Muhammad Ansar

List of Publications by Year
in descending order

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119	4,087	159358	138251
papers	citations	30 h-index	58 g-index

123	123	123	6989
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Loss of Function Variants in the XPC Causes Severe Xeroderma Pigmentosum in Three Large Consanguineous Families. Klinische Padiatrie, 2022, 234, 123-129.	0.2	1
2	Phenotype Expansion for Atypical Gaucher Disease Due to Homozygous Missense PSAP Variant in a Large Consanguineous Pakistani Family. Genes, 2022, 13, 662.	1.0	3
3	Genetic Analysis of Consanguineous Pakistani Families with Congenital Stationary Night Blindness. Ophthalmic Research, 2022, 65, 104-110.	1.0	1
4	Dominant monoallelic variant in the PAK2 gene causes Knobloch syndrome type 2. Human Molecular Genetics, 2021, 31, 1-9.	1.4	6
5	Wolfram-like syndrome with bicuspid aortic valve due to a homozygous missense variant in CDK13. Journal of Human Genetics, 2021, 66, 1009-1018.	1.1	4
6	ADAMTS1, MPDZ, MVD, and SEZ6: candidate genes for autosomal recessive nonsyndromic hearing impairment. European Journal of Human Genetics, 2021, , .	1.4	6
7	A novel homozygous frameshift variant in the C3orf52 gene underlying isolated hair loss in a consanguineous family. European Journal of Dermatology, 2021, 31, 409-411.	0.3	1
8	Exome sequencing identifies novel and known mutations in families with intellectual disability. BMC Medical Genomics, 2021, 14, 211.	0.7	5
9	Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. Npj Genomic Medicine, 2021, 6, 92.	1.7	11
10	Lapachol-Induced Upregulation of Sirt1/Sirt3 is linked with Improved Skin Wound Healing in Alloxan-induced Diabetic Mice.. Iranian Journal of Pharmaceutical Research, 2021, 20, 419-430.	0.3	1
11	Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American Journal of Human Genetics, 2020, 106, 234-245.	2.6	56
12	Taurine treatment of retinal degeneration and cardiomyopathy in a consanguineous family with SLC6A6 taurine transporter deficiency. Human Molecular Genetics, 2020, 29, 618-623.	1.4	29
13	Transient central diabetes insipidus during prolonged sinus surgery: case report and literature review. Otolaryngology Case Reports, 2020, 14, 100139.	0.0	1
14	Cohen Syndrome-Associated Cataract Is Explained by VPS13B Functions in Lens Homeostasis and Is Modified by Additional Genetic Factors. , 2020, 61, 18.		3
15	Two Cases of Recessive Intellectual Disability Caused by NDST1 and METTL23 Variants. Genes, 2020, 11, 1021.	1.0	9
16	Biallelic variants in PSMB1 encoding the proteasome subunit Î²6 cause impairment of proteasome function, microcephaly, intellectual disability, developmental delay and short stature. Human Molecular Genetics, 2020, 29, 1132-1143.	1.4	30
17	Naphthoquinones from promote skin wound healing through Sirt3 regulation. Iranian Journal of Basic Medical Sciences, 2020, 23, 1139-1145.	1.0	0
18	Sequence variants in genes causing nonsyndromic hearing loss in a Pakistani cohort. Molecular Genetics & Genomic Medicine, 2019, 7, e917.	0.6	20

