Sulman Basit

List of Publications by Year
in descending order

122 papers	2,222 citations	361296 20 h-index	276775 41 g-index
124 all docs	124 docs citations	124 times ranked	3823 citing authors

#	Article	IF	Citations
1	A Two-Base Pair Deletion in IQ Repeats in ASPM Underlies Microcephaly in a Pakistani Family. Genetic Testing and Molecular Biomarkers, 2022, 26, 37-42.	0.3	4
2	Association between 17q21 variants and asthma predisposition in Pashtun population from Pakistan. Journal of Asthma, 2022, , 1-13.	0.9	3
3	Exome Sequencing Revealed the First Intragenic Deletion in ABCA5 Underlying Autosomal Recessive Hypertrichosis. Clinical and Experimental Dermatology, 2022, , .	0.6	2
4	Ultrastructure abnormalities of collagen and elastin in Arab patients with arterial tortuosity syndrome. Journal of Cutaneous Pathology, 2022, , .	0.7	1
5	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
6	A homozygous missense variant in the MLC1 gene underlies megalencephalic leukoencephalopathy with subcortical cysts in large kindred: Heterozygous carriers show seizure and mild motor function deterioration. American Journal of Medical Genetics, Part A, 2022, 188, 1075-1082.	0.7	1
7	Association of SORD mutation with autosomal recessive asymmetric distal hereditary motor neuropathy. BMC Medical Genomics, 2022, 15, 88.	0.7	2
8	A novel nonsense variant in EXOC8 underlies a neurodevelopmental disorder. Neurogenetics, 2022, 23, 203-212.	0.7	3
9	A novel homozygous frameshift mutation in the DCC gene in a Pakistani family with autosomal recessive horizontal gaze palsy with progressive scoliosisâ€2 with impaired intellectual development. American Journal of Medical Genetics, Part A, 2021, 185, 355-361.	0.7	3
10	A novel frameshift mutation in the ITGB3 gene leading to Glanzmann's thrombasthenia in a Saudi Arabian family. Hematology/ Oncology and Stem Cell Therapy, 2021, , .	0.6	3
11	First evidence of involvement of TBC1D25 in causing human male infertility. European Journal of Medical Genetics, 2021, 64, 104142.	0.7	8
12	Frameshift variant in MITF gene in a large family with Waardenburg syndrome type II and a co-segregation of a C2orf74 variant. PLoS ONE, 2021, 16, e0246607.	1.1	1
13	Novel Homozygous Mutations in the Genes TGM1, SULT2B1, SPINK5 and FLG in Four Families Underlying Congenital Ichthyosis. Genes, 2021, 12, 373.	1.0	6
14	An intrafamilial phenotypic variability in <scp>Ellisâ€Van</scp> Creveld syndrome due to a novel 27â€‰bps deletion mutation. American Journal of Medical Genetics, Part A, 2021, 185, 2888-2894.	0.7	1
15	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
16	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
17	Integrated Genomic Analysis Identifies ANKRD36 Gene as a Novel and Common Biomarker of Disease Progression in Chronic Myeloid Leukemia. Biology, 2021, 10, 1182.	1.3	5
18	A novel missense variant in the RASGRP2 gene in patients with moderate to severe bleeding disorder. Platelets, 2020, 31, 646-651.	1.1	3

