Mohammed Al Balwi

List of Publications by Year in Descending Order

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	Prevalence of Associated Endocrine Diseases in Patients with Neurofibromatosis Type 1.. *Avicenna Journal of Medicine*, **2022**, 12, 16-20	1.1	0
94	Genetic Profile of Epidermolysis Bullosa Cases in King Abdulaziz Medical City, Riyadh, Saudi Arabia.. *Frontiers in Genetics*, **2021**, 12, 753229	4.5	0
93	Combining exome/genome sequencing with data repository analysis reveals novel gene-disease associations for a wide range of genetic disorders. *Genetics in Medicine*, **2021**, 23, 1551-1568	8.1	6
92	The Leukodystrophy Spectrum in Saudi Arabia: Epidemiological, Clinical, Radiological, and Genetic Data. *Frontiers in Pediatrics*, **2021**, 9, 633385	3.4	2
91	Isolation and Establishment of a Highly Proliferative, Cancer Stem Cell-Like, and Naturally Immortalized Triple-Negative Breast Cancer Cell Line, KAIMRC2. *Cells*, **2021**, 10,	7.9	2
90	Clinical course of myeloproliferative leukaemia virus oncogene (MPL) mutation-associated familial thrombocytosis: a review of 64 paediatric and adult patients. *British Journal of Haematology*, **2021**, 194, 893-898	4.5	2
89	CRISPR/Cas9 Knockout Stimulates the Insulin Secretion Pathway Leading to Excessive Insulin Secretion. *Frontiers in Endocrinology*, **2021**, 12, 657873	5.7	0
88	Successful application of genome sequencing in a diagnostic setting: 1007 index cases from a clinically heterogeneous cohort. *European Journal of Human Genetics*, **2021**, 29, 141-153	5.3	18
87	Ichthyosis Prematurity Syndrome: A Rare Form but Easily Recognizable Ichthyosis. *Case Reports in Dermatology*, **2021**, 13, 470-473	1.1	1
86	Generation of induced pluripotent stem cell Line KAIMRCi001-A by reprogramming erythroid progenitors from peripheral blood of a healthy Saudi donor. *Stem Cell Research*, **2021**, 56, 102548	1.6	1
85	Late-onset multiple venous malformations confined to the upper limb: link to somatic mutations. *Journal of Hand Surgery: European Volume*, **2020**, 45, 1023-1027	1.4	2
84	Peeling of skin as presenting manifestation in congenital disorders of glycosylation. *Journal of Dermatology*, **2020**, 47, e335-e336	1.6	
83	A novel gene in early childhood diabetes: EDEM2 silencing decreases SLC2A2 and PXD1 expression, leading to impaired insulin secretion. *Molecular Genetics and Genomics*, **2020**, 295, 1253-1262	3.1	1
82	Exome sequencing revealed DNA variants in NCOR1, IGF2BP1, SGLT2 and NEK11 as potential novel causes of ketotic hypoglycemia in children. *Scientific Reports*, **2020**, 10, 2114	4.9	6
81	A novel interstitial deletion of chromosome 2q21.1-q23.3: Case report and literature review. *Molecular Genetics & Genomic Medicine*, **2020**, 8, e1135	2.3	2
80	Evolving sequence mutations in the Middle East Respiratory Syndrome Coronavirus (MERS-CoV). *Journal of Infection and Public Health*, **2020**, 13, 1544-1550	7.4	8
79	What is the right sequencing approach? Solo VS extended family analysis in consanguineous populations. *BMC Medical Genomics*, **2020**, 13, 103	3.7	4

