syndrome and mimic congenital viral brain infection. Nature Genetics, 2006, 38, 910-916.	9.4	592
4	Null Mutations in LTBP2 Cause Primary Congenital Glaucoma. American Journal of Human Genetics, 2009, 84, 664-671.	2.6	255
5	Mutations in TSPAN12 Cause Autosomal-Dominant Familial Exudative Vitreoretinopathy. American Journal of Human Genetics, 2010, 86, 248-253.	2.6	161
6	ZNF408 is mutated in familial exudative vitreoretinopathy and is crucial for the development of zebrafish retinal vasculature. Proceedings of the National Academy of Sciences of the United States of America, 2013, 110, 9856-9861.	3.3	144
7	Mutations in REEP6 Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2016, 99, 1305-1315.	2.6	121
8	CFH,VEGFandHTRA1promoter genotype may influence the response to intravitreal ranibizumab therapy for neovascular age-related macular degeneration. British Journal of Ophthalmology, 2012, 96, 208-212.	2.1	98
9	Loss of the Metalloprotease ADAM9 Leads to Cone-Rod Dystrophy in Humans and Retinal Degeneration in Mice. American Journal of Human Genetics, 2009, 84, 683-691.	2.6	76
10	Defects in the Cell Signaling Mediator Î²-Catenin Cause the Retinal Vascular Condition FEVR. American Journal of Human Genetics, 2017, 100, 960-968.	2.6	74
11	Mutational Analysis ofMIR184in Sporadic Keratoconus and Myopia. , 2013, 54, 5266.		73
12	Recessive Mutations in SLC38A8 Cause Foveal Hypoplasia and Optic Nerve Misrouting without Albinism. American Journal of Human Genetics, 2013, 93, 1143-1150.	2.6	71
13	Homozygous Mutations in PXDN Cause Congenital Cataract, Corneal Opacity, and Developmental Glaucoma. American Journal of Human Genetics, 2011, 89, 464-473.	2.6	68
14	Next generation sequencing identifies mutations in Atonal homolog 7 (ATOH7) in families with global eye developmental defects. Human Molecular Genetics, 2012, 21, 776-783.	1.4	66
15	Genotype-Phenotype Correlation for Leber Congenital Amaurosis in Northern Pakistan. JAMA Ophthalmology, 2010, 128, 107.	2.6	64
16	Recessive Mutations inTSPAN12Cause Retinal Dysplasia and Severe Familial Exudative Vitreoretinopathy (FEVR). , 2012, 53, 2873.		64
17	Matrix metalloproteinases in keratoconus â€“ Too much of a good thing?. Experimental Eye Research, 2019, 182, 137-143.	1.2	49
18	Congenital Hereditary Endothelial Dystrophy Caused by SLC4A11 Mutations Progresses to Harboyan Syndrome. Cornea, 2014, 33, 247-251.	0.9	47

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19	Association Between Missense Mutations in theBBS2Gene and Nonsyndromic Retinitis Pigmentosa. JAMA Ophthalmology, 2015, 133, 312.	1.4	43
20	Association of Steroid 5Î±-Reductase Type 3 Congenital Disorder of Glycosylation With Early-Onset Retinal Dystrophy. JAMA Ophthalmology, 2017, 135, 339.	1.4	43
21	Biallelic Mutations in the Autophagy Regulator DRAM2 Cause Retinal Dystrophy with Early Macular Involvement. American Journal of Human Genetics, 2015, 96, 948-954.	2.6	42
22	Mutation in the Guanine Nucleotideâ€“Binding Protein Î²-3 Causes Retinal Degeneration and Embryonic Mortality in Chickens. , 2006, 47, 4714.		41
23	DYNC2H1 hypomorphic or retina-predominant variants cause nonsyndromic retinal degeneration. Genetics in Medicine, 2020, 22, 2041-2051.	1.1	38
24	A multi-ethnic genome-wide association study implicates collagen matrix integrity and cell differentiation pathways in keratoconus. Communications Biology, 2021, 4, 266.	2.0	36
25	Rheumatoid arthritis synovial T cells regulate transcription of several genes associated with antigen-induced anergy. Journal of Clinical Investigation, 2001, 107, 519-528.	3.9	36
26	Specific Alleles of CLN7/MFSD8, a Protein That Localizes to Photoreceptor Synaptic Terminals, Cause a Spectrum of Nonsyndromic Retinal Dystrophy. , 2017, 58, 2906.		35
27	Ultrastructural changes in the retinopathy, globe enlarged (rge) chick cornea. Journal of Structural Biology, 2009, 166, 195-204.	1.3	33
28	Homozygous FOXE3 mutations cause non-syndromic, bilateral, total sclerocornea, aphakia, microphthalmia and optic disc coloboma. Molecular Vision, 2010, 16, 1162-8.	1.1	33
29	The effect of <scp>COMT</scp> Val158Met and <scp>DRD</scp>2 C957T polymorphisms on executive function and the impact of early life stress. Brain and Behavior, 2017, 7, e00695.	1.0	31
30	Collagen organization in the chicken cornea and structural alterations in the retinopathy, globe enlarged (rge) phenotypeâ€”An X-ray diffraction study. Journal of Structural Biology, 2008, 161, 1-8.	1.3	30
31	A missense variant in CST3 exerts a recessive effect on susceptibility to age-related macular degeneration resembling its association with Alzheimerâ€™s disease. Human Genetics, 2015, 134, 705-715.	1.8	30
32	Association of Genetic Variants With Response to Antiâ€“Vascular Endothelial Growth Factor Therapy in Age-Related Macular Degeneration. JAMA Ophthalmology, 2018, 136, 875.	1.4	30
33	Mutations in the polyglutamylase geneTTLL5, expressed in photoreceptor cells and spermatozoa, are associated with cone-rod degeneration and reduced male fertility. Human Molecular Genetics, 2016, 25, ddw282.	1.4	27
34	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.	2.5	25
35	A missense mutation in the nuclear localization signal sequence of CERKL (p.R106S) causes autosomal recessive retinal degeneration. Molecular Vision, 2008, 14, 1960-4.	1.1	25
36	MpdzNull Allele in an Avian Model of Retinal Degeneration and Mutations in Human Leber Congenital Amaurosis and Retinitis Pigmentosa. , 2011, 52, 7432.		24

