Khalda S Amr

List of Publications by Year
in descending order

57 papers	1,278 citations	516215 16 h-index	377514 34 g-index
57 all docs	57 docs citations	57 times ranked	2540 citing authors

#	Article	IF	Citations
1	Identification of a Frameshift Mutation in Osterix in a Patient with Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2010, 87, 110-114.	2.6	246
2	LRP4 Mutations Alter Wnt/Î²-Catenin Signaling and Cause Limb and Kidney Malformations in Cenani-Lenz Syndrome. American Journal of Human Genetics, 2010, 86, 696-706.	2.6	151
3	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, 757-767.	2.6	86
4	Mutations in PLOD2 cause autosomal-recessive connective tissue disorders within the Bruck syndrome-Osteogenesis imperfecta phenotypic spectrum. Human Mutation, 2012, 33, 1444-1449.	1.1	77
5	Early diagnostic evaluation of miR-122 and miR-224 as biomarkers for hepatocellular carcinoma. Genes and Diseases, 2017, 4, 215-221.	1.5	68
6	Long interspersed nuclear element-1 (LINE1)-mediated deletion ofEVC,EVC2,C4orf6, andSTK32B in Ellisâ€“van Creveld syndrome with borderline intelligence. Human Mutation, 2008, 29, 931-938.	1.1	55
7	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.	1.5	48
8	Expanding the phenome and variome of skeletal dysplasia. Genetics in Medicine, 2018, 20, 1609-1616.	1.1	46
9	Clinical and molecular analysis in families with autosomal recessive osteogenesis imperfecta identifies mutations in five genes and suggests genotypeâ€“phenotype correlations. American Journal of Medical Genetics, Part A, 2013, 161, 1354-1369.	0.7	42
10	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.	0.5	35
11	Expanding the phenotypic and mutational spectrum in microcephalic osteodysplastic primordial dwarfism type I. American Journal of Medical Genetics, Part A, 2012, 158A, 1455-1461.	0.7	30
12	Early-infantile onset epilepsy and developmental delay caused by bi-allelic GAD1 variants. Brain, 2020, 143, 2388-2397.	3.7	28
13	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.	0.8	23
14	Assessment of the Ã¢Ë†â€™174G/C (rs1800795) and Ã¢Ë†â€™572G/C (rs1800796) Interleukin 6 Gene Polymorphisms in Egyptian Patients with Rheumatoid Arthritis. Open Access Macedonian Journal of Medical Sciences, 2016, 4, 574-577.	0.1	22
15	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.	0.5	20
16	Role of nanoparticles in osteogenic differentiation of bone marrow mesenchymal stem cells. Cytotechnology, 2020, 72, 1-22.	0.7	20
17	Screening of Dystrophin Gene Deletions in Egyptian Patients with DMD/BMD Muscular Dystrophies. Disease Markers, 2000, 16, 125-129.	0.6	15
18	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.	1.4	15

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19	Apolipoprotein A5 T-1131C variant and risk for metabolic syndrome in obese adolescents. Gene, 2014, 534, 44-47.	1.0	15
20	Mutational spectrum of Xeroderma pigmentosum group A in Egyptian patients. Gene, 2014, 533, 52-56.	1.0	15
21	Schistosoma hematobium soluble egg antigens induce proliferation of urothelial and endothelial cells. World Journal of Urology, 2001, 19, 263-266.	1.2	14
22	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, 2281-2286.	1.0	14
23	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.	1.2	13
24	Association of PTPN22 1858Câ†'T polymorphism, HLA-DRB1 shared epitope and autoantibodies with rheumatoid arthritis. Rheumatology International, 2016, 36, 1167-1175.	1.5	12
25	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.	0.7	11
26	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, 167, 3054-3061.	0.7	10
27	Transforming growth factor-β1 gene polymorphism in psoriasis vulgaris. Clinical, Cosmetic and Investigational Dermatology, 2018, Volume 11, 415-419.	0.8	10
28	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, 481-487.	0.3	10
29	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.	0.1	10
30	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.	1.5	10
31	Epigenetic effects toward new insights as potential therapeutic target in B-thalassemia. Journal of Genetic Engineering and Biotechnology, 2021, 19, 51.	1.5	9
32	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.	0.4	8
33	Genetic assessment of ten Egyptian patients with SjÃ¶grenâ€"Larsson syndrome: expanding the clinical spectrum and reporting a novel ALDH3A2 mutation. Archives of Dermatological Research, 2019, 311, 721-730.	1.1	8
34	A novel mutation in the leptin gene (W121X) in an Egyptian family. Molecular Genetics and Metabolism Reports, 2014, 1, 474-476.	0.4	7
35	Whole exome sequencing identifies a new mutation in the SLC19A2 gene leading to thiamineâ€responsive megaloblastic anemia in an Egyptian family. Molecular Genetics & Genomic Medicine, 2019, 7, e00777.	0.6	7
36	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.	0.0	6