78	The effect of the VKORC1 promoter variant on warfarin responsiveness in the Saudi WArfarin Pharmacogenetic (SWAP) cohort. *Scientific Reports*, **2020**, 10, 11613	4.9	2
77	A classification system for split-hand/ foot malformation (SHFM): A proposal based on 3 pedigrees with WNT10B mutations. *European Journal of Medical Genetics*, **2020**, 63, 103738	2.6	4
76	Oxidative stress, caloric intake and outcomes of critically ill patients. *Clinical Nutrition ESPEN*, **2019**, 29, 103-111	1.3	4
75	Permissive underfeeding, cytokine profiles and outcomes in critically ill patients. *PLoS ONE*, **2019**, 14, e0209669	3.7	3
74	Lessons Learned from Large-Scale, First-Tier Clinical Exome Sequencing in a Highly Consanguineous Population. *American Journal of Human Genetics*, **2019**, 104, 1182-1201	11	95
73	Atypical influenza A(H1N1)pdm09 strains caused an influenza virus outbreak in Saudi Arabia during the 2009-2011 pandemic season. *Journal of Infection and Public Health*, **2019**, 12, 557-567	7.4	4
72	Proteomic and Molecular Assessment of the Common Saudi Variant in Gene Through Mesenchymal Stem Cells. *Frontiers in Cell and Developmental Biology*, **2019**, 7, 365	5.7	2
71	De Novo Variants in TAOK1 Cause Neurodevelopmental Disorders. *American Journal of Human Genetics*, **2019**, 105, 213-220	11	16
70	Targeted SLC19A3 gene sequencing of 3000 Saudi newborn: a pilot study toward newborn screening. *Annals of Clinical and Translational Neurology*, **2019**, 6, 2097-2103	5.3	6
69	Whole-genome sequencing offers additional but limited clinical utility compared with reanalysis of whole-exome sequencing. *Genetics in Medicine*, **2018**, 20, 1328-1333	8.1	73
68	Ibrutinib therapy is effective in B-cell prolymphocytic leukemia exhibiting MYC aberrations. *Leukemia and Lymphoma*, **2018**, 59, 739-742	1.9	6
67	Histopathology of Middle East respiratory syndrome coronovirus (MERS-CoV) infection - clinicopathological and ultrastructural study. *Histopathology*, **2018**, 72, 516-524	7.3	193
66	Coexistence of chronic myeloid leukemia and diffuse large B-cell lymphoma with antecedent chronic lymphocytic leukemia: a case report and review of the literature. *Journal of Medical Case Reports*, **2018**, 12, 64	1.2	7
65	KIF16B is a candidate gene for a novel autosomal-recessive intellectual disability syndrome. *American Journal of Medical Genetics, Part A*, **2018**, 176, 1602-1609	2.5	5
64	HLA class II polymorphism in Saudi patients with multiple sclerosis. *Hla*, **2018**, 91, 17-22	1.9	6
63	Prevalence of BRCA1 and BRCA2 Mutations Among High-Risk Saudi Patients With Breast Cancer. *Journal of Global Oncology*, **2018**, 4, 1-9	2.6	5
62	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. *Scientific Reports*, **2018**, 8, 17984	4.9	2
61	Tetrasomy 18p: case report and review of literature. *The Application of Clinical Genetics*, **2018**, 11, 9-14	3.1	9

60	Upper limb muscle overgrowth with hypoplasia of the index finger: a new over-growth syndrome caused by the somatic PIK3CA mutation c.3140A>G. *BMC Medical Genetics*, **2018**, 19, 158	2.1	8
59	Analysis of CCR5 gene polymorphisms in 321 healthy Saudis using Next Generation Sequencing. *Human Immunology*, **2017**, 78, 384-386	2.3	2
58	Haploinsufficiency of the E3 ubiquitin-protein ligase gene TRIP12 causes intellectual disability with or without autism spectrum disorders, speech delay, and dysmorphic features. *Human Genetics*, **2017**, 136, 377-386	6.3	21
57	Mutations in DONSON disrupt replication fork stability and cause microcephalic dwarfism. *Nature Genetics*, **2017**, 49, 537-549	36.3	52
56	Tracing the epidemic history of hepatitis C virus genotypes in Saudi Arabia. *Infection, Genetics and Evolution*, **2017**, 52, 82-88	4.5	4
55	A multicenter clinical exome study in unselected cohorts from a consanguineous population of Saudi Arabia demonstrated a high diagnostic yield. *Molecular Genetics and Metabolism*, **2017**, 121, 91-95	3.7	41
54	Isolation and characterization of a new naturally immortalized human breast carcinoma cell line, KAIMRC1. *BMC Cancer*, **2017**, 17, 803	4.8	19
53	Screening for glucose-6-phosphate dehydrogenase deficiency in neonates: a comparison between cord and peripheral blood samples. *BMC Pediatrics*, **2017**, 17, 159	2.6	4
52	Hepatitis C virus genotypes in Saudi Arabia: a future prediction and laboratory profile. *Virology Journal*, **2017**, 14, 208	6.1	11
51	X-linked ichthyosis associated with psychosis and behavioral abnormalities: a case report. *Journal of Medical Case Reports*, **2017**, 11, 267	1.2	14
50	Clinical exome sequencing: results from 2819 samples reflecting 1000 families. *European Journal of Human Genetics*, **2017**, 25, 176-182	5.3	201
49	CD95-mediated apoptosis in Burkitt's lymphoma B-cells is associated with Pim-1 down-regulation. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2017**, 1863, 239-252	6.9	7
48	Clinical characteristics and genetic subtypes of Fanconi anemia in Saudi patients. *Cancer Genetics*, **2016**, 209, 171-6	2.3	10
47	Axial Spondylometaphyseal Dysplasia Is Caused by C21orf2 Mutations. *PLoS ONE*, **2016**, 11, e0150555	3.7	21
46	Thirteen year retrospective review of the spectrum of inborn errors of metabolism presenting in a tertiary center in Saudi Arabia. *Orphanet Journal of Rare Diseases*, **2016**, 11, 126	4.2	40
45	A novel homozygous mutation in the SLCO2A1 gene is associated with severe primary hypertrophic osteoarthropathy phenotype in a Saudi patient. *International Journal of Dermatology*, **2015**, 54, e233-5	1.7	6
44	Two novel homozygous missense mutations in the GDF5 gene cause brachydactyly type C. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1621-6	2.5	9
43	Molecular diagnosis of fragile X syndrome using methylation sensitive techniques in a cohort of patients with intellectual disability. *Pediatric Neurology*, **2014**, 50, 368-76	2.9	2

