Salma M Wakil

List of Publications by Year
in descending order

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77 papers	2,323 citations	270111 25 h-index	263392 45 g-index
82 all docs	82 docs citations	82 times ranked	5946 citing authors

#	Article	IF	Citations
1	Telomerase reverse transcriptase promoter mutations in cancers derived from multiple organ sites among middle eastern population. Genomics, 2020, 112, 1746-1753.	1.3	10
2	New Insights into the Impact of Genome-Wide Copy Number Variations on Complex Congenital Heart Disease in Saudi Arabia. OMICS A Journal of Integrative Biology, 2020, 24, 16-28.	1.0	5
3	Comprehensive Genomic Analysis of Noonan Syndrome and Acute Myeloid Leukemia in Adults: A Review and Future Directions. Acta Haematologica, 2020, 143, 583-593.	0.7	4
4	Interleukin-8 Dedifferentiates Primary Human Luminal Cells to Multipotent Stem Cells. Molecular and Cellular Biology, 2020, 40, .	1.1	1
5	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
6	Truncating ARL6IP1 variant as the genetic cause of fatal complicated hereditary spastic paraplegia. BMC Medical Genetics, 2019, 20, 119.	2.1	14
7	A novel KIT mutation in a family with expanded syndrome of piebaldism. JAAD Case Reports, 2019, 5, 627-631.	0.4	4
8	Associations of autozygosity with a broad range of human phenotypes. Nature Communications, 2019, 10, 4957.	5.8	84
9	Combining CDKN1A gene expression and genome-wide SNPs in a twin cohort to gain insight into the heritability of individual radiosensitivity. Functional and Integrative Genomics, 2019, 19, 575-585.	1.4	9
10	Update on hereditary, autosomal dominant cathepsin-A-related arteriopathy with strokes and leukoencephalopathy (CARASAL). Acta Neurologica Belgica, 2019, 119, 299-303.	0.5	9
11	First report of two successive deletions on chromosome 15q13 cytogenetic bands in a boy and girl: additional data to 15q13.3 syndrome with a report of high IQ patient. Molecular Cytogenetics, 2019, 12, 21.	0.4	2
12	Familial, long-term pollakisuria as initial manifestation of HSP4 due to the SPAST variant c.683-2A>C. Journal of Clinical Neuroscience, 2019, 64, 4-5.	0.8	2
13	Pattern Recognition Receptor Polymorphisms as Predictors of Oxaliplatin Benefit in Colorectal Cancer. Journal of the National Cancer Institute, 2019, 111, 828-836.	3.0	10
14	Genetic investigation of 93 families with microphthalmia or posterior microphthalmos. Clinical Genetics, 2018, 93, 1210-1222.	1.0	38
15	Design of Arab Diabetes Gene-Centric Array (ADGCA) in population with an epidemic of Type 2 Diabetes: A population specific SNP evaluation. Gene, 2018, 663, 157-164.	1.0	5
16	Genomeâ€wide association study and metaâ€analysis in Northern European populations replicate multiple colorectal cancer risk loci. International Journal of Cancer, 2018, 142, 540-546.	2.3	26
17	Exome Sequencing: Mutilating Sensory Neuropathy with Spastic Paraplegia due to a Mutation in FAM134B Gene. Case Reports in Genetics, 2018, 2018, 1-5.	0.1	12
18	Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism. American Journal of Human Genetics, 2018, 103, 948-967.	2.6	18