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19	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
20	Novel heterozygous sequence variant in the GLI1 underlies postaxial polydactyly. Congenital Anomalies (discontinued), 2020, 60, 115-119.	0.3	7
21	Centromere protein I (CENPI) is a candidate gene for X-linked steroid sensitive nephrotic syndrome. Journal of Nephrology, 2020, 33, 763-769.	0.9	6
22	Identification of TMC1 as a relatively common cause for nonsyndromic hearing loss in the Saudi population. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 172-180.	1.1	14
23	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
24	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
25	A homozygous missense variant in the homeobox domain of the NKX6â€2 results in progressive spastic ataxia type 8 associated with lower limb weakness and neurological manifestations. Journal of Gene Medicine, 2020, 22, e3196.	1.4	3
26	Novel and recurrent germline mutations in the VHL gene in 5 Arab patients with Von Hippel-Lindau disease. Cancer Genetics, 2020, 243, 1-6.	0.2	2
27	Homozygosity mapping and whole exome sequencing provide exact diagnosis of Cohen syndrome in a Saudi family. Brain and Development, 2020, 42, 587-593.	0.6	5
28	A novel nonsense mutation in the STS gene in a Pakistani family with X-linked recessive ichthyosis: including a very rare case of two homozygous female patients. BMC Medical Genetics, 2020, 21, 20.	2.1	4
29	Mutation screening of genes associated with congenital talipes equinovarus in pakistani families. Journal of Musculoskeletal Surgery and Research, 2020, 4, 25.	0.2	3
30	Further Evidence of a Recessive Variant in COL1A1 as an Underlying Cause of Ehlersâ€“Danlos Syndrome: A Report of a Saudi Founder Mutation. Global Medical Genetics, 2020, 07, 109-112.	0.4	1
31	Apparent Missense Variant in COL7A1 Causes a Severe Form of Recessive Dystrophic Epidermolysis Bullosa via Effects on Splicing. Acta Dermato-Venereologica, 2020, 100, adv00275.	0.6	2
32	Isolated congenital vertical talus: Genetics and genomics. Journal of Musculoskeletal Surgery and Research, 2020, 4, 66.	0.2	0
33	Missense Mutations in theCTSCGene in Saudi Families Segregating Papillon-LefÃ¨vre Syndrome. Annals of Dermatology, 2020, 32, 77.	0.3	3
34	Whole exome sequencing identifies a novel FANCD2 gene splice site mutation associated with disease progression in chronic myeloid leukemia: Implication in targeted therapy of advanced phase CML. Pakistan Journal of Pharmaceutical Sciences, 2020, 33, 1419-1426.	0.2	3
35	Thymic Stromal Lymphopoietin (TSLP) gene variant rs1837253 is significantly associated with Asthma prevalence in Pakistani Pashtun women. Pakistan Journal of Pharmaceutical Sciences, 2020, 33, 2729-2737.	0.2	1
36	Kleineâ€Levin syndrome is associated with LMOD3 variants. Journal of Sleep Research, 2019, 28, e12718.	1.7	12

