

Mohammed Al Balwi

List of Publications by Citations

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

95
papers

2,259
citations

24
h-index

46
g-index

108
ext. papers

2,730
ext. citations

4.4
avg, IF

4.37
L-index

#	Paper	IF	Citations
95	Homozygosity for a missense mutation in SERPINH1, which encodes the collagen chaperone protein HSP47, results in severe recessive osteogenesis imperfecta. <i>American Journal of Human Genetics</i> , 2010 , 86, 389-98	11	253
94	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. <i>European Journal of Human Genetics</i> , 2017 , 25, 176-182	5.3	201
93	Histopathology of Middle East respiratory syndrome coronavirus (MERS-CoV) infection - clinicopathological and ultrastructural study. <i>Histopathology</i> , 2018 , 72, 516-524	7.3	193
92	Alteration of fatty-acid-metabolizing enzymes affects mitochondrial form and function in hereditary spastic paraplegia. <i>American Journal of Human Genetics</i> , 2012 , 91, 1051-64	11	150
91	The genetic basis of a craniofacial disease provides insight into COPII coat assembly. <i>Developmental Cell</i> , 2007 , 13, 623-634	10.2	147
90	Evidence for the role of PWCR1/HBII-85 C/D box small nucleolar RNAs in Prader-Willi syndrome. <i>American Journal of Human Genetics</i> , 2002 , 71, 669-78	11	104
89	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. <i>American Journal of Human Genetics</i> , 2019 , 104, 1182-1201	11	95
88	Biotin-responsive basal ganglia disease should be renamed biotin-thiamine-responsive basal ganglia disease: a retrospective review of the clinical, radiological and molecular findings of 18 new cases. <i>Orphanet Journal of Rare Diseases</i> , 2013 , 8, 83	4.2	90
87	Whole-genome sequencing offers additional but limited clinical utility compared with reanalysis of whole-exome sequencing. <i>Genetics in Medicine</i> , 2018 , 20, 1328-1333	8.1	73
86	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. <i>Nature Genetics</i> , 2017 , 49, 537-549	36.3	52
85	ARNT2 mutation causes hypopituitarism, post-natal microcephaly, visual and renal anomalies. <i>Brain</i> , 2013 , 136, 3096-105	11.2	48
84	Molecular breakpoint cloning and gene expression studies of a novel translocation t(4;15)(q27;q11.2) associated with Prader-Willi syndrome. <i>BMC Medical Genetics</i> , 2005 , 6, 18	2.1	48
83	Clinical, biochemical, cellular and molecular characterization of mitochondrial DNA depletion syndrome due to novel mutations in the MPV17 gene. <i>European Journal of Human Genetics</i> , 2014 , 22, 184-91	5.3	43
82	A multicenter clinical exome study in unselected cohorts from a consanguineous population of Saudi Arabia demonstrated a high diagnostic yield. <i>Molecular Genetics and Metabolism</i> , 2017 , 121, 91-95	3.7	41
81	Thirteen year retrospective review of the spectrum of inborn errors of metabolism presenting in a tertiary center in Saudi Arabia. <i>Orphanet Journal of Rare Diseases</i> , 2016 , 11, 126	4.2	40
80	Clinical and molecular characteristics of mitochondrial DNA depletion syndrome associated with neonatal cholestasis and liver failure. <i>Journal of Pediatrics</i> , 2014 , 164, 553-9.e1-2	3.6	34
79	Hereditary deletion of the entire FAM20C gene in a patient with Raine syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2013 , 161A, 3155-60	2.5	30