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37	Expression of B and T lymphocyte attenuator, retinoidâ€related orphan receptor gammaâ€isoformâ€ and interleukin 7 in psoriasis vulgaris. Australasian Journal of Dermatology, 2019, 60, e132-e137.	0.4	5
38	Association of antiâ€cyclic citrullinated peptide antibodies and rheumatoid factor isotypes with HLAâ€DRB1 shared epitope alleles in Egyptian rheumatoid arthritis patients. International Journal of Rheumatic Diseases, 2020, 23, 647-653.	0.9	5
39	Mutational spectrum of NF1 gene in 24 unrelated Egyptian families with neurofibromatosis type 1. Molecular Genetics & Genomic Medicine, 2021, 9, e1631.	0.6	5
40	Analysis of NPHS2 Gene Mutations in Egyptian Children with Nephrotic Syndrome. Open Access Macedonian Journal of Medical Sciences, 2019, 7, 3145-3148.	0.1	5
41	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.	0.7	4
42	Clinical and Mutational Spectrum of Xeroderma Pigmentosum in Egypt: Identification of Six Novel Mutations and Implications for Ancestral Origins. Genes, 2021, 12, 295.	1.0	4
43	Gene Mutations of the Three Ectodysplasin Pathway Key Players (EDA, EDAR, and EDARADD) Account for More than 60% of Egyptian Ectodermal Dysplasia: A Report of Seven Novel Mutations. Genes, 2021, 12, 1389.	1.0	4
44	Association of IL-12 B Gene Polymorphism with Staging of Liver Disease in Chronic HCV Patients. Infectious Disorders - Drug Targets, 2018, 18, 122-128.	0.4	4
45	Mutational Analysis of the Î±-L-Iduronidase Gene in Three Egyptian Families: Identification of Three Novel Mutations and Five Novel Polymorphisms. Genetic Testing and Molecular Biomarkers, 2009, 13, 761-764.	0.3	3
46	Mutational pattern in the 5Î± reductase 2 (SRD5A2) gene in 46,XY Egyptian DSD patients. Middle East Journal of Medical Genetics, 2015, 4, 77-82.	0.0	3
47	Thalassemia â€" From Genotype to Phenotype. , 0, , .		3
48	Lipoid proteinosis: A clinical and molecular study in Egyptian patients. Gene, 2017, 628, 308-314.	1.0	3
49	Higher Expression of Toll-like Receptors 3, 7, 8, and 9 in Pityriasis Rosea. Journal of Pathology and Translational Medicine, 2017, 51, 148-151.	0.4	3
50	Nextâ€generation sequencing in identification of pathogenic variants in primary hyperoxaluria among 21 Egyptian families: Identification of two novel AGXT gene mutations. Molecular Genetics & Genomic Medicine, 0, , .	0.6	3
51	Genomic alterations in the F8 gene correlating with severe hemophilia A in Egyptian patients. Molecular Genetics & Genomic Medicine, 2021, 9, e1575.	0.6	2
52	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.	1.5	2
53	Genetic susceptibility for insulin resistance among Egyptian women. Journal of Genetic Engineering and Biotechnology, 2016, 14, 189-193.	1.5	1
54	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.	1.1	1

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55	Study of DYRK1B gene expression and its association with metabolic syndrome in a small cohort of Egyptians. Molecular Biology Reports, 2021, 48, 5497-5502.	1.0	1
56	Nanomaterial-induced mesenchymal stem cell differentiation into osteoblast for counteracting bone resorption in the osteoporotic rats. Tissue and Cell, 2021, 73, 101645.	1.0	1
57	Assessment of Multiplex Ligation-Dependent Probe Amplification (MLPA) as a diagnostic test for Egyptian patients with Williams-Beuren syndrome. Gene Reports, 2020, 20, 100767.	0.4	0