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37	An 18 bps in-frame deletion mutation in RUNX2 gene is a population polymorphism rather than a pathogenic variant. European Journal of Medical Genetics, 2019, 62, 124-128.	0.7	9
38	UV-sensitive syndrome: Whole exome sequencing identified a nonsense mutation in the gene UVSSA in two consanguineous pedigrees from Pakistan. Journal of Dermatological Science, 2019, 95, 113-118.	1.0	6
39	KMT2C, a histone methyltransferase, is mutated in a family segregating non-syndromic primary failure of tooth eruption. Scientific Reports, 2019, 9, 16469.	1.6	15
40	Whole genome genotyping mapped regions on chromosome 2 and 18 in a family segregating Waardenburg syndrome type II. Saudi Journal of Ophthalmology, 2019, 33, 326-331.	0.3	2
41	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
42	Biallelic mutations in the <scp>LPAR</scp>6 gene causing autosomal recessive wooly hair/hypotrichosis phenotype in five Pakistani families. International Journal of Dermatology, 2019, 58, 946-952.	0.5	4
43	Detection of rifampicin resistance of Mycobacterium tuberculosis using multiplex allele specific polymerase chain reaction (MAS-PCR) in Pakistan. Infection, Genetics and Evolution, 2019, 71, 42-46.	1.0	6
44	A Heterozygous Mutation in the Triple Helical Region of the Alpha 1 (II) Chain of the COL2A1 Protein Causes Non-Lethal Spondyloepiphyseal Dysplasia Congenita. Genetic Testing and Molecular Biomarkers, 2019, 23, 310-315.	0.3	5
45	Genetic Basis of Polycystic Ovary Syndrome (PCOS): Current Perspectives . The Application of Clinical Genetics, 2019, Volume 12, 249-260.	1.4	159
46	Novel homozygous loss-of-function mutations in RP1 and RP1L1 genes in retinitis pigmentosa patients. Ophthalmic Genetics, 2019, 40, 507-513.	0.5	19
47	XPC gene mutations in families with xeroderma pigmentosum from Pakistan; prevalent founder effect. Congenital Anomalies (discontinued), 2019, 59, 18-21.	0.3	9
48	Genetics of developmental dysplasia of the hip: Recent progress and future perspectives. Journal of Musculoskeletal Surgery and Research, 2019, 3, 245.	0.2	1
49	A Homozygous Missense Variant in the APOB gene in Patients from Hypercholesterolemia Families. Egyptian Academic Journal of Biological Sciences C Physiology and Molecular Biology, 2019, 11, 31-37.	0.0	1
50	Investigations on Novel Gene Variants Associated with Longterm Response to Tyrosine Kinase Inhibitors (TKIs) in Chronic Myeloid Leukemia: Implication in TKI-Cessation Clinical Trails. Blood, 2019, 134, 2939-2939.	0.6	0
51	A novel mutation in the HPGD gene causing primary hypertrophic osteoarthropathy with digital clubbing in a Pakistani family. Annals of Human Genetics, 2018, 82, 171-176.	0.3	9
52	Whole exome sequencing identified a novel single base pair insertion mutation in the EYS gene in a six generation family with retinitis pigmentosa. Congenital Anomalies (discontinued), 2018, 58, 10-15.	0.3	11
53	Whole genome SNP genotyping in a family segregating developmental dysplasia of the hip detected runs of homozygosity on chromosomes 15q13.3 and 19p13.2. Congenital Anomalies (discontinued), 2018, 58, 56-61.	0.3	17
54	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

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55	Genetics of clubfoot; recent progress and future perspectives. European Journal of Medical Genetics, 2018, 61, 107-113.	0.7	42
56	Pakistan Genetic Mutation Database (PGMD); A centralized Pakistani mutome data source. European Journal of Medical Genetics, 2018, 61, 204-208.	0.7	19
57	Exome sequencing revealed a novel nonsense variant in ALX3 gene underlying frontorhiny. Journal of Human Genetics, 2018, 63, 97-100.	1.1	6
58	X-linked ADGRG2 mutation and obstructive azoospermia in a large Pakistani family. Scientific Reports, 2018, 8, 16280.	1.6	26
59	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
60	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
61	Whole Genome Sequencing instead of Whole Exome Sequencing is required to identify the Genetic Causes of Polycystic Ovary Syndrome in Pakistani families. Pakistan Journal of Medical Sciences, 2018, 34, 540-545.	0.3	6
62	Latest perspectives of orally bioavailable 2,4-diarylaminopyrimidine analogues (DAAPalogues) as anaplastic lymphoma kinase inhibitors: discovery and clinical developments. RSC Advances, 2018, 8, 16470-16493.	1.7	5
63	Targeted Next-Generation Sequencing of 406 Genes Identified Genetic Defects Underlying Congenital Heart Disease in Down Syndrome Patients. Pediatric Cardiology, 2018, 39, 1676-1680.	0.6	19
64	Whole exome sequencing identification of a novel insertion mutation in the phospholipase C Îµâ€‘1 gene in a family with steroid resistant inherited nephrotic syndrome. Molecular Medicine Reports, 2018, 18, 5095-5100.	1.1	9
65	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
66	A novel homozygous variant in the SMOC1 gene underlying Waardenburg anophthalmia syndrome. Ophthalmic Genetics, 2017, 38, 335-339.	0.5	8
67	First macrocyclic 3 rd -generation ALK inhibitor for treatment of ALK/ROS1 cancer: Clinical and designing strategy update of lorlatinib. European Journal of Medicinal Chemistry, 2017, 134, 348-356.	2.6	79
68	Exome sequencing identified rare variants in genes HSPG2 and ATP2B4 in a family segregating developmental dysplasia of the hip. BMC Medical Genetics, 2017, 18, 34.	2.1	32
69	A homozygous potentially pathogenic variant in the <scp>PAXBP1</scp> gene in a large family with global developmental delay and myopathic hypotonia. Clinical Genetics, 2017, 92, 579-586.	1.0	16
70	Next-Generation sequencing and molecular diagnosis in musculoskeletal disorders. Journal of Musculoskeletal Surgery and Research, 2017, 1, 23.	0.2	2
71	Developmental dysplasia of the hip: usefulness of next generation genomic tools for characterizing the underlying genes â€“ a mini review. Clinical Genetics, 2016, 90, 16-20.	1.0	18
72	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

