# 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 g-index |
| 77 all docs | 77 docs citations | 77 times ranked | 1302 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Syndactyly: phenotypes, genetics and current classification. European Journal of Human Genetics, 2012, 20, 817-824. | 2.8 | 136 |
| 2 | Polydactyly: phenotypes, genetics and classification. Clinical Genetics, 2014, 85, 203-212. | 2.0 | 131 |
| 3 | Multiple Familial Trichoepithelioma Caused by Mutations in the Cylindromatosis Tumor Suppressor Gene. Cancer Research, 2004, 64, 5113-5117. | 0.9 | 70 |
| 4 | Synpolydactyly: clinical and molecular advances. Clinical Genetics, 2008, 73, 113-120. | 2.0 | 58 |
| 5 | Prevalence of multi-drug resistant uropathogenic Escherichia coli in Potohar region of Pakistan. Asian Pacific Journal of Tropical Biomedicine, 2016, 6, 60-66. | 1.2 | 44 |
| 6 | Human GLI3 Intragenic Conserved Non-Coding Sequences Are Tissue-Specific Enhancers. PLoS ONE, 2007, 2, e366. | 2.5 | 39 |
| 7 | Exome Sequencing and Rare Variant Analysis RevealsÂMultiple Filaggrin Mutations in BangladeshiÂFamilies with Atopic Eczema andÂAdditional Risk Genes. Journal of Investigative Dermatology, 2018, 138, 2674-2677. | 0.7 | 37 |
| 8 | Ultraconserved nonâ€coding sequence element controls a subset of spatiotemporal GLI3 expression. Development Growth and Differentiation, 2007, 49, 543-553. | 1.5 | 35 |
| 9 | Clinical and descriptive genetic study of polydactyly: a Pakistani experience of 313 cases. Clinical Genetics, 2014, 85, 482-486. | 2.0 | 33 |
| 10 | Human intronic enhancers control distinct sub-domains of Gli3 expression during mouse CNS and limb development. BMC Developmental Biology, 2010, 10, 44. | 2.1 | 32 |
| 11 | A locus for hereditary hypotrichosis localized to human chromosome 18q21.1. European Journal of Human Genetics, 2003, 11, 623-628. | 2.8 | 30 |
| 12 | 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 |
| 13 | Homozygous mutation inCEP19,a gene mutated in morbid obesity, in Bardet-Biedl syndrome with predominant postaxial polydactyly. Journal of Medical Genetics, 2018, 55, 189-197. | 3.2 | 25 |
| 14 | Genetic heterogeneity of synpolydactyly: a novel locus SPD3 maps to chromosome 14q11.2-q12. Clinical Genetics, 2006, 69, 518-524. | 2.0 | 23 |
| 15 | 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 |
| 16 | Evidence for clinical and genetic heterogeneity of syndactyly type I: the phenotype of second and third toe syndactyly maps to chromosome 3p21.31. European Journal of Human Genetics, 2005, 13, 1268-1274. | 2.8 | 22 |
| 17 | Autosomal recessive mesoaxial synostotic syndactyly with phalangeal reduction maps to chromosome 17p13.3. American Journal of Medical Genetics, Part A, 2005, 134A, 404-408. | 1.2 | 20 |
| 18 | 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 |

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|---|---|---|---|
| 19 | Progressive SCAR14 with unclear speech, developmental delay, tremor, and behavioral problems caused by a homozygous deletion of the SPTBN2 pleckstrin homology domain. American Journal of Medical Genetics, Part A, 2017, 173, 2494-2499. | 1.2 | 19 |
| 20 | A novel type of autosomal recessive syndactyly: Clinical and molecular studies in a family of Pakistani origin. American Journal of Medical Genetics Part A, 2004, 126A, 61-67. | 2.4 | 18 |
| 21 | Synpolydactyly and HOXD13 polyalanine repeat: addition of 2 alanine residues is without clinical consequences. BMC Medical Genetics, 2007, 8, 78. | 2.1 | 17 |
| 22 | Congenital, low penetrance lymphedema of lower limbs maps to chromosome 6q16.2â€“q22.1 in an inbred Pakistani family. Human Genetics, 2008, 123, 197-205. | 3.8 | 16 |
| 23 | A novel ZRS variant causes preaxial polydactyly type I by increased sonic hedgehog expression in the developing limb bud. Genetics in Medicine, 2020, 22, 189-198. | 2.4 | 16 |
| 24 | PATTERN OF CONSANGUINITY AND INBREEDING COEFFICIENT IN SARGODHA DISTRICT, PUNJAB, PAKISTAN. Journal of Biosocial Science, 2015, 47, 803-811. | 1.2 | 15 |