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19	A novel pathogenic missense variant in CNNM4 underlying Jalili syndrome: Insights from molecular dynamics simulations. Molecular Genetics & Genomic Medicine, 2019, 7, e902.	0.6	11
20	Bi-allelic Variants in IQSEC1 Cause Intellectual Disability, Developmental Delay, and Short Stature. American Journal of Human Genetics, 2019, 105, 907-920.	2.6	22
21	Bi-allelic Variants in DYNC1I2 Cause Syndromic Microcephaly with Intellectual Disability, Cerebral Malformations, and Dysmorphic Facial Features. American Journal of Human Genetics, 2019, 104, 1073-1087.	2.6	19
22	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. Human Genetics, 2019, 138, 593-600.	1.8	16
23	Wholeâ€exome sequencing revealed a nonsense mutation in STKLD1 causing nonâ€syndromic preâ€axial polydactyly type A affecting only upper limb. Clinical Genetics, 2019, 96, 134-139.	1.0	7
24	FAM92A Underlies Nonsyndromic Postaxial Polydactyly in Humans and an Abnormal Limb and Digit Skeletal Phenotype in Mice. Journal of Bone and Mineral Research, 2019, 34, 375-386.	3.1	27
25	Biallelic variants in FBXL3 cause intellectual disability, delayed motor development and short stature. Human Molecular Genetics, 2019, 28, 972-979.	1.4	17
26	Identification of CACNA1D variants associated with sinoatrial node dysfunction and deafness in additional Pakistani families reveals a clinical significance. Journal of Human Genetics, 2019, 64, 153-160.	1.1	32
27	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. Human Mutation, 2019, 40, 53-72.	1.1	48
28	A Novel Homozygous Nonsense Mutation p.Cys366* in the WNT10B Gene Underlying Split-Hand/Split Foot Malformation in a Consanguineous Pakistani Family. Frontiers in Pediatrics, 2019, 7, 526.	0.9	4
29	Mapping autosomal recessive intellectual disability: combined microarray and exome sequencing identifies 26 novel candidate genes in 192 consanguineous families. Molecular Psychiatry, 2018, 23, 973-984.	4.1	147
30	Biallelic variants in LINGO1 are associated with autosomal recessive intellectual disability, microcephaly, speech and motor delay. Genetics in Medicine, 2018, 20, 778-784.	1.1	21
31	Bi-allelic Loss-of-Function Variants in DNMBP Cause Infantile Cataracts. American Journal of Human Genetics, 2018, 103, 568-578.	2.6	29
32	Confirmation of the Role ofDHX38in the Etiology of Early-Onset Retinitis Pigmentosa. , 2018, 59, 4552.		16
33	Novel missense and 3â€²-UTR splice site variants in LHFPL5 cause autosomal recessive nonsyndromic hearing impairment. Journal of Human Genetics, 2018, 63, 1099-1107.	1.1	3
34	Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. Human Genetics, 2018, 137, 735-752.	1.8	42
35	Visual impairment and progressive phthisis bulbi caused by recessive pathogenic variant in MARK3. Human Molecular Genetics, 2018, 27, 2703-2711.	1.4	21
36	Biallelic missense variants in ZBTB11 can cause intellectual disability in humans. Human Molecular Genetics, 2018, 27, 3177-3188.	1.4	19

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37	Recessive progressive symmetric erythrokeratoderma results from a homozygous loss-of-function mutation ofKRT83and is allelic with dominant monilethrix. Journal of Medical Genetics, 2017, 54, 186-189.	1.5	11
38	Sequence variants in nine different genes underlying rare skin disorders in 10 consanguineous families. International Journal of Dermatology, 2017, 56, 1406-1413.	0.5	6
39	Novel homozygous sequence variants in the GDF5 gene underlie acromesomelic dysplasia typeâ€grebe in consanguineous families. Congenital Anomalies (discontinued), 2017, 57, 45-51.	0.3	20
40	Role of alternative phosphorylation and O-glycosylation of erythropoietinreceptor in modulating its function: an in silico study. Turkish Journal of Biology, 2017, 41, 816-825.	2.1	1
41	Segregation of Incomplete Achromatopsia and Alopecia Due to PDE6H and LPAR6 Variants in a Consanguineous Family from Pakistan. Genes, 2016, 7, 41.	1.0	8
42	Exome sequencing discloses KALRN homozygous variant as likely cause of intellectual disability and short stature in a consanguineous pedigree. Human Genomics, 2016, 10, 26.	1.4	13
43	Novel mutations in the genes <scp>TGM</scp>1 and <scp>ALOXE</scp>3 underlying autosomal recessive congenital ichthyosis. International Journal of Dermatology, 2016, 55, 524-530.	0.5	6
44	Intragenic deletion mutation in the gene desmoglein 4 underlies autosomal recessive hypotrichosis in six consanguineous families. Journal of Taibah University Medical Sciences, 2016, 11, 203-210.	0.5	0
45	Mitral regurgitation as a phenotypic manifestation of nonphotosensitive trichothiodystrophy due to a splice variant in MPLKIP. BMC Medical Genetics, 2016, 17, 13.	2.1	5
46	Autosomal-Recessive Hearing Impairment Due to Rare Missense Variants within S1PR2. American Journal of Human Genetics, 2016, 98, 331-338.	2.6	43
47	Genetic analysis of consanguineous families presenting with congenital ocular defects. Experimental Eye Research, 2016, 146, 163-171.	1.2	21
48	Inability of the most commonly used forensic genetic markers to distinguish between samples belonging to different ethnicities of Pakistan with diverse genetic background. Forensic Science International: Genetics, 2016, 22, e7-e8.	1.6	3
49	Expansion of the spectrum of ITGB6-related disorders to adolescent alopecia, dentogingival abnormalities and intellectual disability. European Journal of Human Genetics, 2016, 24, 1223-1227.	1.4	20
50	A Novel Locus for Ectodermal Dysplasia of Hair, Nail and Skin Pigmentation Anomalies Maps to Chromosome 18p11.32-p11.31. PLoS ONE, 2015, 10, e0129811.	1.1	2
51	Novel VPS13B Mutations in Three Large Pakistani Cohen Syndrome Families Suggests a Baloch Variant with Autistic-Like Features. BMC Medical Genetics, 2015, 16, 41.	2.1	23
52	Mutation of ATF6 causes autosomal recessive achromatopsia. Human Genetics, 2015, 134, 941-950.	1.8	69
53	A homozygous missense variant in type I keratinKRT25causes autosomal recessive woolly hair. Journal of Medical Genetics, 2015, 52, 676-680.	1.5	23
54	Homozygosity mapping reveals novel and known mutations in Pakistani families with inherited retinal dystrophies. Scientific Reports, 2015, 5, 9965.	1.6	28