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73	Sequence analysis of the VSX1 and SOD1 genes in families with Keratoconus and a review of the literature. Journal of Taibah University Medical Sciences, 2016, 11, 115-120.	0.5	3
74	Xq21.31â€“q21.32 duplication underlies intellectual disability in a large family with five affected males. American Journal of Medical Genetics, Part A, 2016, 170, 87-93.	0.7	5
75	CIT, a gene involved in neurogenic cytokinesis, is mutated in human primary microcephaly. Human Genetics, 2016, 135, 1199-1207.	1.8	45
76	High-resolution SNP genotyping platform identified recurrent and novel CNVs in autism multiplex families. Neuroscience, 2016, 339, 561-570.	1.1	17
77	Sequence analysis of four vitamin D family genes (VDR, CYP24A1, CYP27B1 and CYP2R1) in Vogt-Koyanagi-Harada (VKH) patients: identification of a potentially pathogenic variant in CYP2R1. BMC Ophthalmology, 2016, 16, 172.	0.6	10
78	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
79	Genetics of human isolated acromesomelic dysplasia. European Journal of Medical Genetics, 2016, 59, 198-203.	0.7	18
80	Homozygous sequence variants in the FKBP10 gene underlie osteogenesis imperfecta in consanguineous families. Journal of Human Genetics, 2016, 61, 207-213.	1.1	32
81	A novel splice-site mutation in the ASPM gene underlies autosomal recessive primary microcephaly. Annals of Saudi Medicine, 2016, 36, 391-396.	0.5	19
82	Prognostic Stratification of Acute Myeloid Leukemia and Mylodysplastic Syndrome Patients on the Basis of Genetic Variations. Blood, 2016, 128, 5239-5239.	0.6	1
83	The need for population-based studies to estimate the rate of consanguinity in Almadinah Almunawwarah. Journal of Taibah University Medical Sciences, 2015, 10, 509-511.	0.5	0
84	Genetics of human isolated hereditary nail disorders. British Journal of Dermatology, 2015, 173, 922-929.	1.4	20
85	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
86	Whole-genome SNP genotyping mapped a novel locus for hereditary hypotrichosis on chromosome 2q31.1â€“q32.2. Journal of Dermatological Science, 2015, 79, 173-175.	1.0	0
87	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
88	Genetics of human isolated hereditary hair loss disorders. Clinical Genetics, 2015, 88, 203-212.	1.0	10
89	Exome sequencing reveals MCM8 mutation underlies ovarian failure and chromosomal instability. Journal of Clinical Investigation, 2015, 125, 258-262.	3.9	178
90	Linkage analysis coupled with exome sequencing identified defects in gene â€˜Xâ€™ causing premature ovarian insufficiency. BMC Genomics, 2014, 15, .	1.2	0

