Muhammad Asif Naeem

List of Publications by Year in descending order

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#	Article	IF	CITATIONS
1	CLCC1 c. 75C>A Mutation in Pakistani Derived Retinitis Pigmentosa Families Likely Originated With a Single Founder Mutation 2,000–5,000 Years Ago. Frontiers in Genetics, 2022, 13, 804924.	2.3	1
2	Genetic Analysis of Consanguineous Pakistani Families with Congenital Stationary Night Blindness. Ophthalmic Research, 2022, 65, 104-110.	1.9	1
3	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
4	Whole genome sequencing data of multiple individuals of Pakistani descent. Scientific Data, 2020, 7, 350.	5.3	1
5	Mutations in CERKL and RP1 cause retinitis pigmentosa in Pakistani families. Human Genome Variation, 2020, 7, 14.	0.7	2
6	A founder RDH5 splice site mutation leads to retinitis punctata albescens in two inbred Pakistani kindreds. Ophthalmic Genetics, 2020, 41, 7-12.	1.2	0
7	Novel mutations in identified in familial cases of primary congenital glaucoma. Molecular Vision, 2020, 26, 14-25.	1.1	4
8	Mutations in identified in families with congenital cataracts. Molecular Vision, 2020, 26, 334-344.	1.1	7
9	Autosomal recessive congenital cataracts linked to HSF4 in a consanguineous Pakistani family. PLoS ONE, 2019, 14, e0225010.	2.5	5
10	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
11	Whole genome sequencing data for two individuals of Pakistani descent. Scientific Data, 2018, 5, 180174.	5.3	2
12	Mutation in the intracellular chloride channel CLCC1 associated with autosomal recessive retinitis pigmentosa. PLoS Genetics, 2018, 14, e1007504.	3.5	25
13	A mutation in IFT43 causes non-syndromic recessive retinal degeneration. Human Molecular Genetics, 2017, 26, 4741-4751.	2.9	10
14	Molecular Genetic Analysis of Pakistani Families With Autosomal Recessive Congenital Cataracts by Homozygosity Screening ., 2017, 58, 2207.		45
15	Homozygosity Mapping and Genetic Analysis of Autosomal Recessive Retinal Dystrophies in 144 Consanguineous Pakistani Families. , 2017, 58, 2218.		34
16	Mutation in LIM2 Is Responsible for Autosomal Recessive Congenital Cataracts. PLoS ONE, 2016, 11, e0162620.	2.5	17
17	A spectrum of CYP1B1 mutations associated with primary congenital glaucoma in families of Pakistani descent. Human Genome Variation, 2016, 3, 16021.	0.7	20
18	Mutations in phosphodiesterase 6 identified in familial cases of retinitis pigmentosa. Human Genome Variation, 2016, 3, 16036.	0.7	8

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19	A missense mutation inASRGL1is involved in causing autosomal recessive retinal degeneration. Human Molecular Genetics, 2016, 25, ddw113.	2.9	16
20	Deletion at the GCNT2 Locus Causes Autosomal Recessive Congenital Cataracts. PLoS ONE, 2016, 11, e0167562.	2.5	9
21	Loss of function mutations in RP1 are responsible for retinitis pigmentosa in consanguineous familial cases. Molecular Vision, 2016, 22, 610-25.	1.1	11
22	Pathogenic mutations in TULP1 responsible for retinitis pigmentosa identified in consanguineous familial cases. Molecular Vision, 2016, 22, 797-815.	1.1	12
23	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
24	Phenotypic Variability Associated with the D226N Allele of IMPDH1. Ophthalmology, 2015, 122, 429-431.	5.2	8
25	Splice-site mutations identified in PDE6A responsible for retinitis pigmentosa in consanguineous Pakistani families. Molecular Vision, 2015, 21, 871-82.	1.1	20
26	Mutations in GRM6 identified in consanguineous Pakistani families with congenital stationary night blindness. Molecular Vision, 2015, 21, 1261-71.	1.1	7
27	exomeSuite: Whole exome sequence variant filtering tool for rapid identification of putative disease causing SNVs/indels. Genomics, 2014, 103, 169-176.	2.9	22
28	AIPL1 implicated in the pathogenesis of two cases of autosomal recessive retinal degeneration. Molecular Vision, 2014, 20, 1-14.	1.1	6
29	Perrault Syndrome Is Caused by Recessive Mutations in CLPP, Encoding a Mitochondrial ATP-Dependent Chambered Protease. American Journal of Human Genetics, 2013, 92, 605-613.	6.2	186
30	Novel mutations in RPE65 identified in consanguineous Pakistani families with retinal dystrophy. Molecular Vision, 2013, 19, 1554-64.	1.1	5
31	<i>CNAT1</i> Associated with Autosomal Recessive Congenital Stationary Night Blindness. , 2012, 53, 1353.		54
32	Association of Pathogenic Mutations in <emph type="ital">TULP1</emph> With Retinitis Pigmentosa in Consanguineous Pakistani Families. JAMA Ophthalmology, 2011, 129, 1351.	2.4	14