Raheel Qamar

List of Publications by Year
in descending order

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90 papers	3,009 citations	257450 24 h-index	182427 51 g-index
94 all docs	94 docs citations	94 times ranked	6648 citing authors

#	Article	IF	Citations
1	The Genetic Legacy of the Mongols. American Journal of Human Genetics, 2003, 72, 717-721.	6.2	512
2	Y-Chromosomal DNA Variation in Pakistan. American Journal of Human Genetics, 2002, 70, 1107-1124.	6.2	213
3	Next generation sequencing-based molecular diagnosis of retinitis pigmentosa: identification of a novel genotype-phenotype correlation and clinical refinements. Human Genetics, 2014, 133, 331-345.	3.8	204
4	Genome-wide association study identifies five new susceptibility loci for primary angle closure glaucoma. Nature Genetics, 2016, 48, 556-562.	21.4	147
5	ABCA4 midigenes reveal the full splice spectrum of all reported noncanonical splice site variants in Stargardt disease. Genome Research, 2018, 28, 100-110.	5.5	134
6	Y-Chromosome Lineages Trace Diffusion of People and Languages in Southwestern Asia. American Journal of Human Genetics, 2001, 68, 537-542.	6.2	131
7	Genetic association study of exfoliation syndrome identifies a protective rare variant at LOXL1 and five new susceptibility loci. Nature Genetics, 2017, 49, 993-1004.	21.4	114
8	Mutations in IMPG2, Encoding Interphotoreceptor Matrix Proteoglycan 2, Cause Autosomal-Recessive Retinitis Pigmentosa. American Journal of Human Genetics, 2010, 87, 199-208.	6.2	98
9	Differential Structuring of Human Populations for Homologous X and Y Microsatellite Loci. American Journal of Human Genetics, 1997, 61, 719-733.	6.2	70
10	Comprehensive genotyping reveals RPE65 as the most frequently mutated gene in Leber congenital amaurosis in Denmark. European Journal of Human Genetics, 2016, 24, 1071-1079.	2.8	69
11	Network Analyses of Y-Chromosomal Types in Europe, Northern Africa, and Western Asia Reveal Specific Patterns of Geographic Distribution. American Journal of Human Genetics, 1998, 63, 847-860.	6.2	63
12	Association of eNOS and HSP70 gene polymorphisms with glaucoma in Pakistani cohorts. Molecular Vision, 2010, 16, 18-25.	1.1	54
13	Genetic Spectrum of Autosomal Recessive Non-Syndromic Hearing Loss in Pakistani Families. PLoS ONE, 2014, 9, e100146.	2.5	52
14	A missense mutation in the splicing factor geneDHX38is associated with early-onset retinitis pigmentosa with macular coloboma. Journal of Medical Genetics, 2014, 51, 444-448.	3.2	48
15	MTHFR gene C677T and A1298C polymorphisms and homocysteine levels in primary open angle and primary closed angle glaucoma. Molecular Vision, 2009, 15, 2268-78.	1.1	41
16	C677T polymorphism in the methylenetetrahydrofolate reductase gene is associated with primary closed angle glaucoma. Molecular Vision, 2008, 14, 661-5.	1.1	39
17	IMPG2-Associated Retinitis Pigmentosa Displays Relatively Early Macular Involvement. , 2014, 55, 3939.		37
18	Screening of a Large Cohort of Leber Congenital Amaurosis and Retinitis Pigmentosa Patients Identifies Novel LCA5 Mutations and New Genotype-Phenotype Correlations. Human Mutation, 2013, 34, 1537-1546.	2.5	32