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91	A founder splice site mutation underlies glycogen storage disease type 3 in consanguineous Saudi families. Annals of Saudi Medicine, 2014, 34, 390-395.	0.5	4
92	A novel WDR62 mutation causes primary microcephaly in a Pakistani family. Molecular Biology Reports, 2013, 40, 591-595.	1.0	16
93	Mutations in KARS, Encoding Lysyl-tRNA Synthetase, Cause Autosomal-Recessive Nonsyndromic Hearing Impairment DFNB89. American Journal of Human Genetics, 2013, 93, 132-140.	2.6	90
94	Novel homozygous mutations in the genes ARL6 and BBS10 underlying Bardetâ€“Biedl syndrome. Gene, 2013, 515, 84-88.	1.0	23
95	Vitamin D in health and disease: a literature review. British Journal of Biomedical Science, 2013, 70, 161-172.	1.2	111
96	Novel <scp>OTOA</scp> mutations cause autosomal recessive nonâ€syndromic hearing impairment in Pakistani families. Clinical Genetics, 2013, 84, 294-296.	1.0	14
97	Five Most Common Prognostically Important Fusion Oncogenes are Detected in the Majority of Pakistani Pediatric Acute Lymphoblastic Leukemia Patients and are Strongly Associated with Disease Biology and Treatment Outcome. Asian Pacific Journal of Cancer Prevention, 2012, 13, 5469-5475.	0.5	17
98	A novel chondroectodermal dysplasia mapped to chromosome 2q24.1-q31.1. European Journal of Medical Genetics, 2012, 55, 455-460.	0.7	5
99	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
100	A novel homozygous missense mutation in WNT10B in familial splitâ€hand/foot malformation. Clinical Genetics, 2012, 82, 48-55.	1.0	38
101	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
102	Prognostically Significant Fusion Oncogenes in Pakistani Patients with Adult Acute Lymphoblastic Leukemia and their Association with Disease Biology and Outcome. Asian Pacific Journal of Cancer Prevention, 2012, 13, 3349-3355.	0.5	14
103	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
104	Digenic inheritance of an autosomal recessive hypotrichosis in two consanguineous pedigrees. Clinical Genetics, 2011, 79, 273-281.	1.0	14
105	Congenital atrichia with papular lesions resulting from novel mutations in human hairless gene in four consanguineous families. Journal of Dermatology, 2011, 38, 755-760.	0.6	11
106	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
107	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
108	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	2.6	108

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109	DFNB89, a novel autosomal recessive nonsyndromic hearing impairment locus on chromosome 16q21-q23.2. Human Genetics, 2011, 129, 379-385.	1.8	11
110	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
111	Mutations in WDR62 gene in Pakistani families with autosomal recessive primary microcephaly. BMC Neurology, 2011, 11, 119.	0.8	26
112	Novel Autosomal Recessive Nonsyndromic Hearing Impairment Locus DFNB90 Maps to 7p22.1-p15.3. Human Heredity, 2011, 71, 106-112.	0.4	4
113	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
114	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
115	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
116	Mapping of a novel locus for an autosomal recessive form of palmoplantar keratoderma on chromosome 3q27.2-q29. British Journal of Dermatology, 2010, 163, 711-718.	1.4	6
117	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
118	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
119	Mutations in theP2RY5gene underlie autosomal recessive hypotrichosis in 13 Pakistani families. British Journal of Dermatology, 2009, 160, 1006-1010.	1.4	20
120	A Homozygous Nonsense Mutation in the Human Desmocollin-3 (DSC3) Gene Underlies Hereditary Hypotrichosis and Recurrent Skin Vesicles. American Journal of Human Genetics, 2009, 85, 515-520.	2.6	75
121	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
122	Recurrent mutation in CDMP1 in a family with Grebe chondrodysplasia: broadening the phenotypic manifestation of syndrome in Pakistani population. Pakistan Journal of Medical Sciences, 1969, 31, 1542-4.	0.3	0