

Sajid Malik

List of Publications by Year in descending order

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76
papers

1,139
citations

430874

18
h-index

434195

31
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77
docs citations

77
times ranked

1302
citing authors

#	ARTICLE	IF	CITATIONS
1	A Two-Base Pair Deletion in IQ Repeats in ASPM Underlies Microcephaly in a Pakistani Family. <i>Genetic Testing and Molecular Biomarkers</i> , 2022, 26, 37-42.	0.7	4
2	Mutation in protein disulfide isomerase A3 causes neurodevelopmental defects by disturbing endoplasmic reticulum proteostasis. <i>EMBO Journal</i> , 2022, 41, e105531.	7.8	11
3	KERATIN 17-related recessive atypical pachyonychia congenita with variable hair and tooth anomalies. <i>European Journal of Human Genetics</i> , 2022, 30, 1292-1296.	2.8	4
4	Biallelic loss-of-function variants in NEMF cause central nervous system impairment and axonal polyneuropathy. <i>Human Genetics</i> , 2021, 140, 579-592.	3.8	14
5	Homozygous deletion of MYADML2 in cranial asymmetry, reduced bone maturation, multiple dislocations, lumbar lordosis, and prominent clavicles. <i>Journal of Human Genetics</i> , 2021, 66, 171-179.	2.3	2
6	A splice-site variant (c.3289-1G>T) in OTOF underlies profound hearing loss in a Pakistani kindred. <i>BMC Medical Genomics</i> , 2021, 14, 2.	1.5	3
7	Consanguinity, inbreeding coefficient, fertility and birth-outcome in population of Okara district, Pakistan. <i>Pakistan Journal of Medical Sciences</i> , 2021, 37, 770-775.	0.6	3
8	Prevalence-pattern and risk factors of Cesarean section in multiethnic cohort. <i>Pakistan Journal of Medical Sciences</i> , 2021, 37, 711-715.	0.6	2
9	The first adolescent case of Fraser syndrome 3, with a novel nonsense variant in <i>GRIP1</i> . <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1858-1863.	1.2	4
10	CRADD and USP44 mutations in intellectual disability, mild lissencephaly, brain atrophy, developmental delay, strabismus, behavioural problems and skeletal anomalies. <i>European Journal of Medical Genetics</i> , 2021, 64, 104181.	1.3	8
11	Congenital limb defects in a married female population of the Rahim Yar Khan District in Pakistan. <i>Asian Biomedicine</i> , 2021, 15, 137-144.	0.3	0
12	Frequencies of ABO and Rh (D) Blood Group Phenotypes in Pashtuns of North-Western Pakistan: A population undergoing huge demographic changes. <i>International Journal of Immunogenetics</i> , 2021, 48, 336-339.	1.8	0
13	A homozygous ROR2 variant in a family with atypical Robinow syndrome and tetramelic transverse deficiency of autopods. <i>American Journal of Medical Genetics, Part A</i> , 2021, , .	1.2	1
14	A novel ZRS variant causes preaxial polydactyly type I by increased sonic hedgehog expression in the developing limb bud. <i>Genetics in Medicine</i> , 2020, 22, 189-198.	2.4	16
15	Biallelic ZNF407 mutations in a neurodevelopmental disorder with ID, short stature and variable microcephaly, hypotonia, ocular anomalies and facial dysmorphism. <i>Journal of Human Genetics</i> , 2020, 65, 1115-1123.	2.3	5
16	Novel EDAR mutation in tooth agenesis and variable associated features. <i>European Journal of Medical Genetics</i> , 2020, 63, 103926.	1.3	6
17	A founder RDH5 splice site mutation leads to retinitis punctata albescens in two inbred Pakistani kindreds. <i>Ophthalmic Genetics</i> , 2020, 41, 7-12.	1.2	0
18	Talipes equinovarus or Clubfoot: A review of study approaches, management and trends in Pakistan. <i>Pakistan Journal of Medical Sciences</i> , 2020, 36, 1414-1420.	0.6	2

