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	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
2	Exome sequencing reveals MCM8 mutation underlies ovarian failure and chromosomal instability. Journal of Clinical Investigation, 2015, 125, 258-262.	3.9	178
3	Genetic Basis of Polycystic Ovary Syndrome (PCOS): Current Perspectives . The Application of Clinical Genetics, 2019, Volume 12, 249-260.	1.4	159
4	Vitamin D in health and disease: a literature review. British Journal of Biomedical Science, 2013, 70, 161-172.	1.2	111
5	Loss-of-Function Mutations of ILDR1 Cause Autosomal-Recessive Hearing Impairment DFNB42. American Journal of Human Genetics, 2011, 88, 127-137.	2.6	108
6	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
7	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
8	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
9	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
10	CIT, a gene involved in neurogenic cytokinesis, is mutated in human primary microcephaly. Human Genetics, 2016, 135, 1199-1207.	1.8	45
11	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
12	Genetics of clubfoot; recent progress and future perspectives. European Journal of Medical Genetics, 2018, 61, 107-113.	0.7	42
13	A novel homozygous missense mutation in WNT10B in familial splitâ€hand/foot malformation. Clinical Genetics, 2012, 82, 48-55.	1.0	38
14	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
15	Homozygous sequence variants in the FKBP10 gene underlie osteogenesis imperfecta in consanguineous families. Journal of Human Genetics, 2016, 61, 207-213.	1.1	32
16	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
17	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
18	Mutations in WDR62 gene in Pakistani families with autosomal recessive primary microcephaly. BMC Neurology, 2011, 11, 119.	0.8	26

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19	X-linked ADGRG2 mutation and obstructive azoospermia in a large Pakistani family. Scientific Reports, 2018, 8, 16280.	1.6	26
20	Novel homozygous mutations in the genes ARL6 and BBS10 underlying Bardetâ€“Biedl syndrome. Gene, 2013, 515, 84-88.	1.0	23
21	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
22	Mutations in theP2RY5gene underlie autosomal recessive hypotrichosis in 13 Pakistani families. British Journal of Dermatology, 2009, 160, 1006-1010.	1.4	20
23	Genetics of human isolated hereditary nail disorders. British Journal of Dermatology, 2015, 173, 922-929.	1.4	20
24	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
25	Pakistan Genetic Mutation Database (PGMD); A centralized Pakistani mutome data source. European Journal of Medical Genetics, 2018, 61, 204-208.	0.7	19
26	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
27	Novel homozygous loss-of-function mutations in RP1 and RP1L1 genes in retinitis pigmentosa patients. Ophthalmic Genetics, 2019, 40, 507-513.	0.5	19
28	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
29	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
30	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
31	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
32	Genetics of human isolated acromesomelic dysplasia. European Journal of Medical Genetics, 2016, 59, 198-203.	0.7	18
33	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
34	High-resolution SNP genotyping platform identified recurrent and novel CNVs in autism multiplex families. Neuroscience, 2016, 339, 561-570.	1.1	17
35	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
36	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

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37	A novel WDR62 mutation causes primary microcephaly in a Pakistani family. Molecular Biology Reports, 2013, 40, 591-595.	1.0	16
38	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
39	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
40	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
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	Digenic inheritance of an autosomal recessive hypotrichosis in two consanguineous pedigrees. Clinical Genetics, 2011, 79, 273-281.	1.0	14
43	Novel <scp>OTOA</scp> mutations cause autosomal recessive nonâ€syndromic hearing impairment in Pakistani families. Clinical Genetics, 2013, 84, 294-296.	1.0	14
44	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
45	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
46	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
47	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
48	Kleineâ€Levin syndrome is associated with LMOD3 variants. Journal of Sleep Research, 2019, 28, e12718.	1.7	12
49	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
50	DFNB89, a novel autosomal recessive nonsyndromic hearing impairment locus on chromosome 16q21-q23.2. Human Genetics, 2011, 129, 379-385.	1.8	11
51	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
52	Genetics of human isolated hereditary hair loss disorders. Clinical Genetics, 2015, 88, 203-212.	1.0	10
53	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
54	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