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19	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
20	Replication of Type 2 diabetes-associated variants in a Saudi Arabian population. Physiological Genomics, 2018, 50, 296-297.	1.0	3
21	Identification of novel genomic imbalances in Saudi patients with congenital heart disease. Molecular Cytogenetics, 2018, 11, 9.	0.4	4
22	GZF1 Mutations Expand the Genetic Heterogeneity of Larsen Syndrome. American Journal of Human Genetics, 2017, 100, 831-836.	2.6	14
23	Mendelian randomisation implicates hyperlipidaemia as a risk factor for colorectal cancer. International Journal of Cancer, 2017, 140, 2701-2708.	2.3	76
24	Exome sequencing identifies novel NTRK1 mutations in patients with HSANâ€IV phenotype. American Journal of Medical Genetics, Part A, 2017, 173, 1009-1016.	0.7	20
25	Pro-inflammatory fatty acid profile and colorectal cancer risk: A Mendelian randomisation analysis. European Journal of Cancer, 2017, 84, 228-238.	1.3	81
26	A common variant association study in ethnic Saudi Arabs reveals novel susceptibility loci for hypertriglyceridemia. Clinical Genetics, 2017, 91, 371-378.	1.0	7
27	Clinical genomics expands the morbid genome of intellectual disability and offers a high diagnostic yield. Molecular Psychiatry, 2017, 22, 615-624.	4.1	187
28	Identification of a novel genetic locus underlying tremor and dystonia. Human Genomics, 2017, 11, 25.	1.4	6
29	Pregnancy reduces severity and frequency of attacks in hyperkalemic periodic paralysis due to the mutation c.2111C>T in the SCN4A gene. Annals of Indian Academy of Neurology, 2017, 20, 75.	0.2	3
30	A European Spectrum of Pharmacogenomic Biomarkers: Implications for Clinical Pharmacogenomics. PLoS ONE, 2016, 11, e0162866.	1.1	96
31	A common variant association study reveals novel susceptibility loci for low <scp>HDL</scp>â€cholesterol levels in ethnic Arabs. Clinical Genetics, 2016, 90, 518-525.	1.0	23
32	Mendelian randomisation analysis strongly implicates adiposity with risk of developing colorectal cancer. British Journal of Cancer, 2016, 115, 266-272.	2.9	57
33	Novel mutations in <scp>TGM</scp>1 and <scp>ABCA</scp>12 cause autosomal recessive congenital ichthyosis in five Saudi families. International Journal of Dermatology, 2016, 55, 673-679.	0.5	9
34	Novel copy number variants and major limb reduction malformation: Report of three cases. American Journal of Medical Genetics, Part A, 2016, 170, 1245-1250.	0.7	8
35	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
36	Mitochondrial implications in bulbospinal muscular atrophy (Kennedy disease). Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration, 2016, 17, 112-118.	1.1	6

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37	Stroke-like episodes, peri-episodic seizures, and MELAS mutations. European Journal of Paediatric Neurology, 2016, 20, 824-829.	0.7	18
38	A substitution mutation in cardiac ubiquitin ligase, FBXO32, is associated with an autosomal recessive form of dilated cardiomyopathy. BMC Medical Genetics, 2016, 17, 3.	2.1	19
39	Data on common variants associated with coronary artery disease/myocardial infarction in ethnic Arabs. Data in Brief, 2016, 7, 172-176.	0.5	0
40	Accelerating matchmaking of novel dysmorphology syndromes through clinical and genomic characterization of a large cohort. Genetics in Medicine, 2016, 18, 686-695.	1.1	55
41	New analytical application of antibody-based biosensor in estimation of thyroid-stimulating hormone in serum. Bioanalysis, 2016, 8, 625-632.	0.6	11
42	Mutations in SMG9, Encoding an Essential Component of Nonsense-Mediated Decay Machinery, Cause a Multiple Congenital Anomaly Syndrome in Humans and Mice. American Journal of Human Genetics, 2016, 98, 643-652.	2.6	51
43	Variation at 2q35 (PNKDandTMBIM1) influences colorectal cancer risk and identifies a pleiotropic effect with inflammatory bowel disease. Human Molecular Genetics, 2016, 25, 2349-2359.	1.4	37
44	Unbiased targeted next-generation sequencing molecular approach for primary immunodeficiency diseases. Journal of Allergy and Clinical Immunology, 2016, 137, 1780-1787.	1.5	115
45	A genome-wide association study reveals susceptibility loci for myocardial infarction/coronary artery disease in Saudi Arabs. Atherosclerosis, 2016, 245, 62-70.	0.4	61
46	The Affymetrix DMET Plus Platform Reveals Unique Distribution of ADME-Related Variants in Ethnic Arabs. Disease Markers, 2015, 2015, 1-8.	0.6	8
47	Association of a Mutation in LACC1 With a Monogenic Form of Systemic Juvenile Idiopathic Arthritis. Arthritis and Rheumatology, 2015, 67, 288-295.	2.9	111
48	Genotyping of CYP2C19 polymorphisms and its clinical validation in the ethnic Arab population. Journal of Pharmacy and Pharmacology, 2015, 67, 972-979.	1.2	1
49	A novelAPCmutation defines a second locus for Cenaniâ€“Lenz syndrome. Journal of Medical Genetics, 2015, 52, 317-321.	1.5	20
50	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
51	A new GWAS and meta-analysis with 1000Genomes imputation identifies novel risk variants for colorectal cancer. Scientific Reports, 2015, 5, 10442.	1.6	109
52	Positional mapping ofPRKD1,NRP1andPRDM1as novel candidate disease genes in truncus arteriosus. Journal of Medical Genetics, 2015, 52, 322-329.	1.5	30
53	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
54	The clinical utility of molecular karyotyping for neurocognitive phenotypes in a consanguineous population. Genetics in Medicine, 2015, 17, 719-725.	1.1	19

