Wasim Ahmad

List of Publications by Year
in descending order

162	3,099	185998 28	253896 43
papers	citations	h-index	g-index

164	164	164	4045
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Desmoglein 4 in Hair Follicle Differentiation and Epidermal Adhesion. Cell, 2003, 113, 249-260.	13.5	301
2	Mutations in TBC1D24, a Gene Associated With Epilepsy, Also Cause Nonsyndromic Deafness DFNB86. American Journal of Human Genetics, 2014, 94, 144-152.	2.6	72
3	Mutational Spectrum ofMYO15Aand the Molecular Mechanisms of DFNB3 Human Deafness. Human Mutation, 2016, 37, 991-1003.	1.1	67
4	Clinical Genetics of Polydactyly: An Updated Review. Frontiers in Genetics, 2018, 9, 447.	1.1	66
5	A mutation in the lipase H (LIPH) gene underlie autosomal recessive hypotrichosis. Human Genetics, 2007, 121, 319-325.	1.8	61
6	A novel missense mutation in MSX1 underlies autosomal recessive oligodontia with associated dental anomalies in Pakistani families. Journal of Human Genetics, 2006, 51, 872-878.	1.1	59
7	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
8	Exome sequencing revealed a splice site variant in the IQCE gene underlying post-axial polydactyly type A restricted to lower limb. European Journal of Human Genetics, 2017, 25, 960-965.	1.4	53
9	Whole exome sequencing identified a novel zinc-finger geneZNF141associated with autosomal recessive postaxial polydactyly type A. Journal of Medical Genetics, 2013, 50, 47-53.	1.5	51
10	GLI1 inactivation is associated with developmental phenotypes overlapping with Ellisâ€“van Creveld syndrome. Human Molecular Genetics, 2017, 26, 4556-4571.	1.4	50
11	A novel deletion mutation in CENPJ gene in a Pakistani family with autosomal recessive primary microcephaly. Journal of Human Genetics, 2006, 51, 760-764.	1.1	48
12	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
13	Global genetic insight contributed by consanguineous Pakistani families segregating hearing loss. Human Mutation, 2019, 40, 53-72.	1.1	48
14	Splice-site mutations in the TRIC gene underlie autosomal recessive nonsyndromic hearing impairment in Pakistani families. Journal of Human Genetics, 2008, 53, 101-105.	1.1	45
15	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
16	Mutational and phenotypic spectra of KCNE1 deficiency in Jervell and Langeâ€Nielsen Syndrome and Romanoâ€Ward Syndrome. Human Mutation, 2019, 40, 162-176.	1.1	44
17	Autosomal-Recessive Hearing Impairment Due to Rare Missense Variants within S1PR2. American Journal of Human Genetics, 2016, 98, 331-338.	2.6	43
18	Novel candidate genes and variants underlying autosomal recessive neurodevelopmental disorders with intellectual disability. Human Genetics, 2018, 137, 735-752.	1.8	42