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19	Epidemiological study of congenital and hereditary anomalies in Stalkot District of Pakistan revealed a high incidence of limb and neurological disorders. <i>Asian Biomedicine</i> , 2019, 13, 49-60.	0.3	2
20	Novel Polymorphism in the Promoter Region of HLA-DQB1 Is a Predictor of Anti-HCV Therapy Response. <i>Jundishapur Journal of Microbiology</i> , 2019, 12, .	0.5	1
21	Homozygous <i>CHST11</i> mutation in chondrodysplasia, brachydactyly, overriding digits, clinodactyly and synpolydactyly. <i>Journal of Medical Genetics</i> , 2018, 55, 489-496.	3.2	12
22	Mesoaxial synostotic syndactyly with phalangeal reduction (MSSD): syndactyly type IX. <i>Skeletal Radiology</i> , 2018, 47, 149-149.	2.0	2
23	Linked homozygous <i>BMPR1B</i> and <i>PDHA2</i> variants in a consanguineous family with complex digit malformation and male infertility. <i>European Journal of Human Genetics</i> , 2018, 26, 876-885.	2.8	10
24	Homozygous mutation in <i>CEP19</i> , a gene mutated in morbid obesity, in Bardet-Biedl syndrome with predominant postaxial polydactyly. <i>Journal of Medical Genetics</i> , 2018, 55, 189-197.	3.2	25
25	Exome Sequencing and Rare Variant Analysis Reveals Multiple Filaggrin Mutations in Bangladeshi Families with Atopic Eczema and Additional Risk Genes. <i>Journal of Investigative Dermatology</i> , 2018, 138, 2674-2677.	0.7	37
26	Progressive SCAR14 with unclear speech, developmental delay, tremor, and behavioral problems caused by a homozygous deletion of the <i>SPTBN2</i> pleckstrin homology domain. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2494-2499.	1.2	19
27	Novel splice mutation in <i>LRP4</i> causes severe type of Cenani-Lenz syndactyly syndrome with oro-facial and skeletal symptoms. <i>European Journal of Medical Genetics</i> , 2017, 60, 421-425.	1.3	13
28	Complex postaxial polydactyly types A and B with camptodactyly, hypoplastic third toe, zygodactyly and other digit anomalies caused by a novel <i>GLI3</i> mutation. <i>European Journal of Medical Genetics</i> , 2017, 60, 268-274.	1.3	6
29	Analysis of Genetic Differentiation at ABO and Rh Loci among the Pashtun Populations Inhabiting Lower Khyber Pakhtunkhwa, Pakistan. <i>Pakistan Journal of Zoology</i> , 2017, 49, 387-389.	0.2	1
30	Determinants of consanguinity and inbreeding coefficient in the multiethnic population of Mardan, Khyber Pakhtunkhwa, Pakistan. <i>Asian Biomedicine</i> , 2017, 11, 451-460.	0.3	3
31	TRANSITION IN CONSANGUINITY IN DIR LOWER DISTRICT, A VICTIM OF WAR, NATURAL DISASTER AND POPULATION DISPLACEMENT, IN NORTH-WEST PAKISTAN – A RESPONSE TO STHANADAR ET AL. (2015). <i>Journal of Biosocial Science</i> , 2016, 48, 421-426.	1.2	7
32	Consanguinity and its socio-biological parameters in Rahim Yar Khan District, Southern Punjab, Pakistan. <i>Journal of Health, Population and Nutrition</i> , 2016, 35, 14.	2.0	14
33	CONSANGUINITY AND INBREEDING COEFFICIENT IN TRIBAL PASHTUNS INHABITING THE TURBULENT AND WAR-AFFECTED TERRITORY OF BAJAUR AGENCY, NORTH-WEST PAKISTAN. <i>Journal of Biosocial Science</i> , 2016, 48, 113-128.	1.2	12
34	Distribution of ABO and Rh(D) Allelic Polymorphisms North Waziristan Agency, (Federally) Tj ETQq0 0 0,rgBT /Overlock 10 T	0.3	1
35	Seroprevalence of HDV among non-hospitalized HBsAg positive patients from KPK-region of Pakistan. <i>Asian Pacific Journal of Tropical Biomedicine</i> , 2016, 6, 609-613.	1.2	4
36	Prevalence of multi-drug resistant uropathogenic <i>Escherichia coli</i> in Potohar region of Pakistan. <i>Asian Pacific Journal of Tropical Biomedicine</i> , 2016, 6, 60-66.	1.2	44

