

Musharraf Jelani

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/4033692/publications.pdf>

Version: 2024-02-01

51
papers

713
citations

471061

17
h-index

580395

25
g-index

51
all docs

51
docs citations

51
times ranked

1383
citing authors

#	ARTICLE	IF	CITATIONS
1	Association of cytochromes P450 3A4*22 and 3A5*3 genotypes and polymorphism with response to simvastatin in hypercholesterolemia patients. <i>PLoS ONE</i> , 2022, 17, e0260824.	1.1	3
2	Whole exome sequencing identified a novel missense alteration in CC2D2A causing Joubert syndrome 9 in a Pakhtun family. <i>Journal of Gene Medicine</i> , 2021, 23, e3279.	1.4	2
3	Whole exome sequencing reveals a homozygous SCGB variant in a Pakhtun family with limb girdle muscular dystrophy (LGMDR4) phenotype. <i>Gene Reports</i> , 2021, 22, 101014.	0.4	0
4	Two missense mutations in GPNMB cause autosomal recessive amyloidosis cutis dyschromica in the consanguineous pakistani families. <i>Genes and Genomics</i> , 2021, 43, 471-478.	0.5	3
5	A novel variant in the DSE gene leads to Ehlers-Danlos musculocontractural type 2 in a Pakistani family. <i>Congenital Anomalies (discontinued)</i> , 2021, 61, 177-182.	0.3	3
6	Whole Exome Sequencing Confirms Molecular Diagnostics of Three Pakhtun Families With Autosomal Recessive Epidermolysis Bullosa. <i>Frontiers in Pediatrics</i> , 2021, 9, 727288.	0.9	2
7	Biallelic inheritance in a single Pakistani family with intellectual disability implicates new candidate gene RDH14. <i>Scientific Reports</i> , 2021, 11, 23113.	1.6	2
8	Novel missense alteration in <i>LRP4</i> gene underlies Cenani-Lenz syndactyly syndrome in a consanguineous family. <i>Journal of Gene Medicine</i> , 2020, 22, e3143.	1.4	5
9	Identification of a recurrent nonsense mutation in HR gene responsible for atrichia with papular lesions in two Kashmiri families. <i>Journal of Gene Medicine</i> , 2020, 22, e3167.	1.4	2
10	Genetic variations in drug-metabolizing enzyme CYP2C9 among major ethnic groups of Pakistani population. <i>Gene</i> , 2020, 746, 144659.	1.0	7
11	Novel compound heterozygous and homozygous variants of laminin subunit $\beta 3$ gene underlie non-Herlitz junctional epidermolysis bullosa in two paternal half-brothers from Saudi Arabia. <i>Congenital Anomalies (discontinued)</i> , 2019, 59, 99-101.	0.3	0
12	A recurrent missense mutation in the EDAR gene causes severe autosomal recessive hypohidrotic ectodermal dysplasia in two consanguineous Kashmiri families. <i>Journal of Gene Medicine</i> , 2019, 21, e3113.	1.4	2
13	Exome Analysis Identifies a Novel Compound Heterozygous Alteration in TGM1 Gene Leading to Lamellar Ichthyosis in a Child From Saudi Arabia: Case Presentation. <i>Frontiers in Pediatrics</i> , 2019, 7, 44.	0.9	7
14	A mutation in the major autophagy gene, WIPI2, associated with global developmental abnormalities. <i>Brain</i> , 2019, 142, 1242-1254.	3.7	28
15	Novel insertion and a previously reported nonsense variant of ALOXE3 gene lead to autosomal recessive ichthyosis in two Balochi families. <i>Congenital Anomalies (discontinued)</i> , 2019, 59, 179-180.	0.3	0
16	A missense mutation in TRAPPC6A leads to build-up of the protein, in patients with a neurodevelopmental syndrome and dysmorphic features. <i>Scientific Reports</i> , 2018, 8, 2053.	1.6	30
17	Subtractive genome analysis for in silico identification and characterization of novel drug targets in <i>Streptococcus pneumoniae</i> strain JJA. <i>Microbial Pathogenesis</i> , 2018, 115, 194-198.	1.3	37
18	Selective glycosidase inhibitors: A patent review (2012-present). <i>International Journal of Biological Macromolecules</i> , 2018, 111, 82-91.	3.6	38