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55	Abnormalities of Skin and Cutaneous Appendages in Neuromuscular Disorders. Pediatric Neurology, 2015, 53, 301-308.	1.0	5
56	A New Susceptibility Locus for Myocardial Infarction, Hypertension, Type 2 Diabetes Mellitus, and Dyslipidemia on Chromosome 12q24. Disease Markers, 2014, 2014, 1-10.	0.6	17
57	Infantile-onset ascending hereditary spastic paraplegia with bulbar involvement due to the novel ALS2 mutation c.2761C > T. Gene, 2014, 536, 217-220.	1.0	23
58	Novel <scp>B4GALNT1</scp> mutations in a complicated form of hereditary spastic paraplegia. Clinical Genetics, 2014, 86, 500-501.	1.0	38
59	Clinical and pathological heterogeneity of a congenital disorder of glycosylation manifesting as a myasthenic/myopathic syndrome. Neuromuscular Disorders, 2014, 24, 353-359.	0.3	25
60	A study of the role of GATA2 gene polymorphism in coronary artery disease risk traits. Gene, 2014, 544, 152-158.	1.0	33
61	Clinical, Immunological and Molecular Characterization of DOCK8 and DOCK8-like Deficient Patients: Single Center Experience of Twenty Five Patients. Journal of Clinical Immunology, 2013, 33, 55-67.	2.0	81
62	New susceptibility locus for obesity and dyslipidaemia on chromosome 3q22.3. Human Genomics, 2013, 7, 15.	1.4	20
63	A novel splice site mutation in ERLIN2 causes hereditary spastic paraplegia in a Saudi family. European Journal of Medical Genetics, 2013, 56, 43-45.	0.7	21
64	Novel FGD1 mutation underlying Aarskogâ€“Scott syndrome with myopathy and distal arthropathy. Clinical Dysmorphology, 2013, 22, 13-17.	0.1	14
65	A study of the role of GATA4 polymorphism in cardiovascular metabolic disorders. Human Genomics, 2013, 7, 25.	1.4	9
66	Autozygome maps dispensable DNA and reveals potential selective bias against nullizygosity. Genetics in Medicine, 2012, 14, 515-519.	1.1	10
67	Novel homozygous mutation in DSP causing skin fragility-woolly hair syndrome: report of a large family and review of the desmoplakin-related phenotypes. Clinical Genetics, 2011, 80, 50-58.	1.0	27
68	Subcutaneous nodules, arthropathy, coarse face, cataract and glaucoma. Clinical Dysmorphology, 2011, 20, 50-52.	0.1	0
69	Genetics of type 2 diabetes in Arabs: What we know to date. International Journal of Diabetes Mellitus, 2009, 1, 32-34.	0.6	3
70	Identification of loci conferring risk for premature CAD and heterozygous familial hyperlipidemia in the LDLR, APOB and PCSK9 genes. International Journal of Diabetes Mellitus, 2009, 1, 16-21.	0.6	5
71	Syndromic congenital sensorineural deafness, microtia and microdontia resulting from a novel homoallelic mutation in fibroblast growth factor 3 (FGF3). European Journal of Human Genetics, 2009, 17, 14-21.	1.4	31
72	Genetic Study of Saudi Diabetes (GSSD): significant association of the KCNJ11 E23K polymorphism with type 2 diabetes. Diabetes/Metabolism Research and Reviews, 2008, 24, 137-140.	1.7	50

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73	Phenotypic Variation and Allelic Heterogeneity in Young Patients with Papillonâ€LefÃ¨vre Syndrome. Acta Dermato-Venereologica, 2005, -1, 1-1.	0.6	3
74	Mutation of the slow myosin heavy chain rod domain underlies hyaline body myopathy. Neurology, 2004, 62, 1518-1521.	1.5	78
75	Vertical transmission of hepatitis B virus despite maternal lamivudine therapy. Lancet, The, 2002, 359, 1488-1489.	6.3	64
76	Prevalence and profile of mutations associated with lamivudine therapy in Indian patients with chronic hepatitis B in the surface and polymerase genes of hepatitis B virus. Journal of Medical Virology, 2002, 68, 311-318.	2.5	45
77	Beneficial effects of lamivudine in hepatitis B virus-related decompensated cirrhosis. Journal of Hepatology, 2000, 33, 308-312.	1.8	152