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55	Genetic analysis of Xp22.3 micro-deletions in seventeen families segregating isolated form of X-linked ichthyosis. Journal of Dermatological Science, 2015, 80, 214-217.	1.0	3
56	Mutations in the lipaseâ€<scp>H</scp> gene causing autosomal recessive hypotrichosis and woolly hair. Australasian Journal of Dermatology, 2015, 56, e66-70.	0.4	10
57	Challenges and solutions for gene identification in the presence of familial locus heterogeneity. European Journal of Human Genetics, 2015, 23, 1207-1215.	1.4	35
58	Novel C8orf37 mutations cause retinitis pigmentosa in consanguineous families of Pakistani origin. Molecular Vision, 2015, 21, 236-43.	1.1	10
59	Biallelic Truncating Mutations in FMN2, Encoding the Actin-Regulatory Protein Formin 2, Cause Nonsyndromic Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2014, 95, 721-728.	2.6	62
60	Genetic analysis of a consanguineous Pakistani family with Leber congenital amaurosis identifies a novel mutation in GUCY2D gene. Journal of Genetics, 2014, 93, 527-530.	0.4	2
61	Adenylate cyclase 1 (ADCY1) mutations cause recessive hearing impairment in humans and defects in hair cell function and hearing in zebrafish. Human Molecular Genetics, 2014, 23, 3289-3298.	1.4	48
62	Reduced Euchromatin histone methyltransferase 1 causes developmental delay, hypotonia, and cranial abnormalities associated with increased bone gene expression in Kleefstra syndrome mice. Developmental Biology, 2014, 386, 395-407.	0.9	65
63	A missense mutation in the PISA domain of HsSAS-6 causes autosomal recessive primary microcephaly in a large consanguineous Pakistani family. Human Molecular Genetics, 2014, 23, 5940-5949.	1.4	63
64	Application of Short Tandem Repeat markers in diagnosis of chromosomal aneuploidies and forensic DNA investigation in Pakistan. Gene, 2014, 548, 217-222.	1.0	8
65	A novel WDR62 mutation causes primary microcephaly in a Pakistani family. Molecular Biology Reports, 2013, 40, 591-595.	1.0	16
66	Homozygous and heterozygous disruptions of ANK3: at the crossroads of neurodevelopmental and psychiatric disorders. Human Molecular Genetics, 2013, 22, 1960-1970.	1.4	137
67	Stable Huh-7 cell lines expressing non-structural proteins of genotype 1a of hepatitis C virus. Journal of Virological Methods, 2013, 189, 65-69.	1.0	7
68	Novel mutations in the geneHOXC13underlying pure hair and nail ectodermal dysplasia in consanguineous families. British Journal of Dermatology, 2013, 169, 478-480.	1.4	15
69	Mutations in the gene phospholipase C, delta-1 (PLCD1) underlying hereditary leukonychia. European Journal of Dermatology, 2012, 22, 736-739.	0.3	15
70	Alterations of the CIB2 calcium- and integrin-binding protein cause Usher syndrome type 1J and nonsyndromic deafness DFNB48. Nature Genetics, 2012, 44, 1265-1271.	9.4	217
71	Mutation in NSUN2, which Encodes an RNA Methyltransferase, Causes Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2012, 90, 856-863.	2.6	189
72	Novel TMPRSS3 variants in Pakistani families with autosomal recessive nonâ€syndromic hearing impairment. Clinical Genetics, 2012, 82, 56-63.	1.0	14