#	ARTICLE	IF	CITATIONS
19	Whole-exome sequencing analysis reveals co-segregation of a COL20A1 missense mutation in a Pakistani family with striate palmoplantar keratoderma. <i>Genes and Genomics</i> , 2018, 40, 789-795.	0.5	5
20	Novel splice site mutation in <i>EIF2AK3</i> gene causes Wolcott-Rallison syndrome in a consanguineous family from Saudi Arabia. <i>Congenital Anomalies (discontinued)</i> , 2018, 58, 39-40.	0.3	3
21	The prevalence of APOL1 gene variants in a cohort of renal disease patients in Western Saudi Arabia. <i>Saudi Journal of Kidney Diseases and Transplantation: an Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia</i> , 2018, 29, 793.	0.4	1
22	Whole exome sequencing analysis identifies a missense variant in COL1A2 gene which causes osteogenesis imperfecta Type IV in a family from Saudi Arabia. <i>Journal of Musculoskeletal Surgery and Research</i> , 2017, 1, 33.	0.2	3
23	A novel homozygous PTH1R variant identified through whole-exome sequencing further expands the clinical spectrum of primary failure of tooth eruption in a consanguineous Saudi family. <i>Archives of Oral Biology</i> , 2016, 67, 28-33.	0.8	21
24	Whole-exome sequencing reveals a recurrent mutation in the cathepsin C gene that causes Papillon-Lefevre syndrome in a Saudi family. <i>Saudi Journal of Biological Sciences</i> , 2016, 23, 571-576.	1.8	6
25	Whole exome analysis reveals a novel missense PNPLA1 variant that causes autosomal recessive congenital ichthyosis in a Pakistani family. <i>Journal of Dermatological Science</i> , 2016, 82, 46-48.	1.0	14
26	The alkylglycerol monooxygenase (AGMO) gene previously involved in autism also causes a novel syndromic form of primary microcephaly in a consanguineous Saudi family. <i>Journal of the Neurological Sciences</i> , 2016, 363, 240-244.	0.3	18
27	A Novel Missense Mutation in the CLPP Gene Causing Perrault Syndrome Type 3 in a Turkish Family. <i>JCRPE Journal of Clinical Research in Pediatric Endocrinology</i> , 2016, 8, 472-477.	0.4	19
28	Prognostic Stratification of Acute Myeloid Leukemia and Myelodysplastic Syndrome Patients on the Basis of Genetic Variations. <i>Blood</i> , 2016, 128, 5239-5239.	0.6	1
29	Whole-exome sequencing identifies a novel LRAT mutation underlying retinitis punctata albescens in a consanguineous Pakistani family. <i>Genes and Genomics</i> , 2015, 37, 845-849.	0.5	3
30	Case of Sjögren-Larsson syndrome with a large deletion in the <i>ALDH3A2</i> gene confirmed by single nucleotide polymorphism array analysis. <i>Journal of Dermatology</i> , 2015, 42, 706-709.	0.6	16
31	Identification of Two Homozygous Sequence Variants in the <i>COL7A1</i> Gene Underlying Dystrophic Epidermolysis Bullosa by Whole-Exome Analysis in a Consanguineous Family. <i>Annals of Human Genetics</i> , 2015, 79, 350-356.	0.3	3
32	Familial Primary Localized Cutaneous Amyloidosis Results from Either Dominant or Recessive Mutations in OSMR. <i>Acta Dermato-Venereologica</i> , 2015, 95, 1005-1007.	0.6	10
33	Novel nonsense mutation in the PTRF gene underlies congenital generalized lipodystrophy in a consanguineous Saudi family. <i>European Journal of Medical Genetics</i> , 2015, 58, 216-221.	0.7	31
34	Human semen quality and sperm DNA damage assessed by comet assay in clinical groups. <i>Turkish Journal of Medical Sciences</i> , 2015, 45, 729-737.	0.4	10
35	Exome analysis identified a novel missense mutation in the CLPP gene in a consanguineous Saudi family expanding the clinical spectrum of Perrault Syndrome type-3. <i>Journal of the Neurological Sciences</i> , 2015, 353, 149-154.	0.3	37
36	Truncating mutation in intracellular phospholipase A1 gene (DDHD2) in hereditary spastic paraplegia with intellectual disability (SPG54). <i>BMC Research Notes</i> , 2015, 8, 271.	0.6	17

#	ARTICLE	IF	CITATIONS
37	Insight into the serum kisspeptin levels in infertile males. Archives of Iranian Medicine, 2015, 18, 12-7.	0.2	29
38	Deletion mutation in BSCL2 gene underlies congenital generalized lipodystrophy in a Pakistani family. Diagnostic Pathology, 2013, 8, 78.	0.9	17
39	Congenital cutis laxa syndrome maps to a novel locus on chromosome 9q13-q21.32. Journal of Dermatological Science, 2011, 61, 134-136.	1.0	0
40	Digenic inheritance of an autosomal recessive hypotrichosis in two consanguineous pedigrees. Clinical Genetics, 2011, 79, 273-281.	1.0	14
41	Novel Autosomal Recessive Nonsyndromic Hearing Impairment Locus DFNB90 Maps to 7p22.1-p15.3. Human Heredity, 2011, 71, 106-112.	0.4	4
42	Mutation in PVRL4 gene encoding nectin-4 underlies ectodermal-dysplasia-syndactyly syndrome (EDSS1). Journal of Human Genetics, 2011, 56, 352-357.	1.1	27
43	Mutation Analysis of the <i>ASPM</i> Gene in 18 Pakistani Families With Autosomal Recessive Primary Microcephaly. Journal of Child Neurology, 2010, 25, 715-720.	0.7	18
44	Mutations in the <i>P2RY5</i> gene underlie autosomal recessive hypotrichosis in 13 Pakistani families. British Journal of Dermatology, 2009, 160, 1006-1010.	1.4	20
45	A novel splice-site mutation in the <i>CDH3</i> gene in hypotrichosis with juvenile macular dystrophy. Clinical and Experimental Dermatology, 2009, 34, 68-73.	0.6	25
46	A novel deletion mutation in the human hairless (<i>HR</i>) gene in an Iranian family with atrichia and papular lesions. Clinical and Experimental Dermatology, 2009, 34, e498-e500.	0.6	4
47	A Homozygous Nonsense Mutation in the Human Desmocollin-3 (<i>DSC3</i>) Gene Underlies Hereditary Hypotrichosis and Recurrent Skin Vesicles. American Journal of Human Genetics, 2009, 85, 515-520.	2.6	75
48	Novel mutations in G protein-coupled receptor gene (<i>P2RY5</i>) in families with autosomal recessive hypotrichosis (LAH3). Human Genetics, 2008, 123, 515-519.	1.8	37
49	A novel deletion mutation in <i>LIPH</i> gene causes autosomal recessive hypotrichosis (LAH2). Clinical Genetics, 2008, 74, 184-188.	1.0	28
50	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
51	Whole exome sequencing identifies a novel compound heterozygous <i>CFM1</i> variant underlying developmental delay, dystonia, polymicrogyria, and severe intellectual disability in a Pakhtun family. American Journal of Medical Genetics, Part A, 0, , .	0.7	3