42	Clinical, biochemical, cellular and molecular characterization of mitochondrial DNA depletion syndrome due to novel mutations in the MPV17 gene. *European Journal of Human Genetics*, **2014**, 22, 184-91	5.3	43
41	Clinical and molecular characteristics of mitochondrial DNA depletion syndrome associated with neonatal cholestasis and liver failure. *Journal of Pediatrics*, **2014**, 164, 553-9.e1-2	3.6	34
40	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. *Orphanet Journal of Rare Diseases*, **2013**, 8, 83	4.2	90
39	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. *International Journal of Cancer*, **2013**, 133, 2864-71	7.5	25
38	Transaldolase deficiency: report of 12 new cases and further delineation of the phenotype. *Journal of Inherited Metabolic Disease*, **2013**, 36, 997-1004	5.4	26
37	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. *Gene*, **2013**, 526, 471-3	3.8	2
36	82-P. *Human Immunology*, **2013**, 74, 108	2.3	3
35	Liebenberg syndrome is caused by a deletion upstream to the PITX1 gene resulting in transformation of the upper limbs to reflect lower limb characteristics. *Gene*, **2013**, 524, 65-71	3.8	20
34	A report of two cases of Al-Awadi Raas-Rothschild syndrome (AARRS) supporting that "apparent" Phocomelia differentiates AARRS from Schinzel Phocomelia syndrome (SPS). *Gene*, **2013**, 527, 371-5	3.8	8
33	ARNT2 mutation causes hypopituitarism, post-natal microcephaly, visual and renal anomalies. *Brain*, **2013**, 136, 3096-105	11.2	48
32	Genome-wide association study of chronic hepatitis B virus infection reveals a novel candidate risk allele on 11q22.3. *Journal of Medical Genetics*, **2013**, 50, 725-32	5.8	20
31	Hereditary deletion of the entire FAM20C gene in a patient with Raine syndrome. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3155-60	2.5	30
30	HLA-A, -B, -C, -DRB1 and -DQB1 allele and haplotype frequencies in Saudis using next generation sequencing technique. *Tissue Antigens*, **2013**, 82, 252-8		26
29	Novel frameshift mutations in ADAMTS13 in two families with hereditary thrombotic thrombocytopenic purpura. *Pediatric Blood and Cancer*, **2013**, 60, 1559-60	3	
28	HCV genotypes among 1013 Saudi nationals: a multicenter study. *Annals of Saudi Medicine*, **2013**, 33, 10-2	1.6	14
27	Mucolipidosis II: first report from Saudi Arabia. *Annals of Saudi Medicine*, **2013**, 33, 382-6	1.6	5
26	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	
25	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. *Multiple Sclerosis Journal*, **2012**, 18, 1395-400	5	8