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73	A novel homozygous missense mutation in WNT10B in familial splitâ€hand/foot malformation. Clinical Genetics, 2012, 82, 48-55.	1.0	38
74	Novel CLDN14 mutations in Pakistani families with autosomal recessive nonâ€syndromic hearing loss. American Journal of Medical Genetics, Part A, 2012, 158A, 315-321.	0.7	28
75	Genetic mapping of an autosomal recessive postaxial polydactyly type A to chromosome 13q13.3â€“q21.2 and screening of the candidate genes. Human Genetics, 2012, 131, 415-422.	1.8	44
76	Mapping the NPHP-JBTS-MKS Protein Network Reveals Ciliopathy Disease Genes and Pathways. Cell, 2011, 145, 513-528.	13.5	531
77	Post-transcriptional inhibition of hepatitis C virus replication through small interference RNA. Virology Journal, 2011, 8, 112.	1.4	12
78	Inhibition of full length Hepatitis C Virus particles of 1a genotype through small interference RNA. Virology Journal, 2011, 8, 203.	1.4	17
79	NS4A protein as a marker of HCV history suggests that different HCV genotypes originally evolved from genotype 1b. Virology Journal, 2011, 8, 317.	1.4	9
80	A Novel Deletion Mutation in Proteoglycan-4 Underlies Camptodactyly-Arthropathy-Coxa-Vara-Pericarditis Syndrome in a Consanguineous Pakistani Family. Archives of Medical Research, 2011, 42, 110-114.	1.5	16
81	Functional Null Mutations of MSRB3 Encoding Methionine Sulfoxide Reductase Are Associated with Human Deafness DFNB74. American Journal of Human Genetics, 2011, 88, 19-29.	2.6	107
82	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	2.6	108
83	Mutations in the Alpha 1,2-Mannosidase Gene, MAN1B1, Cause Autosomal-Recessive Intellectual Disability. American Journal of Human Genetics, 2011, 89, 176-182.	2.6	73
84	DFNB89, a novel autosomal recessive nonsyndromic hearing impairment locus on chromosome 16q21-q23.2. Human Genetics, 2011, 129, 379-385.	1.8	11
85	Novel mutations in the keratin-74 (KRT74) gene underlie autosomal dominant woolly hair/hypotrichosis in Pakistani families. Human Genetics, 2011, 129, 419-424.	1.8	29
86	Genetic analysis of four Pakistani families with achromatopsia and a novel S4 motif mutation of CNGA3. Japanese Journal of Ophthalmology, 2011, 55, 676-680.	0.9	16
87	A novel deletion mutation in the TUSC3 gene in a consanguineous Pakistani family with autosomal recessive nonsyndromic intellectual disability. BMC Medical Genetics, 2011, 12, 56.	2.1	33
88	Mutations in WDR62 gene in Pakistani families with autosomal recessive primary microcephaly. BMC Neurology, 2011, 11, 119.	0.8	26
89	A Novel ESRRB Deletion Is a Rare Cause of Autosomal Recessive Nonsyndromic Hearing Impairment among Pakistani Families. Genetics Research International, 2011, 2011, 1-4.	2.0	8
90	Novel Autosomal Recessive Nonsyndromic Hearing Impairment Locus DFNB90 Maps to 7p22.1-p15.3. Human Heredity, 2011, 71, 106-112.	0.4	4