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37	Determinants of Consanguinity and Inbreeding Coefficient F in Dir Lower District, North-West Pakistan: A Multivariate Approach. Iranian Journal of Public Health, 2016, 45, 537-9.	0.5	3
38	<i>RBBP8</i> syndrome with microcephaly, intellectual disability, short stature and brachydactyly. American Journal of Medical Genetics, Part A, 2015, 167, 3148-3152.	1.2	5
39	Descriptive epidemiology of hereditary musculoskeletal and limb defects in the isolated population of Chitral, North-West Pakistan.. Pakistan Journal of Medical Sciences, 2015, 31, 1047-52.	0.6	1
40	Genetic Diversity at ABO and Rh (D) Loci in the Tribal Groups of Mohmand Agency (Federally) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 622	0.1	1
41	Molecular Diagnosis of Fragile X Syndrome in Subjects with Intellectual Disability of Unknown Origin: Implications of Its Prevalence in Regional Pakistan. PLoS ONE, 2015, 10, e0122213.	2.5	8
42	PATTERN OF CONSANGUINITY AND INBREEDING COEFFICIENT IN SARGODHA DISTRICT, PUNJAB, PAKISTAN. Journal of Biosocial Science, 2015, 47, 803-811.	1.2	15
43	Congenital constriction ring of limbs in subjects with history of maternal substance use. Journal of the College of Physicians and Surgeons–Pakistan: JCPSP, 2015, 25, 383-5.	0.4	0
44	Prevalence and pattern of traumatic limb amputations in female population of District Bhimber, Azad Jammu and Kashmir, Pakistan. Pakistan Journal of Medical Sciences, 2014, 31, 54-9.	0.6	4
45	Mutations Affecting the BHLHA9 DNA-Binding Domain Cause MSSD, Mesoaxial Synostotic Syndactyly with Phalangeal Reduction, Malik-Percin Type. American Journal of Human Genetics, 2014, 95, 649-659.	6.2	27
46	Clinical and descriptive genetic study of polydactyly: a Pakistani experience of 313 cases. Clinical Genetics, 2014, 85, 482-486.	2.0	33
47	Prevalence of hepatitis C virus infection among thalassemia patients: a perspective from a multi-ethnic population of Pakistan. Asian Pacific Journal of Tropical Medicine, 2014, 7, S127-S133.	0.8	23
48	Polydactyly: phenotypes, genetics and classification. Clinical Genetics, 2014, 85, 203-212.	2.0	131
49	ABO and Rh (D) Blood Groups Polymorphism in Four Tehsils of Bajaur Agency (Federally Administered) Tj ETQq1 1 0,784314 rgBT /Overlock 10 Tf 50 622	0.1	5
50	Longitudinal deficiency of upper limb: similar case presentation of two subjects with unilateral ulnar hemimelia, carpal and metacarpal deficiency, and severe oligodactyly. Asian Biomedicine, 2014, 8, 569-575.	0.3	1
51	Consanguinity and its sociodemographic differentials in Bhimber District, Azad Jammu and Kashmir, Pakistan. Journal of Health, Population and Nutrition, 2014, 32, 301-13.	2.0	20
52	Prevalence of Congenital Anomalies and Non-Communicable Diseases in Women of Age 12-75 Years in District Bhimber, Azad Jammu and Kashmir, Pakistan. Iranian Journal of Public Health, 2014, 43, 42-9.	0.5	3
53	Ulnar aplasia, dysplastic radius and preaxial oligodactyly: Rare longitudinal limb defect in a sporadic male child. Journal of Research in Medical Sciences, 2013, 18, 818-21.	0.9	3
54	Congenital terminal transverse deformity of upper limb: clinical and radiological findings in a sporadic care. Journal of the College of Physicians and Surgeons–Pakistan: JCPSP, 2013, 23, 219-20.	0.4	3