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55	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
56	XPC gene mutations in families with xeroderma pigmentosum from Pakistan; prevalent founder effect. Congenital Anomalies (discontinued), 2019, 59, 18-21.	0.3	9
57	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
58	A novel homozygous variant in the SMOC1 gene underlying Waardenburg anophthalmia syndrome. Ophthalmic Genetics, 2017, 38, 335-339.	0.5	8
59	First evidence of involvement of TBC1D25 in causing human male infertility. European Journal of Medical Genetics, 2021, 64, 104142.	0.7	8
60	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
61	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
62	Novel heterozygous sequence variant in the GLI1 underlies postaxial polydactyly. Congenital Anomalies (discontinued), 2020, 60, 115-119.	0.3	7
63	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
64	Exome sequencing revealed a novel nonsense variant in ALX3 gene underlying frontorhiny. Journal of Human Genetics, 2018, 63, 97-100.	1.1	6
65	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
66	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
67	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
68	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
69	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
70	Novel Homozygous Mutations in the Genes TGM1, SULT2B1, SPINK5 and FLG in Four Families Underlying Congenital Ichthyosis. Genes, 2021, 12, 373.	1.0	6
71	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
72	A novel chondroectodermal dysplasia mapped to chromosome 2q24.1-q31.1. European Journal of Medical Genetics, 2012, 55, 455-460.	0.7	5

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73	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
74	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
75	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
76	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
77	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
78	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
79	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
80	Novel Autosomal Recessive Nonsyndromic Hearing Impairment Locus DFNB90 Maps to 7p22.1-p15.3. Human Heredity, 2011, 71, 106-112.	0.4	4
81	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
82	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
83	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
84	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
85	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
86	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
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	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
89	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
90	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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91	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
92	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
93	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
94	Mutation screening of genes associated with congenital talipes equinovarus in pakistani families. Journal of Musculoskeletal Surgery and Research, 2020, 4, 25.	0.2	3
95	Missense Mutations in theCTSCGene in Saudi Families Segregating Papillon-LefÃ¨vre Syndrome. Annals of Dermatology, 2020, 32, 77.	0.3	3
96	Association between 17q21 variants and asthma predisposition in Pashtun population from Pakistan. Journal of Asthma, 2022, , 1-13.	0.9	3
97	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
98	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
99	A novel nonsense variant in EXOC8 underlies a neurodevelopmental disorder. Neurogenetics, 2022, 23, 203-212.	0.7	3
100	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
101	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
102	Next-Generation sequencing and molecular diagnosis in musculoskeletal disorders. Journal of Musculoskeletal Surgery and Research, 2017, 1, 23.	0.2	2
103	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
104	Exome Sequencing Revealed the First Intragenic Deletion in ABCA5 Underlying Autosomal Recessive Hypertrichosis. Clinical and Experimental Dermatology, 2022, , .	0.6	2
105	Association of SORD mutation with autosomal recessive asymmetric distal hereditary motor neuropathy. BMC Medical Genomics, 2022, 15, 88.	0.7	2
106	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
107	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
108	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

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109	Prognostic Stratification of Acute Myeloid Leukemia and Mylodysplastic Syndrome Patients on the Basis of Genetic Variations. Blood, 2016, 128, 5239-5239.	0.6	1
110	Genetics of developmental dysplasia of the hip: Recent progress and future perspectives. Journal of Musculoskeletal Surgery and Research, 2019, 3, 245.	0.2	1
111	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
112	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
113	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
114	Ultrastructure abnormalities of collagen and elastin in Arab patients with arterial tortuosity syndrome. Journal of Cutaneous Pathology, 2022, , .	0.7	1
115	A homozygous missense variant in the ‹i›MLC1‹/i› 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
116	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
117	Linkage analysis coupled with exome sequencing identified defects in gene â€˜Xâ€™ causing premature ovarian insufficiency. BMC Genomics, 2014, 15, .	1.2	0
118	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
119	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
120	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
121	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
122	Isolated congenital vertical talus: Genetics and genomics. Journal of Musculoskeletal Surgery and Research, 2020, 4, 66.	0.2	0