| 25 | Consanguinity and its socio-biological parameters in Rahim Yar Khan District, Southern Punjab, Pakistan. Journal of Health, Population and Nutrition, 2016, 35, 14. | 2.0 | 14 |
| 26 | Biallelic loss-of-function variants in NEMF cause central nervous system impairment and axonal polyneuropathy. Human Genetics, 2021, 140, 579-592. | 3.8 | 14 |
| 27 | A novel ZRS mutation in a Balochi tribal family with triphalangeal thumb, preâ€axial polydactyly, postâ€axial polydactyly, and syndactyly. American Journal of Medical Genetics, Part A, 2012, 158A, 2031-2035. | 1.2 | 13 |
| 28 | Novel splice mutation in LRP4 causes severe type of Cenani-Lenz syndactyly syndrome with oro-facial and skeletal symptoms. European Journal of Medical Genetics, 2017, 60, 421-425. | 1.3 | 13 |
| 29 | Fifth finger camptodactyly maps to chromosome 3q11.2â€“q13.12 in a large German kindred. European Journal of Human Genetics, 2008, 16, 265-269. | 2.8 | 12 |
| 30 | CONSANGUINITY AND INBREEDING COEFFICIENT IN TRIBAL PASHTUNS INHABITING THE TURBULENT AND WAR-AFFECTED TERRITORY OF BAJAUR AGENCY, NORTH-WEST PAKISTAN. Journal of Biosocial Science, 2016, 48, 113-128. | 1.2 | 12 |
| 31 | Homozygous CHST11 mutation in chondrodysplasia, brachydactyly, overriding digits, clino-symphalangism and synpolydactyly. Journal of Medical Genetics, 2018, 55, 489-496. | 3.2 | 12 |
| 32 | Mutation in protein disulfide isomerase A3 causes neurodevelopmental defects by disturbing endoplasmic reticulum proteostasis. EMBO Journal, 2022, 41, e105531. | 7.8 | 11 |
| 33 | Linked homozygous BMPR1B and PDHA2 variants in a consanguineous family with complex digit malformation and male infertility. European Journal of Human Genetics, 2018, 26, 876-885. | 2.8 | 10 |
| 34 | 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 |
| 35 | CRADD and USP44 mutations in intellectual disability, mild lissencephaly, brain atrophy, developmental delay, strabismus, behavioural problems and skeletal anomalies. European Journal of Medical Genetics, 2021, 64, 104181. | 1.3 | 8 |
| 36 | TRANSITION IN CONSANGUINITY IN DIR LOWER DISTRICT, A VICTIM OF WAR, NATURAL DISASTER AND POPULATION DISPLACEMENT, IN NORTH-WEST PAKISTAN â€“ A RESPONSE TO STHANADAR ET AL. (2015). Journal of Biosocial Science, 2016, 48, 421-426. | 1.2 | 7 |

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|---|---|---|---|
| 37 | A Novel Locus of Ectodermal Dysplasia Maps to Chromosome 10q24.32â€“q25.1. Journal of Investigative Dermatology, 2005, 124, 338-342. | 0.7 | 6 |
| 38 | Complex postaxial polydactyly types A and B with camptodactyly, hypoplastic third toe, zygodactyly and other digit anomalies caused by a novel GLI3 mutation. European Journal of Medical Genetics, 2017, 60, 268-274. | 1.3 | 6 |
| 39 | Novel EDAR mutation in tooth agenesis and variable associated features. European Journal of Medical Genetics, 2020, 63, 103926. | 1.3 | 6 |
| 40 | ABO and Rh (D) Blood Groups Polymorphism in Four Tehsils of Bajaur Agency (Federally Administered) Tj ETQq0 0 0 rgBT /Overlock 10 T | 0.1 | 5 |
| 41 | RBBP8 syndrome with microcephaly, intellectual disability, short stature and brachydactyly. American Journal of Medical Genetics, Part A, 2015, 167, 3148-3152. | 1.2 | 5 |
| 42 | Biallelic ZNF407 mutations in a neurodevelopmental disorder with ID, short stature and variable microcephaly, hypotonia, ocular anomalies and facial dysmorphism. Journal of Human Genetics, 2020, 65, 1115-1123. | 2.3 | 5 |
| 43 | 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 |
| 44 | Seroprevalence of HDV among non-hospitalized HBsAg positive patients from KPK-region of Pakistan. Asian Pacific Journal of Tropical Biomedicine, 2016, 6, 609-613. | 1.2 | 4 |
| 45 | The first adolescent case of Fraser syndrome 3, with a novel nonsense variant in GRIP1. American Journal of Medical Genetics, Part A, 2021, 185, 1858-1863. | 1.2 | 4 |
| 46 | 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. | 0.7 | 4 |
| 47 | Zygodactyly with thumb aplasia: an unusual variant in a male subject. Journal of the College of Physicians and Surgeons--Pakistan: JCPSP, 2011, 21, 710-2. | 0.4 | 4 |