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55	Syndactyly: phenotypes, genetics and current classification. <i>European Journal of Human Genetics</i> , 2012, 20, 817-824.	2.8	136
56	A novel ZRS mutation in a Balochi tribal family with triphalangeal thumb, preaxial polydactyly, postaxial polydactyly, and syndactyly. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2031-2035.	1.2	13
57	Zygodactyly with thumb aplasia: an unusual variant in a male subject. <i>Journal of the College of Physicians and Surgeons–Pakistan: JCPSP</i> , 2011, 21, 710-2.	0.4	4
58	Autosomal dominant syndrome of camptodactyly, clinodactyly, syndactyly, and bifid toes. <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 2313-2317.	1.2	1
59	Human intronic enhancers control distinct sub-domains of Gli3 expression during mouse CNS and limb development. <i>BMC Developmental Biology</i> , 2010, 10, 44.	2.1	32
60	Synpolydactyly: clinical and molecular advances. <i>Clinical Genetics</i> , 2008, 73, 113-120.	2.0	58
61	Congenital, low penetrance lymphedema of lower limbs maps to chromosome 6q16.2–q22.1 in an inbred Pakistani family. <i>Human Genetics</i> , 2008, 123, 197-205.	3.8	16
62	Fifth finger camptodactyly maps to chromosome 3q11.2–q13.12 in a large German kindred. <i>European Journal of Human Genetics</i> , 2008, 16, 265-269.	2.8	12
63	Synpolydactyly and HOXD13 polyalanine repeat: addition of 2 alanine residues is without clinical consequences. <i>BMC Medical Genetics</i> , 2007, 8, 78.	2.1	17
64	Ultraconserved non-coding sequence element controls a subset of spatiotemporal <i>GLI3</i> expression. <i>Development Growth and Differentiation</i> , 2007, 49, 543-553.	1.5	35
65	Human GLI3 Intragenic Conserved Non-Coding Sequences Are Tissue-Specific Enhancers. <i>PLoS ONE</i> , 2007, 2, e366.	2.5	39
66	Genetic heterogeneity of synpolydactyly: a novel locus SPD3 maps to chromosome 14q11.2-q12. <i>Clinical Genetics</i> , 2006, 69, 518-524.	2.0	23
67	Evidence for clinical and genetic heterogeneity of syndactyly type I: the phenotype of second and third toe syndactyly maps to chromosome 3p21.31. <i>European Journal of Human Genetics</i> , 2005, 13, 1268-1274.	2.8	22
68	A Novel Locus of Ectodermal Dysplasia Maps to Chromosome 10q24.32–q25.1. <i>Journal of Investigative Dermatology</i> , 2005, 124, 338-342.	0.7	6
69	Autosomal recessive mesoaxial synostotic syndactyly with phalangeal reduction maps to chromosome 17p13.3. <i>American Journal of Medical Genetics, Part A</i> , 2005, 134A, 404-408.	1.2	20
70	Multiple Familial Trichoepithelioma Caused by Mutations in the Cylindromatosis Tumor Suppressor Gene. <i>Cancer Research</i> , 2004, 64, 5113-5117.	0.9	70
71	A novel type of autosomal recessive syndactyly: Clinical and molecular studies in a family of Pakistani origin. <i>American Journal of Medical Genetics Part A</i> , 2004, 126A, 61-67.	2.4	18
72	A locus for hereditary hypotrichosis localized to human chromosome 18q21.1. <i>European Journal of Human Genetics</i> , 2003, 11, 623-628.	2.8	30

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73	Study of non-syndromic thumb aplasia in six independent cases.. Pakistan Journal of Medical Sciences, 1969, 30, 677-81.	0.6	2
74	Phenotypic manifestation of congenital transverse amputation of autopod in Pakistani subjects. Pakistan Journal of Medical Sciences, 1969, 32, 519-22.	0.6	0
75	Congenital hypoplasia of first digital ray of hands as an isolated presentation in four subjects. Pakistan Journal of Medical Sciences, 1969, 30, 1428-31.	0.6	1
76	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.	0.6	0