Khalda S Amr

List of Publications by Citations

55 papers	952 citations	13 h-index	30 g-index
57 ext. papers	1,115 ext. citations	3.5 avg, IF	3.81 L-index

#	Paper	IF	Citations
55	Identification of a frameshift mutation in Osterix in a patient with recessive osteogenesis imperfecta. *American Journal of Human Genetics*, **2010**, 87, 110-4	11	212
54	LRP4 mutations alter Wnt/beta-catenin signaling and cause limb and kidney malformations in Cenani-Lenz syndrome. *American Journal of Human Genetics*, **2010**, 86, 696-706	11	127
53	Temtamy preaxial brachydactyly syndrome is caused by loss-of-function mutations in chondroitin synthase 1, a potential target of BMP signaling. *American Journal of Human Genetics*, **2010**, 87, 757-67	11	77
52	Mutations in PLOD2 cause autosomal-recessive connective tissue disorders within the Bruck syndrome--osteogenesis imperfecta phenotypic spectrum. *Human Mutation*, **2012**, 33, 1444-9	4.7	67
51	Early diagnostic evaluation of miR-122 and miR-224 as biomarkers for hepatocellular carcinoma. *Genes and Diseases*, **2017**, 4, 215-221	6.6	50
50	Long interspersed nuclear element-1 (LINE1)-mediated deletion of EVC, EVC2, C4orf6, and STK32B in Ellis-van Creveld syndrome with borderline intelligence. *Human Mutation*, **2008**, 29, 931-8	4.7	43
49	Clinical and molecular analysis in families with autosomal recessive osteogenesis imperfecta identifies mutations in five genes and suggests genotype-phenotype correlations. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 1354-69	2.5	39
48	Association of vitamin D receptor gene polymorphism (VDR) with vitamin D deficiency, metabolic and inflammatory markers in Egyptian obese women. *Genes and Diseases*, **2017**, 4, 176-182	6.6	29
47	Expanding the phenotypic and mutational spectrum in microcephalic osteodysplastic primordial dwarfism type I. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1455-61	2.5	28
46	Assay for hepatitis C virus in peripheral blood mononuclear cells enhances sensitivity of diagnosis and monitoring of HCV-associated hepatitis. *Clinica Chimica Acta*, **1999**, 283, 1-14	6.2	27
45	Expanding the phenome and variome of skeletal dysplasia. *Genetics in Medicine*, **2018**, 20, 1609-1616	8.1	20
44	Assessment of the (rs1800795) and (rs1800796) Gene Polymorphisms in Egyptian Patients with Rheumatoid Arthritis. *Open Access Macedonian Journal of Medical Sciences*, **2016**, 4, 574-577	1	18
43	Evaluation of the correlation between serum levels of vitamin D and vitamin D receptor gene polymorphisms in an Egyptian population. *International Journal of Dermatology*, **2016**, 55, 1329-1335	1.7	16
42	Mutational spectrum of Xeroderma pigmentosum group A in Egyptian patients. *Gene*, **2014**, 533, 52-6	3.8	13
41	C242T polymorphism of NADPH oxidase p22phox gene reduces the risk of coronary artery disease in a random sample of Egyptian population. *Molecular Biology Reports*, **2014**, 41, 2281-6	2.8	12
40	Association of PTPN22 1858C-T polymorphism, HLA-DRB1 shared epitope and autoantibodies with rheumatoid arthritis. *Rheumatology International*, **2016**, 36, 1167-75	3.6	11
39	Apolipoprotein A5 T-1131C variant and risk for metabolic syndrome in obese adolescents. *Gene*, **2014**, 534, 44-7	3.8	11

38	Schistosoma hematobium soluble egg antigens induce proliferation of urothelial and endothelial cells. *World Journal of Urology*, **2001**, 19, 263-6	4	11
37	Role of nanoparticles in osteogenic differentiation of bone marrow mesenchymal stem cells. *Cytotechnology*, **2020**, 72, 1-22	2.2	11
36	Screening of dystrophin gene deletions in Egyptian patients with DMD/BMD muscular dystrophies. *Disease Markers*, **2000**, 16, 125-9	3.2	10
35	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. *Brain*, **2020**, 143, 2388-2397	11.2	10
34	Prediction of relapse after interferon therapy in hepatitis C virus-infected patients by the use of triple assay. *Journal of Gastroenterology and Hepatology (Australia)*, **2003**, 18, 68-73	4	9
33	Clinical and molecular characterization of seven Egyptian families with autosomal recessive robinow syndrome: Identification of four novel ROR2 gene mutations. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 3054-61	2.5	7
32	Reduction of RANTES expression in lesional psoriatic skin after narrow band ultraviolet therapy: a possible marker of therapeutic efficacy. *European Journal of Dermatology*, **2012**, 22, 481-7	0.8	7
31	Evaluation of serum and gingival crevicular fluid microRNA-223, microRNA-203 and microRNA-200b expression in chronic periodontitis patients with and without diabetes type 2. *Archives of Oral Biology*, **2021**, 121, 104949	2.8	7
30	Hereditary 1,25-dihydroxyvitamin D-resistant rickets with alopecia in four Egyptian families: report of three novel mutations in the vitamin D receptor gene. *Journal of Pediatric Endocrinology and Metabolism*, **2014**, 27, 873-8	1.6	6
29	Definition of the phenotypic spectrum of Temtamy preaxial brachydactyly syndrome associated with autosomal recessive CHYS1 mutations. *Middle East Journal of Medical Genetics*, **2012**, 1, 64-70		6
28	Association of the Pro12Ala Polymorphism with the Metabolic Parameters in Women with Polycystic Ovary Syndrome. *Open Access Macedonian Journal of Medical Sciences*, **2017**, 5, 275-280	1	6
27	Transforming growth factor-β1 gene polymorphism in psoriasis vulgaris. *Clinical, Cosmetic and Investigational Dermatology*, **2018**, 11, 415-419	2.9	6
26	Expression of B and T lymphocyte attenuator, retinoid-related orphan receptor gamma-isoform-t and interleukin 7 in psoriasis vulgaris. *Australasian Journal of Dermatology*, **2019**, 60, e132-e137	1.3	5
25	Whole exome sequencing identifies a new mutation in the SLC19A2 gene leading to thiamine-responsive megaloblastic anemia in an Egyptian family. *Molecular Genetics & Genomic Medicine*, **2019**, 7, e00777	2.3	5
24	A novel mutation in the leptin gene (W121X) in an Egyptian family. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 474-476	1.8	5
23	Genetic assessment of ten Egyptian patients with Sjögren-Larsson syndrome: expanding the clinical spectrum and reporting a novel ALDH3A2 mutation. *Archives of Dermatological Research*, **2019**, 311, 721-730	2.3	4
22	Advances in genomic diagnosis of a large cohort of Egyptian patients with disorders of sex development. *American Journal of Medical Genetics, Part A*, **2021**, 185, 1666-1677	2.5	4
21	Association of anti-cyclic citrullinated peptide antibodies and rheumatoid factor isotypes with HLA-DRB1 shared epitope alleles in Egyptian rheumatoid arthritis patients. *International Journal of Rheumatic Diseases*, **2020**, 23, 647-653	2.3	3