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19	Patient HLA-DRB1* and -DQB1* allele and haplotype association with hepatitis C virus persistence and clearance. Journal of General Virology, 2010, 91, 1931-1938.	2.9	31
20	Taurine treatment of retinal degeneration and cardiomyopathy in a consanguineous family with SLC6A6 taurine transporter deficiency. Human Molecular Genetics, 2020, 29, 618-623.	2.9	29
21	Whole exome sequencing identifies a heterozygous missense variant in the PRDM5 gene in a family with Axenfeldâ€“Rieger syndrome. Neurogenetics, 2016, 17, 17-23.	1.4	28
22	Novel and recurrent CIB2 variants, associated with nonsyndromic deafness, do not affect calcium buffering and localization in hair cells. European Journal of Human Genetics, 2016, 24, 542-549.	2.8	28
23	Homozygosity Mapping and Targeted Sanger Sequencing Reveal Genetic Defects Underlying Inherited Retinal Disease in Families from Pakistan. PLoS ONE, 2015, 10, e0119806.	2.5	27
24	Association of known common genetic variants with primary open angle, primary angle closure, and pseudoexfoliation glaucoma in Pakistani cohorts. Molecular Vision, 2014, 20, 1471-9.	1.1	27
25	Exome sequencing identifies a novel and a recurrent BBS1 mutation in Pakistani families with Bardet-Biedl syndrome. Molecular Vision, 2013, 19, 644-53.	1.1	26
26	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	3.5	25
27	Polymorphisms in matrix metalloproteinases MMP1 and MMP9 are associated with primary open-angle and angle closure glaucoma in a Pakistani population. Molecular Vision, 2013, 19, 441-7.	1.1	25
28	De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder. Genetics in Medicine, 2020, 22, 538-546.	2.4	24
29	Exome Sequencing Identifies Three Novel Candidate Genes Implicated in Intellectual Disability. PLoS ONE, 2014, 9, e112687.	2.5	23
30	Identification of novel <scp>CYP1B</scp>1 gene mutations in patients with primary congenital and primary openâ€angle glaucoma. Clinical and Experimental Ophthalmology, 2015, 43, 31-39.	2.6	22
31	The protective variant rs7173049 at LOXL1 locus impacts on retinoic acid signaling pathway in pseudoexfoliation syndrome. Human Molecular Genetics, 2019, 28, 2531-2548.	2.9	22
32	A novel mutation in GRK1 causes Oguchi disease in a consanguineous Pakistani family. Molecular Vision, 2009, 15, 1788-93.	1.1	22
33	Association of ABO blood groups with glaucoma in the Pakistani population. Canadian Journal of Ophthalmology, 2009, 44, 582-586.	0.7	21
34	Clinical Utility of a Coronary Heart Disease Risk Prediction Gene Score in UK Healthy Middle Aged Men and in the Pakistani Population. PLoS ONE, 2015, 10, e0130754.	2.5	21
35	The Molecular Basis of Retinal Dystrophies in Pakistan. Genes, 2014, 5, 176-195.	2.4	20
36	Role of tissue plasminogen activator and plasminogen activator inhibitor polymorphism in myocardial infarction. Molecular Biology Reports, 2011, 38, 2541-2548.	2.3	19