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37	Application of differential display to immunological research. Journal of Immunological Methods, 2001, 250, 29-43.	0.6	23
38	Mutation Screening of Retinal Dystrophy Patients by Targeted Capture from Tagged Pooled DNAs and Next Generation Sequencing. PLoS ONE, 2014, 9, e104281.	1.1	20
39	Identification of Inherited Retinal Disease-Associated Genetic Variants in 11 Candidate Genes. Genes, 2018, 9, 21.	1.0	20
40	Clinical phenotype associated with homozygosity forÂaÂHOXD13 7-residue polyalanine tract expansion. European Journal of Medical Genetics, 2006, 49, 396-401.	0.7	19
41	A new recessively inherited disorder composed of foveal hypoplasia, optic nerve decussation defects and anterior segment dysgenesis maps to chromosome 16q23.3-24.1. Molecular Vision, 2013, 19, 2165-72.	1.1	19
42	The Influence of Lamellar Orientation on Corneal Material Behavior: Biomechanical and Structural Changes in an Avian Corneal Disorder. , 2011, 52, 1243.		18
43	Mutation spectrum and clinical investigation of achromatopsia patients with mutations in the GNAT2 gene. Human Mutation, 2019, 40, 1145-1155.	1.1	15
44	Biallelic sequence and structural variants in RAX2 are a novel cause for autosomal recessive inherited retinal disease. Genetics in Medicine, 2019, 21, 1319-1329.	1.1	15
45	Homozygous single base deletion in TUSC3 causes intellectual disability with developmental delay in an Omani family. American Journal of Medical Genetics, Part A, 2016, 170, 1826-1831.	0.7	14
46	PDZD8 Disruption Causes Cognitive Impairment in Humans, Mice, and Fruit Flies. Biological Psychiatry, 2022, 92, 323-334.	0.7	14
47	Genetic Heterogeneity for Recessively Inherited Congenital Cataract Microcornea with Corneal Opacity. , 2011, 52, 4294.		13
48	Spectral domain optical coherence tomography imaging of the posterior segment of the eye in the retinal dysplasia and degeneration chicken, an animal model of inherited retinal degeneration. Veterinary Ophthalmology, 2014, 17, 113-119.	0.6	13
49	Disease Expression in Autosomal Recessive Retinal Dystrophy Associated With Mutations in theDRAM2Gene. , 2015, 56, 8083.		13
50	DETAILED RETINAL IMAGING IN CARRIERS OF OCULAR ALBINISM. Retina, 2018, 38, 620-628.	1.0	13
51	The D153del Mutation in GNB3 Gene Causes Tissue Specific Signalling Patterns and an Abnormal Renal Morphology in Rge Chickens. PLoS ONE, 2011, 6, e21156.	1.1	11
52	Novel C8orf37 mutations cause retinitis pigmentosa in consanguineous families of Pakistani origin. Molecular Vision, 2015, 21, 236-43.	1.1	10
53	Profiling Retinal Biochemistry in the MPDZ Mutant Retinal Dysplasia and Degeneration Chick: A Model of Human RP and LCA. , 2012, 53, 413.		9
54	New variants and in silico analyses in GRK1 associated Oguchi disease. Human Mutation, 2021, 42, 164-176.	1.1	7

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55	LHFPL5 mutation: A rare cause of non-syndromic autosomal recessive hearing loss. European Journal of Medical Genetics, 2019, 62, 103592.	0.7	6
56	An X-Ray Scattering Study into the Structural Basis of Corneal Refractive Function in an Avian Model. Biophysical Journal, 2013, 104, 2586-2594.	0.2	5
57	Reply to Papanikolaou et al. British Journal of Ophthalmology, 2011, 95, 890-890.	2.1	4
58	Identification of autosomal recessive disease loci using out-bred nuclear families. Human Mutation, 2012, 33, 338-342.	1.1	4
59	Simple and Efficient Identification of Rare Recessive Pathologically Important Sequence Variants from Next Generation Exome Sequence Data. Human Mutation, 2013, 34, 945-952.	1.1	4
60	Retinal Phenotypes in Patients Homozygous for the G1961E Mutation in theABCA4Gene. , 2013, 54, 520.		3
61	A Recessively Inherited Risk Locus on Chromosome 13q22-31 Conferring Susceptibility to Schizophrenia. Schizophrenia Bulletin, 2021, 47, 796-802.	2.3	3
62	Rapid Visualisation of Microarray Copy Number Data for the Detection of Structural Variations Linked to a Disease Phenotype. PLoS ONE, 2012, 7, e43466.	1.1	1
63	Use of a gene-based case-control association approach in exome sequencing data to elucidate the molecular basis of a mendelian phenotype. Lancet, The, 2017, 389, S14.	6.3	1
64	Changing the status quo bias. British Journal of Ophthalmology, 2011, 95, 1034-1034.	2.1	0
65	Patterns of inheritance, not always easily visible. BMJ, The, 2013, 347, f6610-f6610.	3.0	0
66	Differential Display Reverse Transcription-Polymerase Chain Reaction to Identify Novel Biomolecules in Arthritis Research. Methods in Molecular Medicine, 2007, 136, 329-347.	0.8	0