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19	In silico analysis of SIGMAR1 variant (rs4879809) segregating in a consanguineous Pakistani family showing amyotrophic lateral sclerosis without frontotemporal lobar dementia. Neurogenetics, 2015, 16, 299-306.	0.7	39
20	A novel homozygous missense mutation in WNT10B in familial splitâ€hand/foot malformation. Clinical Genetics, 2012, 82, 48-55.	1.0	38
21	Novel mutations in G protein-coupled receptor gene (P2RY5) in families with autosomal recessive hypotrichosis (LAH3). Human Genetics, 2008, 123, 515-519.	1.8	37
22	Novel mutations in natriuretic peptide receptor-2 gene underlie acromesomelic dysplasia, type maroteaux. BMC Medical Genetics, 2012, 13, 44.	2.1	37
23	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
24	Exome sequencing reveals a novel homozygous splice site variant in the WNT1 gene underlying osteogenesis imperfecta type 3. Pediatric Research, 2017, 82, 753-758.	1.1	34
25	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
26	Homozygous sequence variants in the FKBP10 gene underlie osteogenesis imperfecta in consanguineous families. Journal of Human Genetics, 2016, 61, 207-213.	1.1	32
27	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
28	Homozygous XYLT2 variants as a cause of spondyloocular syndrome. Clinical Genetics, 2018, 93, 913-918.	1.0	31
29	Association analysis of GWAS and candidate gene loci in a Pakistani population with psoriasis. Molecular Immunology, 2015, 64, 190-194.	1.0	30
30	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
31	A novel deletion mutation in LIPH gene causes autosomal recessive hypotrichosis (LAH2). Clinical Genetics, 2008, 74, 184-188.	1.0	28
32	A novel recessive mutation in the gene ELOVL4 causes a neuro-ichthyotic disorder with variable expressivity. BMC Medical Genetics, 2014, 15, 25.	2.1	28
33	Mutation in PVRL4 gene encoding nectin-4 underlies ectodermal-dysplasia-syndactyly syndrome (EDSS1). Journal of Human Genetics, 2011, 56, 352-357.	1.1	27
34	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
35	Novel mutations in the EDAR gene in two Pakistani consanguineous families with autosomal recessive hypohidrotic ectodermal dysplasia. British Journal of Dermatology, 2005, 153, 46-50.	1.4	26
36	NOVEL PROTEIN-TRUNCATING MUTATIONS IN THEASPMGENE IN FAMILIES WITH AUTOSOMAL RECESSIVE PRIMARY MICROCEPHALY. Journal of Neurogenetics, 2007, 21, 153-163.	0.6	26

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37	Biallelic variants in four genes underlying recessive osteogenesis imperfecta. European Journal of Medical Genetics, 2020, 63, 103954.	0.7	26
38	A novel homozygous sequence variant in GLI1 underlies first case of autosomal recessive preâ€axial polydactyly. Clinical Genetics, 2019, 95, 540-541.	1.0	24
39	Ectodermal dysplasia of hair and nail type: mapping of a novel locus to chromosome 17p12-q21.2. British Journal of Dermatology, 2006, 155, 1184-1190.	1.4	23
40	Novel homozygous mutations in the genes ARL6 and BBS10 underlying Bardetâ€“Biedl syndrome. Gene, 2013, 515, 84-88.	1.0	23
41	A homozygous missense variant in type I keratinKRT25causes autosomal recessive woolly hair. Journal of Medical Genetics, 2015, 52, 676-680.	1.5	23
42	A novel deletion mutation in theDSG4gene underlies autosomal recessive hypotrichosis with variable phenotype in two unrelated consanguineous families. Clinical and Experimental Dermatology, 2015, 40, 78-84.	0.6	23
43	Exome sequencing revealed a novel biallelic deletion in theDCAF17gene underlying Woodhouse Sakati syndrome. Clinical Genetics, 2016, 90, 263-269.	1.0	23
44	Autosomal Dominantly Inherited GREB1L Variants in Individuals with Profound Sensorineural Hearing Impairment. Genes, 2020, 11, 687.	1.0	23
45	A novel splice site mutation in gene C2orf37 underlying Woodhouseâ€“Sakati syndrome (WSS) in a consanguineous family of Pakistani origin. Gene, 2011, 490, 26-31.	1.0	22
46	In Silico Analysis of Missense Mutations in LPAR6 Reveals Abnormal Phospholipid Signaling Pathway Leading to Hypotrichosis. PLoS ONE, 2014, 9, e104756.	1.1	22
47	A Novel Heterozygous Intragenic Sequence Variant in DLX6 Probably Underlies First Case of Autosomal Dominant Split-Hand/Foot Malformation Type 1. Molecular Syndromology, 2017, 8, 79-84.	0.3	22
48	A syndromic form of autosomal recessive congenital microcephaly (Jawad syndrome) maps to chromosome 18p11.22â€“q11.2. Human Genetics, 2008, 123, 77-82.	1.8	21
49	Novel homozygous mutations in the WNT10B gene underlying autosomal recessive split hand/foot malformation in three consanguineous families. Gene, 2014, 534, 265-271.	1.0	21
50	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
51	Intragenic deletions in thedystrophingene in 211 Pakistani Duchenne muscular dystrophy patients. Pediatrics International, 2008, 50, 162-166.	0.2	20
52	Genetics of human isolated hereditary nail disorders. British Journal of Dermatology, 2015, 173, 922-929.	1.4	20
53	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
54	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