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37	Implementation of public health genomics in Pakistan. European Journal of Human Genetics, 2019, 27, 1485-1492.	2.8	19
38	Role of ACE and PAI-1 Polymorphisms in the Development and Progression of Diabetic Retinopathy. PLoS ONE, 2015, 10, e0144557.	2.5	19
39	Identification of Mutations in the PRDM5 Gene in Brittle Cornea Syndrome. Cornea, 2016, 35, 853-859.	1.7	18
40	Role of Lysyl oxidase-like 1 gene polymorphisms in Pakistani patients with pseudoexfoliative glaucoma. Molecular Vision, 2012, 18, 1040-4.	1.1	18
41	A nonsense mutation in S-antigen (p.Glu306*) causes Oguchi disease. Molecular Vision, 2012, 18, 1253-9.	1.1	18
42	Association of tumor necrosis factor alpha gene polymorphism G-308A with pseudoexfoliative glaucoma in the Pakistani population. Molecular Vision, 2009, 15, 2861-7.	1.1	17
43	Missense mutations at homologous positions in the fourth and fifth laminin A G-like domains of eyes shut homolog cause autosomal recessive retinitis pigmentosa. Molecular Vision, 2010, 16, 2753-9.	1.1	17
44	Identification of recurrent and novel mutations in TULP1 in Pakistani families with early-onset retinitis pigmentosa. Molecular Vision, 2012, 18, 1226-37.	1.1	17
45	Identification of a novel FBN1 gene mutation in a large Pakistani family with Marfan syndrome. Molecular Vision, 2012, 18, 1918-26.	1.1	17
46	Association of Pro12Ala polymorphism in peroxisome proliferator activated receptor gamma with proliferative diabetic retinopathy. Molecular Vision, 2013, 19, 710-7.	1.1	17
47	Molecular Mechanisms of Complement System Proteins and Matrix Metalloproteinases in the Pathogenesis of Age-Related Macular Degeneration. Current Molecular Medicine, 2019, 19, 705-718.	1.3	16
48	XRCC1 and XPD DNA repair gene polymorphisms: a potential risk factor for glaucoma in the Pakistani population. Molecular Vision, 2011, 17, 1153-63.	1.1	16
49	Novel mutation in AAA domain of BCS1L causing Bjornstad syndrome. Journal of Human Genetics, 2013, 58, 819-821.	2.3	15
50	A homozygous p.Glu150Lys mutation in the opsin gene of two Pakistani families with autosomal recessive retinitis pigmentosa. Molecular Vision, 2009, 15, 2526-34.	1.1	15
51	The association of glutathione S-transferase GSTT1 and GSTM1 gene polymorphism with pseudoexfoliative glaucoma in a Pakistani population. Molecular Vision, 2010, 16, 2146-52.	1.1	15
52	Novel mutations in RDH5 cause fundus albipunctatus in two consanguineous Pakistani families. Molecular Vision, 2012, 18, 1558-71.	1.1	15
53	Identification of a recurrent insertion mutation in the LDLR gene in a Pakistani family with autosomal dominant hypercholesterolemia. Molecular Biology Reports, 2010, 37, 3869-3875.	2.3	14
54	CBS mutations and MTFHR SNPs causative of hyperhomocysteinemia in Pakistani children. Molecular Biology Reports, 2018, 45, 353-360.	2.3	13

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55	Association of IGF1 and VEGFA polymorphisms with diabetic retinopathy in Pakistani population. Acta Diabetologica, 2020, 57, 237-245.	2.5	13
56	Novel CNGA3 and CNGB3 mutations in two Pakistani families with achromatopsia. Molecular Vision, 2010, 16, 774-81.	1.1	13
57	The genetic spectrum of familial hypercholesterolemia in Pakistan. Clinica Chimica Acta, 2013, 421, 219-225.	1.1	12
58	A complex microcephaly syndrome in a Pakistani family associated with a novel missense mutation in RBBP8 and a heterozygous deletion in NRXN1. Gene, 2014, 538, 30-35.	2.2	11
59	VNTR Polymorphism of the DRD4 Locus in Different Pakistani Ethnic Groups. Genetic Testing and Molecular Biomarkers, 2008, 12, 299-304.	1.7	10
60	A novel homozygous 10 nucleotide deletion in BBS10 causes Bardetâ€“Biedl syndrome in a Pakistani family. Gene, 2013, 519, 177-181.	2.2	10
61	Association of a Polymorphism in the BIRC6 Gene with Pseudoexfoliative Glaucoma. PLoS ONE, 2014, 9, e105023.	2.5	10
62	Variants in the ASB10 Gene Are Associated with Primary Open Angle Glaucoma. PLoS ONE, 2015, 10, e0145005.	2.5	10
63	Zika virus in Pakistan: the tip of the iceberg?. The Lancet Global Health, 2016, 4, e913-e914.	6.3	10
64	Identification of novel potential genetic predictors of urothelial bladder carcinoma susceptibility in Pakistani population. Familial Cancer, 2017, 16, 577-594.	1.9	9
65	Synthesis and separation of a diastereomeric pair of phosphonopeptide inhibitors of the cyclic AMP-dependent protein kinase catalytic subunit. Tetrahedron, 1994, 50, 1919-1926.	1.9	8
66	Molecular evidence for the presence of huanglongbing in Pakistan. Australasian Plant Disease Notes, 2007, 2, 37.	0.7	8
67	A Single SNP Surrogate for Genotyping HLA-C*06:02 in Diverse Populations. Journal of Investigative Dermatology, 2015, 135, 1177-1180.	0.7	8
68	Variants in the PRPF8 Gene are Associated with Glaucoma. Molecular Neurobiology, 2018, 55, 4504-4510.	4.0	8
69	MTHFR polymorphisms as risk for male infertility in Pakistan and its comparison with socioeconomic status in the world. Personalized Medicine, 2019, 16, 35-49.	1.5	8
70	The effect of varied pH on the luminescence characteristics of antibodyâ€“mercaptoacetic acid conjugated ZnS nanowires. Chemical Physics, 2017, 497, 24-31.	1.9	6
71	Electrochemically driven optical and SERS immunosensor for the detection of a therapeutic cardiac drug. RSC Advances, 2022, 12, 2901-2913.	3.6	6
72	Novel and recurrent LDLR gene mutations in Pakistani hypercholesterolemia patients. Molecular Biology Reports, 2012, 39, 7365-7372.	2.3	5