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91	A new autosomal recessive nonsyndromic hearing impairment locus DFNB96 on chromosome 1p36.31â€“p36.13. Journal of Human Genetics, 2011, 56, 866-868.	1.1	5
92	Homozygosity Mapping Reveals Mutations of GRXCR1 as a Cause of Autosomal-Recessive Nonsyndromic Hearing Impairment. American Journal of Human Genetics, 2010, 86, 138-147.	2.6	58
93	Genetic mapping of a novel hypotrichosis locus to chromosome 7p21.3â€“p22.3 in a Pakistani family and screening of the candidate genes. Human Genetics, 2010, 128, 213-220.	1.8	13
94	RNAi as a new therapeutic strategy against HCV. Biotechnology Advances, 2010, 28, 27-34.	6.0	30
95	WDR62 is associated with the spindle pole and is mutated in human microcephaly. Nature Genetics, 2010, 42, 1010-1014.	9.4	255
96	Mapping of three novel loci for nonâ€syndromic autosomal recessive mental retardation (NSâ€ARMR) in consanguineous families from Pakistan. Clinical Genetics, 2010, 78, 478-483.	1.0	17
97	Mutation Analysis of the ASPM Gene in 18 Pakistani Families With Autosomal Recessive Primary Microcephaly. Journal of Child Neurology, 2010, 25, 715-720.	0.7	18
98	Mutations in Lipase H Gene Underlie Autosomal Recessive Hypotrichosis in Five Pakistani Families. Acta Dermato-Venereologica, 2010, 90, 93-94.	0.6	9
99	Recurrent mutations in functionally-related EDA and EDAR genes underlie X-linked isolated hypodontia and autosomal recessive hypohidrotic ectodermal dysplasia. Archives of Dermatological Research, 2009, 301, 625-629.	1.1	15
100	Noncoding Mutations of HGF Are Associated with Nonsyndromic Hearing Loss, DFNB39. American Journal of Human Genetics, 2009, 85, 25-39.	2.6	119
101	Novel missense mutations in lipase H (LIPH) gene causing autosomal recessive hypotrichosis (LAH2). Journal of Dermatological Science, 2009, 54, 12-16.	1.0	16
102	Novel autosomal recessive non-syndromic hearing impairment locus (DFNB71) maps to chromosome 8p22â€“21.3. Journal of Human Genetics, 2009, 54, 141-144.	1.1	6
103	A novel insertion mutation in the cartilage-derived morphogenetic protein-1 (CDMP1) gene underlies Grebe-type chondrodysplasia in a consanguineous Pakistani family. BMC Medical Genetics, 2008, 9, 102.	2.1	32
104	Intragenic deletions in thedystrophingene in 211 Pakistani Duchenne muscular dystrophy patients. Pediatrics International, 2008, 50, 162-166.	0.2	20
105	Mutations of ESRRB Encoding Estrogen-Related Receptor Beta Cause Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB35. American Journal of Human Genetics, 2008, 82, 125-138.	2.6	127
106	Genetic heterogeneity of synpolydactyly: a novel locus SPD3 maps to chromosome 14q11.2-q12. Clinical Genetics, 2006, 69, 518-524.	1.0	23
107	Recurrent intragenic deletion mutation in desmoglein 4 gene underlies autosomal recessive hypotrichosis in two Pakistani families of Balochi and Sindhi origins. Archives of Dermatological Research, 2006, 298, 135-137.	1.1	20
108	Genetic studies of autosomal recessive primary microcephaly in 33 Pakistani families: novel sequence variants in ASPM gene. Neurogenetics, 2006, 7, 105-110.	0.7	55

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109	A novel autosomal recessive non-syndromic hearing impairment locus (DFNB47) maps to chromosome 2p25.1-p24.3. Human Genetics, 2006, 118, 605-610.	1.8	9
110	Novel sequence variants in theTMC1 gene in Pakistani families with autosomal recessive hearing impairment. Human Mutation, 2005, 26, 396-396.	1.1	52
111	A novel autosomal recessive nonsyndromic hearing impairment locus (DFNB42) maps to chromosome 3q13.31-q22.3. American Journal of Medical Genetics, Part A, 2005, 133A, 18-22.	0.7	20
112	Mapping of a novel autosomal recessive nonsyndromic deafness locus (DFNB46) to chromosome 18p11.32-p11.31. American Journal of Medical Genetics, Part A, 2005, 133A, 23-26.	0.7	3
113	DFNB44, a Novel Autosomal Recessive Non-Syndromic Hearing Impairment Locus, Maps to Chromosome 7p14.1-q11.22. Human Heredity, 2004, 57, 195-199.	0.4	11
114	Localization of a novel locus for hereditary nail dysplasia to chromosome 17q25.1-17q25.3. Clinical Genetics, 2004, 66, 73-78.	1.0	5
115	A Recurrent Intragenic Deletion Mutation in DSG4 Gene in Three Pakistani Families with Autosomal Recessive Hypotrichosis. Journal of Investigative Dermatology, 2004, 123, 247-248.	0.3	41
116	A novel autosomal recessive non-syndromic deafness locus (DFNB35) maps to 14q24.1â€“14q24.3 in large consanguineous kindred from Pakistan. European Journal of Human Genetics, 2003, 11, 77-80.	1.4	32
117	A locus for hereditary hypotrichosis localized to human chromosome 18q21.1. European Journal of Human Genetics, 2003, 11, 623-628.	1.4	30
118	DFNB39, a recessive form of sensorineural hearing impairment, maps to chromosome 7q11.22â€“q21.12. European Journal of Human Genetics, 2003, 11, 812-815.	1.4	14
119	Localization of A Novel Autosomal Recessive Non-Syndromic Hearing Impairment Locus (DFNB38) to 6q26-q27 in a Consanguineous Kindred from Pakistan. Human Heredity, 2003, 55, 71-74.	0.4	13