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55	First direct evidence of involvement of a homozygous lossâ€•ofâ€•function variant in the EPS15L1 gene underlying splitâ€•hand/splitâ€•foot malformation. Clinical Genetics, 2018, 93, 699-702.	1.0	20
56	Sequence variants in genes causing nonsyndromic hearing loss in a Pakistani cohort. Molecular Genetics & Genomic Medicine, 2019, 7, e917.	0.6	20
57	Localization of a novel autosomal recessive hypotrichosis locus (LAH3) to chromosome 13q14.11-q21.32. Clinical Genetics, 2007, 72, 23-29.	1.0	19
58	Homozygous Sequence Variants in the NPR2 Gene Underlying Acromesomelic Dysplasia Maroteaux Type (AMDM) in Consanguineous Families. Annals of Human Genetics, 2015, 79, 238-244.	0.3	19
59	Sequence variants in four genes underlying Bardet-Biedl syndrome in consanguineous families. Molecular Vision, 2017, 23, 482-494.	1.1	19
60	Mutations in the LPAR6 and LIPH genes underlie autosomal recessive hypotrichosis/woolly hair in 17 consanguineous families from Pakistan. Clinical and Experimental Dermatology, 2011, 36, 652-654.	0.6	18
61	A Novel Locus Harbouring a Functional CD164 Nonsense Mutation Identified in a Large Danish Family with Nonsyndromic Hearing Impairment. PLoS Genetics, 2015, 11, e1005386.	1.5	18
62	Genetics of human isolated acromesomelic dysplasia. European Journal of Medical Genetics, 2016, 59, 198-203.	0.7	18
63	Homozygous sequence variants in the WNT10B gene underlie split hand/foot malformation. Genetics and Molecular Biology, 2018, 41, 1-8.	0.6	18
64	A variant in LMX1A causes autosomal recessive severe-to-profound hearing impairment. Human Genetics, 2018, 137, 471-478.	1.8	18
65	Novel digenic inheritance of PCDH15 and USH1G underlies profound non-syndromic hearing impairment. BMC Medical Genetics, 2018, 19, 122.	2.1	18
66	Localization of a novel locus for alopecia with mental retardation syndrome to chromosome 3q26.33â€“q27.3. Human Genetics, 2006, 118, 665-667.	1.8	17
67	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
68	Confirmation of the Role ofDHX38in the Etiology of Early-Onset Retinitis Pigmentosa. , 2018, 59, 4552.		16
69	Variants in KIAA0825 underlie autosomal recessive postaxial polydactyly. Human Genetics, 2019, 138, 593-600.	1.8	16
70	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
71	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
72	A Novel Homozygous Nonsense Mutation in the PVRL4 Gene and Expansion of Clinical Spectrum of EDSS1. Annals of Human Genetics, 2015, 79, 92-98.	0.3	15