78	A novel mutation in the SHH long-range regulator (ZRS) is associated with preaxial polydactyly, triphalangeal thumb, and severe radial ray deficiency. <i>American Journal of Medical Genetics, Part A</i> , 2012 , 158A, 2610-5	2.5	29
77	Familial multiple sclerosis: does consanguinity have a role?. <i>Multiple Sclerosis Journal</i> , 2011 , 17, 487-9	5	28
76	Transaldolase deficiency: report of 12 new cases and further delineation of the phenotype. <i>Journal of Inherited Metabolic Disease</i> , 2013 , 36, 997-1004	5.4	26
75	HLA-A, -B, -C, -DRB1 and -DQB1 allele and haplotype frequencies in Saudis using next generation sequencing technique. <i>Tissue Antigens</i> , 2013 , 82, 252-8		26
74	Novel point mutations and mutational complexes in the enhancer II, core promoter and precore regions of hepatitis B virus genotype D1 associated with hepatocellular carcinoma in Saudi Arabia. <i>International Journal of Cancer</i> , 2013 , 133, 2864-71	7.5	25
73	A novel homozygous missense mutation (c.610G>A, p.Gly204Ser) in the WNT7A gene causes tetra-amelia in two Saudi families. <i>American Journal of Medical Genetics, Part A</i> , 2011 , 155A, 599-604	2.5	25
72	Mutation analysis of NPHS1 in a worldwide cohort of congenital nephrotic syndrome patients. <i>Nephron Clinical Practice</i> , 2012 , 120, c139-46		25
71	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. <i>Human Genetics</i> , 2017 , 136, 377-386	6.3	21
70	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. <i>PLoS ONE</i> , 2016 , 11, e0150555	3.7	21
69	Efficacy and safety of treatment of hepatitis C virus infection in renal transplant recipients. <i>World Journal of Gastroenterology</i> , 2012 , 18, 55-63	5.6	21
68	Liebenberg syndrome is caused by a deletion upstream to the PITX1 gene resulting in transformation of the upper limbs to reflect lower limb characteristics. <i>Gene</i> , 2013 , 524, 65-71	3.8	20
67	Genome-wide association study of chronic hepatitis B virus infection reveals a novel candidate risk allele on 11q22.3. <i>Journal of Medical Genetics</i> , 2013 , 50, 725-32	5.8	20
66	Isolation and characterization of a new naturally immortalized human breast carcinoma cell line, KAIMRC1. <i>BMC Cancer</i> , 2017 , 17, 803	4.8	19
65	Validity of two rapid point of care influenza tests and direct fluorescence assay in comparison of real time PCR for swine of origin Influenza virus. <i>Journal of Infection and Public Health</i> , 2011 , 4, 7-11	7.4	18
64	Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort. <i>European Journal of Human Genetics</i> , 2021 , 29, 141-153	5.3	18
63	De Novo Variants in TAOK1 Cause Neurodevelopmental Disorders. <i>American Journal of Human Genetics</i> , 2019 , 105, 213-220	11	16
62	X-linked ichthyosis associated with psychosis and behavioral abnormalities: a case report. <i>Journal of Medical Case Reports</i> , 2017 , 11, 267	1.2	14
61	HCV genotypes among 1013 Saudi nationals: a multicenter study. <i>Annals of Saudi Medicine</i> , 2013 , 33, 10-2	1.6	14

60	Congenital duplication of the palm syndrome. <i>Annals of Plastic Surgery</i> , 2007 , 59, 341-3	1.7	12
59	Hepatitis C virus genotypes in Saudi Arabia: a future prediction and laboratory profile. <i>Virology Journal</i> , 2017 , 14, 208	6.1	11
58	Clinical characteristics and genetic subtypes of Fanconi anemia in Saudi patients. <i>Cancer Genetics</i> , 2016 , 209, 171-6	2.3	10
57	Two novel homozygous missense mutations in the GDF5 gene cause brachydactyly type C. <i>American Journal of Medical Genetics, Part A</i> , 2015 , 167, 1621-6	2.5	9
56	Tetrasomy 18p: case report and review of literature. <i>The Application of Clinical Genetics</i> , 2018 , 11, 9-14	3.1	9
55	A report of two cases of Al-Awadi Raas-Rothschild syndrome (AARRS) supporting that "apparent" Phocomelia differentiates AARRS from Schinzel Phocomelia syndrome (SPS). <i>Gene</i> , 2013 , 527, 371-5	3.8	8
54	Association of SNPs rs6498169 and rs10984447 with multiple sclerosis in Saudi patients: a model of the usefulness of familial aggregates in identifying genetic linkage in a multifactorial disease. <i>Multiple Sclerosis Journal</i> , 2012 , 18, 1395-400	5	8
53	Evolving sequence mutations in the Middle East Respiratory Syndrome Coronavirus (MERS-CoV). <i>Journal of Infection and Public Health</i> , 2020 , 13, 1544-1550	7.4	8
52	Upper limb muscle overgrowth with hypoplasia of the index finger: a new over-growth syndrome caused by the somatic PIK3CA mutation c.3140A>G. <i>BMC Medical Genetics</i> , 2018 , 19, 158	2.1	8
51	Coexistence of chronic myeloid leukemia and diffuse large B-cell lymphoma with antecedent chronic lymphocytic leukemia: a case report and review of the literature. <i>Journal of Medical Case Reports</i> , 2018 , 12, 64	1.2	7
50	CD95-mediated apoptosis in Burkitt's lymphoma B-cells is associated with Pim-1 down-regulation. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2017 , 1863, 239-252	6.9	7
49	Prevalence of mixed hepatitis C virus (HCV) genotypes among recently diagnosed dialysis patients with HCV infection. <i>Saudi Journal of Kidney Diseases and Transplantation: an Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia</i> , 2011 , 22, 712-6	0.6	7
48	A novel homozygous mutation in the SLCO2A1 gene is associated with severe primary hypertrophic osteoarthropathy phenotype in a Saudi patient. <i>International Journal of Dermatology</i> , 2015 , 54, e233-5	1.7	6
47	Exome sequencing revealed DNA variants in NCOR1, IGF2BP1, SGLT2 and NEK11 as potential novel causes of ketotic hypoglycemia in children. <i>Scientific Reports</i> , 2020 , 10, 2114	4.9	6
46	Ibrutinib therapy is effective in B-cell prolymphocytic leukemia exhibiting MYC aberrations. <i>Leukemia and Lymphoma</i> , 2018 , 59, 739-742	1.9	6
45	Targeted SLC19A3 gene sequencing of 3000 Saudi newborn: a pilot study toward newborn screening. <i>Annals of Clinical and Translational Neurology</i> , 2019 , 6, 2097-2103	5.3	6
44	Combining exome/genome sequencing with data repository analysis reveals novel gene-disease associations for a wide range of genetic disorders. <i>Genetics in Medicine</i> , 2021 , 23, 1551-1568	8.1	6
43	HLA class II polymorphism in Saudi patients with multiple sclerosis. <i>Hla</i> , 2018 , 91, 17-22	1.9	6