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73	Comprehensive Registration of DNA Sequence Variants Associated with Inherited Retinal Diseases in Leiden Open Variation Databases. Human Mutation, 2014, 35, 147-148.	2.5	5
74	The Development of Computational Biology in Pakistan: Still a Long Way to Go. PLoS Computational Biology, 2011, 7, e1001135.	3.2	4
75	Founder mutation c.676insC in three unrelated Kindler syndrome families belonging to a particular clan from Pakistan. Journal of Dermatology, 2012, 39, 640-641.	1.2	4
76	A canonical splice site mutation in GIPC3 causes sensorineural hearing loss in a large Pakistani family. Journal of Human Genetics, 2014, 59, 683-686.	2.3	4
77	The adverse role of excess negative ions in reducing the photoluminescence from water soluble MAAâ€“CdSe/ZnS quantum dots in various phosphate buffers. Physical Chemistry Chemical Physics, 2018, 20, 29446-29451.	2.8	4
78	Association of rs10490924 in ARMS2 / HTRA1 with ageâ€related macular degeneration in the Pakistani population. Annals of Human Genetics, 2019, 83, 285-290.	0.8	4
79	A study of ACE, eNOS and MTHFR association with psoriasis in Pakistani population. Meta Gene, 2018, 15, 65-69.	0.6	3
80	A 3â€² untranslated region polymorphism rs2304277 in the DNA repair pathway geneOGG1is a novel risk modulator for urothelial bladder carcinoma. Annals of Human Genetics, 2018, 82, 74-87.	0.8	3
81	Effect of gasotransmitters treatment on expression of hypertension, vascular and cardiac remodeling and hypertensive nephropathy genes in left ventricular hypertrophy. Gene, 2020, 737, 144479.	2.2	3
82	The PlantProm DB: Recent Updates. , 2012, , .		2
83	ANRIL polymorphism rs1333049, a novel genetic predictor for diabetic retinopathy complication. Meta Gene, 2017, 14, 33-37.	0.6	2
84	A 2-year retrospective study of viral and host-associated risk factors in Pakistani hepatocellular carcinoma patients. European Journal of Gastroenterology and Hepatology, 2019, 31, 1103-1109.	1.6	2
85	Phytochemical Screening and Protective Effects of Prunus persica Seeds Extract on Carbon Tetrachloride-Induced Hepatic Injury in Rats. Current Pharmaceutical Biotechnology, 2022, 23, 158-170.	1.6	2
86	ATF6 polymorphisms and protective effect in diabetic retinopathy. Meta Gene, 2018, 17, 56-60.	0.6	1
87	A Method for Counting Active Sites of Cyclic Amp-Dependent Protein Kinase. Journal of Enzyme Inhibition and Medicinal Chemistry, 1993, 7, 151-157.	0.5	0
88	The Spectrum of Mutations In Î²-Thalassaemic Patients and Carriers From Punjab and N.W.F.J. in Pakistan. Natural Product Research, 1998, 12, 199-207.	0.4	0
89	Compound heterozygous mutations p.Q1530X and 6103delG in COL7A1 causing recessive dystrophic epidermolysis bullosa in a Pakistani family. Journal of Dermatology, 2012, 39, 472-474.	1.2	0
90	Glaucoma Genetics in Pakistan. Essentials in Ophthalmology, 2021, , 233-249.	0.1	0