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73	Exome sequencing revealed a novel lossâ€ofâ€function variant in the GLI3 transcriptional activator 2 domain underlies nonsyndromic postaxial polydactyly. Molecular Genetics & Genomic Medicine, 2019, 7, e00627.	0.6	15
74	A novel splice-acceptor site mutation in CDH3 gene in a consanguineous family exhibiting hypotrichosis with juvenile macular dystrophy. Archives of Dermatological Research, 2010, 302, 701-703.	1.1	14
75	Digenic inheritance of an autosomal recessive hypotrichosis in two consanguineous pedigrees. Clinical Genetics, 2011, 79, 273-281.	1.0	14
76	A novel missense variant in the PNPLA1 gene underlies congenital ichthyosis in three consanguineous families. Journal of the European Academy of Dermatology and Venereology, 2016, 30, e210-e213.	1.3	14
77	Molecular and in silico analyses validates pathogenicity of homozygous mutations in the NPR2 gene underlying variable phenotypes of Acromesomelic dysplasia, type Maroteaux. International Journal of Biochemistry and Cell Biology, 2018, 102, 76-86.	1.2	14
78	Atrichia with papular lesions in two Pakistani consanguineous families resulting from mutations in the human hairless gene. Archives of Dermatological Research, 2005, 297, 226-230.	1.1	13
79	A novel locus for alopecia with mental retardation syndrome (APMR2) maps to chromosome 3q26.2-q26.31. Clinical Genetics, 2006, 70, 233-239.	1.0	13
80	Mapping of a Gene for Alopecia with Mental Retardation Syndrome (APMR3) on Chromosome 18q11.2-q12.2. Annals of Human Genetics, 2007, 71, 570-577.	0.3	13
81	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
82	Exome sequencing revealed a novel splice site variant in the <scp>ALX1</scp> gene underlying frontonasal dysplasia. Clinical Genetics, 2017, 91, 494-498.	1.0	13
83	Ellisâ€“van Creveld syndrome and profound deafness resulted by sequence variants in the EVCÂ/ÂEVC2Âand TMC1Âgenes. Journal of Genetics, 2017, 96, 1005-1014.	0.4	13
84	A novel homozygous variant in BMPR1B underlies acromesomelic dysplasia Hunterâ€“Thompson type. Annals of Human Genetics, 2018, 82, 129-134.	0.3	13
85	A homozygous missense mutation in SLC25A16 associated with autosomal recessive isolated fingernail dysplasia in a Pakistani family. British Journal of Dermatology, 2018, 178, 556-558.	1.4	12
86	Homozygous variants in the HEXB and MBOAT7 genes underlie neurological diseases in consanguineous families. BMC Medical Genetics, 2019, 20, 199.	2.1	12
87	Mapping of a novel autosomal recessive hypotrichosis locus on chromosome 10q11.23â€“22.3. Human Genetics, 2010, 127, 395-401.	1.8	11
88	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
89	Novel sequence variants in the MKKS gene cause Bardetâ€Biedl syndrome with intraâ€and interâ€familial variable phenotypes. Congenital Anomalies (discontinued), 2018, 58, 173-175.	0.3	11
90	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

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91	A loss-of-function variant in DNA mismatch repair gene MLH3 underlies severe oligozoospermia. Journal of Human Genetics, 2021, 66, 725-730.	1.1	11
92	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
93	Genetics of human isolated hereditary hair loss disorders. Clinical Genetics, 2015, 88, 203-212.	1.0	10
94	A novel homozygous missense mutation in BHLHA9 causes mesoaxial synostotic syndactyly with phalangeal reduction in a Pakistani family. Human Genome Variation, 2017, 4, 17054.	0.4	10
95	BBS5 and INPP5E mutations associated with ciliopathy disorders in families from Pakistan. Annals of Human Genetics, 2019, 83, 477-482.	0.3	10
96	Biallelic variants in TRAPPC10 cause a microcephalic TRAPPopathy disorder in humans and mice. PLoS Genetics, 2022, 18, e1010114.	1.5	10
97	Hypomorphic MKS1 mutation in a Pakistani family with mild Joubert syndrome and atypical features: Expanding the phenotypic spectrum of MKS1â€related ciliopathies. American Journal of Medical Genetics, Part A, 2016, 170, 3289-3293.	0.7	9
98	A disease-causing novel missense mutation in the ST14 gene underlies autosomal recessive ichthyosis with hypotrichosis syndrome in a consanguineous family. European Journal of Dermatology, 2018, 28, 209-216.	0.3	9
99	A novel insertion and deletion mutation in the BHLHA9 underlies polydactyly and mesoaxial synostotic syndactyly with phalangeal reduction. European Journal of Medical Genetics, 2019, 62, 278-281.	0.7	9
100	Î²2 Integrin Gene (ITGB2) mutation spectra in Pakistani families with leukocyte adhesion deficiency type 1 (LAD1). Immunobiology, 2020, 225, 151938.	0.8	9
101	A novel missense mutation in theTRPS1gene underlies trichorhinophalangeal syndrome type III. British Journal of Dermatology, 2008, 159, 476-478.	1.4	8
102	Novel heterozygous frameshift mutation in distalâ€less homeobox 5 underlies isolated split hand/foot malformation type 1. Pediatrics International, 2016, 58, 1348-1350.	0.2	8
103	A novel homozygous variant in the SMOC1 gene underlying Waardenburg anophthalmia syndrome. Ophthalmic Genetics, 2017, 38, 335-339.	0.5	8
104	HomozygousSLCO2A1translation initiation codon mutation in a Pakistani family with recessive isolated congenital nail clubbing. British Journal of Dermatology, 2017, 177, 546-548.	1.4	8
105	Identification of novel L2HGDH mutation in a large consanguineous Pakistani family- a case report. BMC Medical Genetics, 2018, 19, 25.	2.1	8
106	Identification of novel LEPR mutations in Pakistani families with morbid childhood obesity. BMC Medical Genetics, 2018, 19, 199.	2.1	8
107	Novel nonsense variants in SLURP1 and DSG1 cause palmoplantar keratoderma in Pakistani families. BMC Medical Genetics, 2019, 20, 145.	2.1	8
108	Variants inGLI3Cause Greig Cephalopolysyndactyly Syndrome. Genetic Testing and Molecular Biomarkers, 2019, 23, 744-750.	0.3	8