42	KIF16B is a candidate gene for a novel autosomal-recessive intellectual disability syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018 , 176, 1602-1609	2.5	5
41	Mucopolipidosis II: first report from Saudi Arabia. <i>Annals of Saudi Medicine</i> , 2013 , 33, 382-6	1.6	5
40	Prevalence of BRCA1 and BRCA2 Mutations Among High-Risk Saudi Patients With Breast Cancer. <i>Journal of Global Oncology</i> , 2018 , 4, 1-9	2.6	5
39	Tracing the epidemic history of hepatitis C virus genotypes in Saudi Arabia. <i>Infection, Genetics and Evolution</i> , 2017 , 52, 82-88	4.5	4
38	Oxidative stress, caloric intake and outcomes of critically ill patients. <i>Clinical Nutrition ESPEN</i> , 2019 , 29, 103-111	1.3	4
37	Atypical influenza A(H1N1)pdm09 strains caused an influenza virus outbreak in Saudi Arabia during the 2009-2011 pandemic season. <i>Journal of Infection and Public Health</i> , 2019 , 12, 557-567	7.4	4
36	Screening for glucose-6-phosphate dehydrogenase deficiency in neonates: a comparison between cord and peripheral blood samples. <i>BMC Pediatrics</i> , 2017 , 17, 159	2.6	4
35	Successful transfer from insulin to oral sulfonylurea in a 3-year-old girl with a mutation in the KCNJ11 gene. <i>Annals of Saudi Medicine</i> , 2010 , 30, 162-4	1.6	4
34	Familial glucocorticoid deficiency in five Arab kindreds with homozygous point mutations of the ACTH receptor (MC2R): genotype and phenotype correlations. <i>Hormone Research in Paediatrics</i> , 2011 , 76, 165-71	3.3	4
33	What is the right sequencing approach? Solo VS extended family analysis in consanguineous populations. <i>BMC Medical Genomics</i> , 2020 , 13, 103	3.7	4
32	A classification system for split-hand/ foot malformation (SHFM): A proposal based on 3 pedigrees with WNT10B mutations. <i>European Journal of Medical Genetics</i> , 2020 , 63, 103738	2.6	4
31	Novel human pathological mutations. Gene symbol: WNT7A. Disease: ulnar and fibula absence, with severe limb deficiency. <i>Human Genetics</i> , 2009 , 125, 334	6.3	4
30	Permissive underfeeding, cytokine profiles and outcomes in critically ill patients. <i>PLoS ONE</i> , 2019 , 14, e0209669	3.7	3
29	82-P. <i>Human Immunology</i> , 2013 , 74, 108	2.3	3
28	Analysis of CCR5 gene polymorphisms in 321 healthy Saudis using Next Generation Sequencing. <i>Human Immunology</i> , 2017 , 78, 384-386	2.3	2
27	Late-onset multiple venous malformations confined to the upper limb: link to somatic mutations. <i>Journal of Hand Surgery: European Volume</i> , 2020 , 45, 1023-1027	1.4	2
26	A novel interstitial deletion of chromosome 2q21.1-q23.3: Case report and literature review. <i>Molecular Genetics & Genomic Medicine</i> , 2020 , 8, e1135	2.3	2
25	Proteomic and Molecular Assessment of the Common Saudi Variant in Gene Through Mesenchymal Stem Cells. <i>Frontiers in Cell and Developmental Biology</i> , 2019 , 7, 365	5.7	2

