Musharraf Jelani

List of Publications by Year
in descending order

51	713	471061	580395
papers	citations	17 h-index	25 g-index

51	51	51	1383
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	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
2	Selective glycosidase inhibitors: A patent review (2012â€“present). International Journal of Biological Macromolecules, 2018, 111, 82-91.	3.6	38
3	Novel mutations in G protein-coupled receptor gene (P2RY5) in families with autosomal recessive hypotrichosis (LAH3). Human Genetics, 2008, 123, 515-519.	1.8	37
4	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. Journal of the Neurological Sciences, 2015, 353, 149-154.	0.3	37
5	Subtractive genome analysis for in silico identification and characterization of novel drug targets in Streptococcus pneumonia strain JJA. Microbial Pathogenesis, 2018, 115, 194-198.	1.3	37
6	Novel nonsense mutation in the PTRF gene underlies congenital generalized lipodystrophy in a consanguineous Saudi family. European Journal of Medical Genetics, 2015, 58, 216-221.	0.7	31
7	A missense mutation in TRAPPC6A leads to build-up of the protein, in patients with a neurodevelopmental syndrome and dysmorphic features. Scientific Reports, 2018, 8, 2053.	1.6	30
8	Insight into the serum kisspeptin levels in infertile males. Archives of Iranian Medicine, 2015, 18, 12-7.	0.2	29
9	A novel deletion mutation in LIPH gene causes autosomal recessive hypotrichosis (LAH2). Clinical Genetics, 2008, 74, 184-188.	1.0	28
10	A mutation in the major autophagy gene, WIPI2, associated with global developmental abnormalities. Brain, 2019, 142, 1242-1254.	3.7	28
11	Mutation in PVRL4 gene encoding nectin-4 underlies ectodermal-dysplasia-syndactyly syndrome (EDSS1). Journal of Human Genetics, 2011, 56, 352-357.	1.1	27
12	A novel splice-site mutation in theCDH3gene in hypotrichosis with juvenile macular dystrophy. Clinical and Experimental Dermatology, 2009, 34, 68-73.	0.6	25
13	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
14	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. Archives of Oral Biology, 2016, 67, 28-33.	0.8	21
15	Mutations in theP2RY5gene underlie autosomal recessive hypotrichosis in 13 Pakistani families. British Journal of Dermatology, 2009, 160, 1006-1010.	1.4	20
16	A Novel Missense Mutation in the CLPP Gene Causing Perrault Syndrome Type 3 in a Turkish Family. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2016, 8, 472-477.	0.4	19
17	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
18	The alkylglycerol monooxygenase (AGMO) gene previously involved in autism also causes a novel syndromic form of primary microcephaly in a consanguineous Saudi family. Journal of the Neurological Sciences, 2016, 363, 240-244.	0.3	18

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19	Deletion mutation in BSCL2 gene underlies congenital generalized lipodystrophy in a Pakistani family. Diagnostic Pathology, 2013, 8, 78.	0.9	17
20	Truncating mutation in intracellular phospholipase A1 gene (DDHD2) in hereditary spastic paraplegia with intellectual disability (SPG54). BMC Research Notes, 2015, 8, 271.	0.6	17
21	Case of SjÃ¶grenâ€“Larsson syndrome with a large deletion in the ALDH3A2 gene confirmed by single nucleotide polymorphism array analysis. Journal of Dermatology, 2015, 42, 706-709.	0.6	16
22	Digenic inheritance of an autosomal recessive hypotrichosis in two consanguineous pedigrees. Clinical Genetics, 2011, 79, 273-281.	1.0	14
23	Whole exome analysis reveals a novel missense PNPLA1 variant that causes autosomal recessive congenital ichthyosis in a Pakistani family. Journal of Dermatological Science, 2016, 82, 46-48.	1.0	14
24	Familial Primary Localized Cutaneous Amyloidosis Results from Either Dominant or Recessive Mutations in OSMR. Acta Dermato-Venereologica, 2015, 95, 1005-1007.	0.6	10
25	Human semen quality and sperm DNA damage assessed bycomet assay in clinical groups. Turkish Journal of Medical Sciences, 2015, 45, 729-737.	0.4	10
26	Exome Analysis Identifies a Novel Compound Heterozygous Alteration in TGM1 Gene Leading to Lamellar Ichthyosis in a Child From Saudi Arabia: Case Presentation. Frontiers in Pediatrics, 2019, 7, 44.	0.9	7
27	Genetic variations in drug-metabolizing enzyme CYP2C9 among major ethnic groups of Pakistani population. Gene, 2020, 746, 144659.	1.0	7
28	Whole-exome sequencing reveals a recurrent mutation in the cathepsin C gene that causes Papillonâ€“Lefevre syndrome in a Saudi family. Saudi Journal of Biological Sciences, 2016, 23, 571-576.	1.8	6
29	Whole-exome sequencing analysis reveals co-segregation of a COL20A1 missense mutation in a Pakistani family with striate palmoplantar keratoderma. Genes and Genomics, 2018, 40, 789-795.	0.5	5
30	Novel missense alteration in LRP4 gene underlies Cenaniâ€“Lenz syndactyly syndrome in a consanguineous family. Journal of Gene Medicine, 2020, 22, e3143.	1.4	5
31	A novel deletion mutation in the human hairless (HR) gene in an Iranian family with atrichia and papular lesions. Clinical and Experimental Dermatology, 2009, 34, e498-e500.	0.6	4
32	Novel Autosomal Recessive Nonsyndromic Hearing Impairment Locus DFNB90 Maps to 7p22.1-p15.3. Human Heredity, 2011, 71, 106-112.	0.4	4
33	Whole-exome sequencing identifies a novel LRAT mutation underlying retinitis punctata albescens in a consanguineous Pakistani family. Genes and Genomics, 2015, 37, 845-849.	0.5	3
34	Identification of Two Homozygous Sequence Variants in the COL7A1 Gene Underlying Dystrophic Epidermolysis Bullosa by Wholeâ€Exome Analysis in a Consanguineous Family. Annals of Human Genetics, 2015, 79, 350-356.	0.3	3
35	Novel splice site mutation in EIF2AK3 gene causes Wolcottâ€Rallison syndrome in a consanguineous family from Saudi Arabia. Congenital Anomalies (discontinued), 2018, 58, 39-40.	0.3	3
36	Two missense mutations in GPNMB cause autosomal recessive amyloidosis cutis dyschromica in the consanguineous pakistani families. Genes and Genomics, 2021, 43, 471-478.	0.5	3