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109	First evidence of involvement of TBC1D25 in causing human male infertility. European Journal of Medical Genetics, 2021, 64, 104142.	0.7	8
110	Further confirmation of the association of SLC12A2 with non-syndromic autosomal-dominant hearing impairment. Journal of Human Genetics, 2021, 66, 1169-1175.	1.1	8
111	Disease causing homozygous variants in the human hairless gene. International Journal of Dermatology, 2016, 55, 977-981.	0.5	7
112	Novel homozygous sequence variants in the CDH3 gene encoding P-cadherin underlying hypotrichosis with juvenile macular dystrophy in consanguineous families. European Journal of Dermatology, 2016, 26, 610-612.	0.3	7
113	A novel homozygous missense variant inNECTIN4 (PVRL4)causing ectodermal dysplasia cutaneous syndactyly syndrome. Annals of Human Genetics, 2018, 82, 232-238.	0.3	7
114	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
115	Novel heterozygous sequence variant in the GLI1 underlies postaxial polydactyly. Congenital Anomalies (discontinued), 2020, 60, 115-119.	0.3	7
116	Identification of a novel biallelic missense variant in the KIAA0825 underlies postaxial polydactyly type A. Genomics, 2020, 112, 2729-2733.	1.3	7
117	Ectodermal dysplasia-cutaneous syndactyly syndrome maps to chromosome 7p21.1-p14.3. Human Genetics, 2009, 125, 421-429.	1.8	6
118	The angiotensin-converting enzyme gene insertion polymorphism: a higher risk for psoriasis in male patients. British Journal of Dermatology, 2016, 175, 824-826.	1.4	6
119	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
120	Autosomal recessive transmission of a rare HOXC13 variant causes pure hair and nail ectodermal dysplasia. Clinical and Experimental Dermatology, 2017, 42, 585-589.	0.6	6
121	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
122	Sequence variants in <scp>GDF</scp>5 and <scp>TRPS</scp>1 underlie brachydactyly and trichoâ€rhinoâ€phalangeal syndrome type III. Pediatrics International, 2018, 60, 304-306.	0.2	6
123	Novel sequence variants in the LIPH and LPAR6 genes underlies autosomal recessive woolly hair/hypotrichosis in consanguineous families. Congenital Anomalies (discontinued), 2018, 58, 24-28.	0.3	6
124	Exome sequencing revealed a novel nonsense variant in ALX3 gene underlying frontorhiny. Journal of Human Genetics, 2018, 63, 97-100.	1.1	6
125	Woodhouseâ€“Sakati syndrome in a family is associated with a homozygous start loss mutation in the <scp>DCAF</scp> 17 gene. Clinical and Experimental Dermatology, 2020, 45, 159-164.	0.6	6
126	Further evidence of involvement of TMEM132E in autosomal recessive nonsyndromic hearing impairment. Journal of Human Genetics, 2020, 65, 187-192.	1.1	6