24	Molecular diagnosis of fragile X syndrome using methylation sensitive techniques in a cohort of patients with intellectual disability. <i>Pediatric Neurology</i> , 2014 , 50, 368-76	2.9	2
23	The unclassified variant: c.2044AD>G, p.T682A (het.) in exon 12 of the GLI3 gene in a patient with oral-facial-digital syndrome type II (Mohr syndrome) phenotype. <i>Gene</i> , 2013 , 526, 471-3	3.8	2
22	Distinctive Features of a Saudi Genome		2
21	The effect of the VKORC1 promoter variant on warfarin responsiveness in the Saudi Warfarin Pharmacogenetic (SWAP) cohort. <i>Scientific Reports</i> , 2020 , 10, 11613	4.9	2
20	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. <i>Frontiers in Pediatrics</i> , 2021 , 9, 633385	3.4	2
19	Isolation and Establishment of a Highly Proliferative, Cancer Stem Cell-Like, and Naturally Immortalized Triple-Negative Breast Cancer Cell Line, KAIMRC2. <i>Cells</i> , 2021 , 10,	7.9	2
18	Clinical course of myeloproliferative leukaemia virus oncogene (MPL) mutation-associated familial thrombocytosis: a review of 64 paediatric and adult patients. <i>British Journal of Haematology</i> , 2021 , 194, 893-898	4.5	2
17	Differential Gene Expression in Peripheral White Blood Cells with Permissive Underfeeding and Standard Feeding in Critically Ill Patients: A Descriptive Sub-study of the PerMIT Randomized Controlled Trial. <i>Scientific Reports</i> , 2018 , 8, 17984	4.9	2
16	Gene symbol: LMX1B. Disease: Nail-Patella syndrome. <i>Human Genetics</i> , 2008 , 123, 109-10	6.3	2
15	A novel gene in early childhood diabetes: EDEM2 silencing decreases SLC2A2 and PXD1 expression, leading to impaired insulin secretion. <i>Molecular Genetics and Genomics</i> , 2020 , 295, 1253-1262	3.1	1
14	Homozygous R396H mutation of the RAG1 gene in a Saudi infant with Omenn syndrome: a case report. <i>Cases Journal</i> , 2009 , 2, 8391		1
13	Ichthyosis Prematurity Syndrome: A Rare Form but Easily Recognizable Ichthyosis. <i>Case Reports in Dermatology</i> , 2021 , 13, 470-473	1.1	1
12	Generation of induced pluripotent stem cell Line KAIMRCi001-A by reprogramming erythroid progenitors from peripheral blood of a healthy Saudi donor. <i>Stem Cell Research</i> , 2021 , 56, 102548	1.6	1
11	Genetic Profile of Epidermolysis Bullosa Cases in King Abdulaziz Medical City, Riyadh, Saudi Arabia.. <i>Frontiers in Genetics</i> , 2021 , 12, 753229	4.5	0
10	CRISPR/Cas9 Knockout Stimulates the Insulin Secretion Pathway Leading to Excessive Insulin Secretion. <i>Frontiers in Endocrinology</i> , 2021 , 12, 657873	5.7	0
9	Prevalence of Associated Endocrine Diseases in Patients with Neurofibromatosis Type 1.. <i>Avicenna Journal of Medicine</i> , 2022 , 12, 16-20	1.1	0
8	Peeling of skin as presenting manifestation in congenital disorders of glycosylation. <i>Journal of Dermatology</i> , 2020 , 47, e335-e336	1.6	
7	Novel frameshift mutations in ADAMTS13 in two families with hereditary thrombotic thrombocytopenic purpura. <i>Pediatric Blood and Cancer</i> , 2013 , 60, 1559-60	3	

- 6 PRF1 gene mutation in a Saudi patient with haemophagocytic lymphohistiocytosis. *British Journal of Biomedical Science*, **2010**, 67, 88-9 1.6
- 5 Novel human pathological mutations. Gene symbol: SLC3A1. Disease: Cystinuria. *Human Genetics*, **2009**, 126, 330 6.3
- 4 Multi-colour FISH analysis of gene expression in formalin-fixed paraffin wax-embedded tissue. *Technical Tips Online*, **1996**, 1, 4-6
- 3 Successful Engraftment In Children With Severe Aplastic Anemia After Transplantation Of HLA-Identical Hematopoietic Donor Cells With Shorter Telomere Length. *Blood*, **2013**, 122, 2047-2047 2.2
- 2 Novel human pathological mutations. Gene symbol: MAN2B1. Disease: Mannosidosis, alpha. *Human Genetics*, **2010**, 127, 122 6.3
- 1 Novel human pathological mutations. Gene symbol: EDAR. Disease: Ectodermal dysplasia, hypohidrotic. *Human Genetics*, **2010**, 127, 123 6.3