| 48 | KERATIN 17-related recessive atypical pachyonychia congenita with variable hair and tooth anomalies. European Journal of Human Genetics, 2022, 30, 1292-1296. | 2.8 | 4 |
| 49 | A splice-site variant (c.3289-1G>T) in OTOF underlies profound hearing loss in a Pakistani kindred. BMC Medical Genomics, 2021, 14, 2. | 1.5 | 3 |
| 50 | Consanguinity, inbreeding coefficient, fertility andÂbirth-outcome in population of Okara district, Pakistan. Pakistan Journal of Medical Sciences, 2021, 37, 770-775. | 0.6 | 3 |
| 51 | Determinants of consanguinity and inbreeding coefficient in the multiethnic population of Mardan, Khyber Pakhtunkhwa, Pakistan. Asian Biomedicine, 2017, 11, 451-460. | 0.3 | 3 |
| 52 | 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 |
| 53 | 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 |
| 54 | 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 |

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| 55 | 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 |
| 56 | Study of non-syndromic thumb aplasia in six independent cases.. Pakistan Journal of Medical Sciences, 1969, 30, 677-81. | 0.6 | 2 |
| 57 | Mesoaxial synostotic syndactyly with phalangeal reduction (MSSD): syndactyly type IX. Skeletal Radiology, 2018, 47, 149-149. | 2.0 | 2 |
| 58 | Homozygous deletion of MYADML2 in cranial asymmetry, reduced bone maturation, multiple dislocations, lumbar lordosis, and prominent clavicles. Journal of Human Genetics, 2021, 66, 171-179. | 2.3 | 2 |
| 59 | Prevalence-pattern and risk factors ofÂCesarean sectionÂinÂaÂmultiethnic cohort. Pakistan Journal of Medical Sciences, 2021, 37, 711-715. | 0.6 | 2 |
| 60 | Epidemiological study of congenital and hereditary anomalies in Sialkot District of Pakistan revealed a high incidence of limb and neurological disorders. Asian Biomedicine, 2019, 13, 49-60. | 0.3 | 2 |
| 61 | Talipes equinovarus or Clubfoot: A review of study approaches, management and trends in Pakistan. Pakistan Journal of Medical Sciences, 2020, 36, 1414-1420. | 0.6 | 2 |
| 62 | 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 |
| 63 | Autosomal dominant syndrome of camptodactyly, clinodactyly, syndactyly, and bifid toes. American Journal of Medical Genetics, Part A, 2010, 152A, 2313-2317. | 1.2 | 1 |
| 64 | 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 |
| 65 | Genetic Diversity at ABO and Rh (D) Loci in the Tribal Groups of Mohmand Agency (Federally) Tj ETQq1 1 0.784314 rgBT /Overlock 10 | 0.1 | 1 |
| 66 | Distribution ofABOandRh(D)Allelic Polymorphisms North Waziristan Agency, (Federally) Tj ETQq0 0 0 rgBT /Overlock 10 | 0.1 | 1 |
| 67 | A homozygous ROR2 variant in a family with atypical Robinow syndrome and tetramelic transverse deficiency of autopods. American Journal of Medical Genetics, Part A, 2021, , . | 1.2 | 1 |
| 68 | Analysis of Genetic Differentiation at ABO and Rh Loci among the Pashtun Populations Inhabiting Lower Khyber Pakhtunkhwa, Pakistan. Pakistan Journal of Zoology, 2017, 49, 387-389. | 0.2 | 1 |
| 69 | 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 |
| 70 | Novel Polymorphism in the Promoter Region of HLA-DQB1 Is a Predictor of Anti-HCV Therapy Response. Jundishapur Journal of Microbiology, 2019, 12, . | 0.5 | 1 |
| 71 | Phenotypic manifestation of congenital transverse amputation of autopod in Pakistani subjects. Pakistan Journal of Medical Sciences, 1969, 32, 519-22. | 0.6 | 0 |
| 72 | 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 |

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|---|---|---|---|
| 73 | A founder RDH5 splice site mutation leads to retinitis punctata albescens in two inbred Pakistani kindreds. Ophthalmic Genetics, 2020, 41, 7-12. | 1.2 | 0 |
| 74 | Congenital limb defects in a married female population of the Rahim Yar Khan District in Pakistan. Asian Biomedicine, 2021, 15, 137-144. | 0.3 | 0 |
| 75 | Frequencies of ABO and Rh (D) Blood Group Phenotypes in Pashtuns of Northâ€Western Pakistan: A population undergoing huge demographic changes. International Journal of Immunogenetics, 2021, 48, 336-339. | 1.8 | 0 |
| 76 | 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 |