20	Mutational pattern in the 5reductase 2 (SRD5A2) gene in 46,XY Egyptian DSD patients. *Middle East Journal of Medical Genetics*, **2015**, 4, 77-82		3
19	Thalassemia From Genotype to Phenotype **2015**,		3
18	Mutational Analysis of the alpha-L-iduronidase gene in three Egyptian families: identification of three novel mutations and five novel polymorphisms. *Genetic Testing and Molecular Biomarkers*, **2009**, 13, 761-4	1.6	3
17	Association of IL-12 B Gene Polymorphism with Staging of Liver Disease in Chronic HCV Patients. *Infectious Disorders - Drug Targets*, **2018**, 18, 122-128	1.1	3
16	Epigenetic effects toward new insights as potential therapeutic target in B-thalassemia. *Journal of Genetic Engineering and Biotechnology*, **2021**, 19, 51	3.1	3
15	Impact of type 2 diabetes mellitus on the immunoregulatory characteristics of adipose tissue-derived mesenchymal stem cells. *International Journal of Biochemistry and Cell Biology*, **2021**, 140, 106072	5.6	3
14	Lipoid proteinosis: A clinical and molecular study in Egyptian patients. *Gene*, **2017**, 628, 308-314	3.8	2
13	Possible role of angiotensin-converting enzyme polymorphism on progression of hepatic fibrosis in chronic hepatitis C virus infection. *Transactions of the Royal Society of Tropical Medicine and Hygiene* , **2011**, 105, 396-400	2	2
12	Higher Expression of Toll-like Receptors 3, 7, 8, and 9 in Pityriasis Rosea. *Journal of Pathology and Translational Medicine*, **2017**, 51, 148-151	2.9	2
11	Clinical and Mutational Spectrum of Xeroderma Pigmentosum in Egypt: Identification of Six Novel Mutations and Implications for Ancestral Origins. *Genes*, **2021**, 12,	4.2	2
10	Are single nucleotide polymorphisms rs7903146 and rs12255372 in transcription factor 7-like 2 gene associated with an increased risk for gestational diabetes mellitus in Egyptian women?. *Journal of Genetic Engineering and Biotechnology*, **2021**, 19, 169	3.1	1
9	Correlation of circulating miRNA-33a and miRNA-122 with lipid metabolism among Egyptian patients with metabolic syndrome. *Journal of Genetic Engineering and Biotechnology*, **2021**, 19, 147	3.1	1
8	Analysis of Gene Mutations in Egyptian Children with Nephrotic Syndrome. *Open Access Macedonian Journal of Medical Sciences*, **2019**, 7, 3145-3148	1	1
7	Genetic susceptibility for insulin resistance among Egyptian women. *Journal of Genetic Engineering and Biotechnology*, **2016**, 14, 189-193	3.1	1
6	Genomic alterations in the F8 gene correlating with severe hemophilia A in Egyptian patients. *Molecular Genetics & Genomic Medicine*, **2021**, 9, e1575	2.3	0
5	Mutational spectrum of NF1 gene in 24 unrelated Egyptian families with neurofibromatosis type 1. *Molecular Genetics & Genomic Medicine*, **2021**, e1631	2.3	0
4	Assessment of Multiplex Ligation-Dependent Probe Amplification (MLPA) as a diagnostic test for Egyptian patients with Williams-Beuren syndrome. *Gene Reports*, **2020**, 20, 100767	1.4	
3	Study of DYRK1B gene expression and its association with metabolic syndrome in a small cohort of Egyptians. *Molecular Biology Reports*, **2021**, 48, 5497-5502	2.8	

2	Clinical, Biochemical, and Molecular Characterization of Metachromatic Leukodystrophy Among Egyptian Pediatric Patients: Expansion of the ARSA Mutational Spectrum. *Journal of Molecular Neuroscience*, **2021**, 71, 1112-1130	3.3
1	Nanomaterial-induced mesenchymal stem cell differentiation into osteoblast for counteracting bone resorption in the osteoporotic rats. *Tissue and Cell*, **2021**, 73, 101645	2.7