#	Article	IF	Citations
37	A novel variant in the DSE gene leads to Ehlersâ€“Danlos musculocontractural type 2 in a Pakistani family. Congenital Anomalies (discontinued), 2021, 61, 177-182.	0.3	3
38	Whole exome sequencing analysis identifies a missense variant in COL1A2 gene which causes osteogenesis imperfecta Type IV in a family from Saudi Arabia. Journal of Musculoskeletal Surgery and Research, 2017, 1, 33.	0.2	3
39	Whole exome sequencing identifies a novel compound heterozygous GFM1 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
40	Association of cytochromes P450 3A4*22 and 3A5*3 genotypes and polymorphism with response to simvastatin in hypercholesterolemia patients. PLoS ONE, 2022, 17, e0260824.	1.1	3
41	A recurrent missense mutation in the EDAR gene causes severe autosomal recessive hypohidrotic ectodermal dysplasia in two consanguineous Kashmiri families. Journal of Gene Medicine, 2019, 21, e3113.	1.4	2
42	Identification of a recurrent nonsense mutation in HR gene responsible for atrichia with papular lesions in two Kashmiri families. Journal of Gene Medicine, 2020, 22, e3167.	1.4	2
43	Whole exome sequencing identified a novel missense alteration in CC2D2A causing Joubert syndrome 9 in a Pakhtun family. Journal of Gene Medicine, 2021, 23, e3279.	1.4	2
44	Whole Exome Sequencing Confirms Molecular Diagnostics of Three Pakhtun Families With Autosomal Recessive Epidermolysis Bullosa. Frontiers in Pediatrics, 2021, 9, 727288.	0.9	2
45	Biallelic inheritance in a single Pakistani family with intellectual disability implicates new candidate gene RDH14. Scientific Reports, 2021, 11, 23113.	1.6	2
46	The prevalence of APOL1 gene variants in a cohort of renal disease patients in Western Saudi Arabia. Saudi Journal of Kidney Diseases and Transplantation: an Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia, 2018, 29, 793.	0.4	1
47	Prognostic Stratification of Acute Myeloid Leukemia and Mylodysplastic Syndrome Patients on the Basis of Genetic Variations. Blood, 2016, 128, 5239-5239.	0.6	1
48	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
49	Novel compound heterozygous and homozygous variants of laminin subunit Î²3 gene underlie nonâ€Herlitz junctional epidermolysis bullosa in two paternal halfâ€brothers from Saudi Arabia. Congenital Anomalies (discontinued), 2019, 59, 99-101.	0.3	0
50	Novel insertion and a previously reported nonsense variant of ALOXE3 gene lead to autosomal recessive ichthyosis in two Balochi families. Congenital Anomalies (discontinued), 2019, 59, 179-180.	0.3	0
51	Whole exome sequencing reveals a homozygous SGCB variant in a Pakhtun family with limb girdle muscular dystrophy (LGMDR4) phenotype. Gene Reports, 2021, 22, 101014.	0.4	0