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127	ADAMTS1, MPDZ, MVD, and SEZ6: candidate genes for autosomal recessive nonsyndromic hearing impairment. European Journal of Human Genetics, 2021, , .	1.4	6
128	Localization of a novel locus for hereditary nail dysplasia to chromosome 17q25.1-17q25.3. Clinical Genetics, 2004, 66, 73-78.	1.0	5
129	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
130	Novel autosomal recessive LAMA3 and PLEC variants underlie junctional epidermolysis bullosa generalized intermediate and epidermolysis bullosa simplex with muscular dystrophy in two consanguineous families. Clinical and Experimental Dermatology, 2018, 43, 752-755.	0.6	5
131	A homozygous nonsense variant in DYM underlies Dyggveâ€“Melchiorâ€“Clausen syndrome associated with ectodermal features. Molecular Biology Reports, 2020, 47, 7083-7088.	1.0	5
132	A Novel Missense Variant in theALX4Gene Underlies Mild to Severe Frontonasal Dysplasia in a Consanguineous Family. Genetic Testing and Molecular Biomarkers, 2020, 24, 217-223.	0.3	5
133	Novel variants in the LRP4 underlying Cenani-Lenz Syndactyly syndrome. Journal of Human Genetics, 2021, , .	1.1	5
134	Linkage mapping of a nonspecific form of X-linked mental retardation (MRX53) in a large Pakistani family. American Journal of Medical Genetics Part A, 2001, 100, 62-65.	2.4	4
135	Frameshift Sequence Variants in the Human Lipaseâ€H Gene Causing Hypotrichosis. Pediatric Dermatology, 2016, 33, e40-2.	0.5	4
136	A novel frameshift variant in BHLHA9 underlies mesoaxial synostotic syndactyly associated with postaxial polydactyly. European Journal of Medical Genetics, 2019, 62, 103688.	0.7	4
137	A novel missense variant in the BBS7 gene underlying Bardet-Biedl syndrome in a consanguineous Pakistani family. Clinical Dysmorphology, 2020, 29, 17-23.	0.1	4
138	Sequence Variants in the WNT10B and TP63 Genes Underlying Isolated Split-Hand/Split-Foot Malformation. Genetic Testing and Molecular Biomarkers, 2020, 24, 600-607.	0.3	4
139	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
140	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
141	Homozygous variants of EDAR underlying hypohidrotic ectodermal dysplasia in three consanguineous families. European Journal of Dermatology, 2020, 30, 408-416.	0.3	4
142	A Novel Locus for Ectodermal Dysplasia of Hairs, Nails and Teeth Type Maps to Chromosome 18q22.1â€“22.3. Annals of Human Genetics, 2008, 72, 19-25.	0.3	3
143	A novel homozygous variant in the dsp gene underlies the first case of non-syndromic form of alopecia. Archives of Dermatological Research, 2015, 307, 793-801.	1.1	3
144	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

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145	A novel homozygous mutation disrupting the initiation codon in the SLURP1 gene underlies mal de Meleda in a consanguineous family. Clinical and Experimental Dermatology, 2016, 41, 675-679.	0.6	3
146	Identification of a Novel NonsenseASPMMutation in a Large Consanguineous Pakistani Family Using Targeted Next-Generation Sequencing. Genetic Testing and Molecular Biomarkers, 2018, 22, 159-164.	0.3	3
147	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
148	Identification and Computational Analysis of Novel TYR and SLC45A2 Gene Mutations in Pakistani Families With Identical Non-syndromic Oculocutaneous Albinism. Frontiers in Genetics, 2020, 11, 749.	1.1	3
149	Greig Cephalopolysyndactyly Syndrome: Phenotypic Variability Associated with Variants in Two Different Domains of GLI3. Klinische Padiatrie, 2021, 233, 53-58.	0.2	3
150	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
151	A novel nonsense variant in EXOC8 underlies a neurodevelopmental disorder. Neurogenetics, 2022, 23, 203-212.	0.7	3
152	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
153	Sequence variants in theEDARgene causing hypohidrotic ectodermal dysplasia. Congenital Anomalies (discontinued), 2019, 59, 145-147.	0.3	2
154	Sequence variants in three genes underlying leukodystrophy in Pakistani families. International Journal of Developmental Neuroscience, 2020, 80, 380-388.	0.7	2
155	A Frameshift Variant in <i>KIAA0825</i> Causes Postaxial Polydactyly. Molecular Syndromology, 2021, 12, 20-24.	0.3	2
156	Screening, diagnosis and genetic study of breast cancer patients in Pakistan. Pakistan Journal of Medical Sciences, 2020, 36, 16-20.	0.3	2
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