24	A novel mutation in the SHH long-range regulator (ZRS) is associated with preaxial polydactyly, triphalangeal thumb, and severe radial ray deficiency. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 2610-5	2.5	29
23	Alteration of fatty-acid-metabolizing enzymes affects mitochondrial form and function in hereditary spastic paraplegia. *American Journal of Human Genetics*, **2012**, 91, 1051-64	11	150
22	Mutation analysis of NPHS1 in a worldwide cohort of congenital nephrotic syndrome patients. *Nephron Clinical Practice*, **2012**, 120, c139-46		25
21	Efficacy and safety of treatment of hepatitis C virus infection in renal transplant recipients. *World Journal of Gastroenterology*, **2012**, 18, 55-63	5.6	21
20	Validity of two rapid point of care influenza tests and direct fluorecence assay in comparison of real time PCR for swine of origin Influenza virus. *Journal of Infection and Public Health*, **2011**, 4, 7-11	7.4	18
19	A novel homozygous missense mutation (c.610G>A, p.Gly204Ser) in the WNT7A gene causes tetra-amelia in two Saudi families. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 599-604	2.5	25
18	Familial glucocorticoid deficiency in five Arab kindreds with homozygous point mutations of the ACTH receptor (MC2R): genotype and phenotype correlations. *Hormone Research in Paediatrics*, **2011**, 76, 165-71	3.3	4
17	Familial multiple sclerosis: does consanguinity have a role?. *Multiple Sclerosis Journal*, **2011**, 17, 487-9	5	28
16	Prevalence of mixed hepatitis C virus (HCV) genotypes among recently diagnosed dialysis patients with HCV infection. *Saudi Journal of Kidney Diseases and Transplantation: an Official Publication of the Saudi Center for Organ Transplantation, Saudi Arabia*, **2011**, 22, 712-6	0.6	7
15	PRF1 gene mutation in a Saudi patient with haemophagocytic lymphohistiocytosis. *British Journal of Biomedical Science*, **2010**, 67, 88-9	1.6	
14	Successful transfer from insulin to oral sulfonylurea in a 3-year-old girl with a mutation in the KCNJ11 gene. *Annals of Saudi Medicine*, **2010**, 30, 162-4	1.6	4
13	Homozygosity for a missense mutation in SERPINH1, which encodes the collagen chaperone protein HSP47, results in severe recessive osteogenesis imperfecta. *American Journal of Human Genetics*, **2010**, 86, 389-98	11	253
12	Novel human pathological mutations. Gene symbol: MAN2B1. Disease: Mannosidosis, alpha. *Human Genetics*, **2010**, 127, 122	6.3	
11	Novel human pathological mutations. Gene symbol: EDAR. Disease: Ectodermal dysplasia, hypohidrotic. *Human Genetics*, **2010**, 127, 123	6.3	
10	Novel human pathological mutations. Gene symbol: SLC3A1. Disease: Cystinuria. *Human Genetics*, **2009**, 126, 330	6.3	
9	Homozygous R396H mutation of the RAG1 gene in a Saudi infant with Omenn syndrome: a case report. *Cases Journal*, **2009**, 2, 8391		1
8	Novel human pathological mutations. Gene symbol: WNT7A. Disease: ulnar and fibula absence, with severe limb deficiency. *Human Genetics*, **2009**, 125, 334	6.3	4
7	Gene symbol: LMX1B. Disease: Nail-Patella syndrome. *Human Genetics*, **2008**, 123, 109-10	6.3	2

6	The genetic basis of a craniofacial disease provides insight into COPII coat assembly. *Developmental Cell*, **2007**, 13, 623-634	10.2	147
5	Congenital duplication of the palm syndrome. *Annals of Plastic Surgery*, **2007**, 59, 341-3	1.7	12
4	Molecular breakpoint cloning and gene expression studies of a novel translocation t(4;15)(q27;q11.2) associated with Prader-Willi syndrome. *BMC Medical Genetics*, **2005**, 6, 18	2.1	48
3	Evidence for the role of PWCR1/HBII-85 C/D box small nucleolar RNAs in Prader-Willi syndrome. *American Journal of Human Genetics*, **2002**, 71, 669-78	11	104
2	Multi-colour FISH analysis of gene expression in formalin-fixed paraffin wax-embedded tissue. *Technical Tips Online*, **1996**, 1, 4-6		
1	Distinctive Features of a Saudi Genome		2