Hatem El-Shanti

List of Publications by Year
in descending order

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83 papers	5,303 citations	136940 32 h-index	85537 71 g-index
84 all docs	84 docs citations	84 times ranked	6059 citing authors

#	Article	IF	Citations
1	An Autoinflammatory Disease with Deficiency of the Interleukin-1â€“Receptor Antagonist. New England Journal of Medicine, 2009, 360, 2426-2437.	27.0	892
2	Homozygous mutations in LPIN2 are responsible for the syndrome of chronic recurrent multifocal osteomyelitis and congenital dyserythropoietic anaemia (Majeed syndrome). Journal of Medical Genetics, 2005, 42, 551-557.	3.2	346
3	Independent Introduction of Two Lactase-Persistence Alleles into Human Populations Reflects Different History of Adaptation to Milk Culture. American Journal of Human Genetics, 2008, 82, 57-72.	6.2	301
4	Mutations in the CCN gene family member WISP3 cause progressive pseudorheumatoid dysplasia. Nature Genetics, 1999, 23, 94-98.	21.4	260
5	The infevers autoinflammatory mutation online registry: update with new genes and functions. Human Mutation, 2008, 29, 803-808.	2.5	239
6	A Homozygous Mutation in a Novel Zinc-Finger Protein, ERIS, Is Responsible for Wolfram Syndrome 2. American Journal of Human Genetics, 2007, 81, 673-683.	6.2	233
7	A missense mutation in pstpip2 is associated with the murine autoinflammatory disorder chronic multifocal osteomyelitis. Bone, 2006, 38, 41-47.	2.9	199
8	A Homozygous Mutation in Human PRICKLE1 Causes an Autosomal-Recessive Progressive Myoclonus Epilepsy-Ataxia Syndrome. American Journal of Human Genetics, 2008, 83, 572-581.	6.2	199
9	Autoinflammatory bone disorders. Current Opinion in Rheumatology, 2007, 19, 492-498.	4.3	140
10	Familial Mediterranean fever in Arabs. Lancet, The, 2006, 367, 1016-1024.	13.7	127
11	Chronic Recurrent Multifocal Osteomyelitis. Clinical Orthopaedics and Related Research, 2007, 462, 11-19.	1.5	126
12	Mutations in Prickle Orthologs Cause Seizures in Flies, Mice, and Humans. American Journal of Human Genetics, 2011, 88, 138-149.	6.2	125
13	A Homozygous Mutation in ADAMTSL4 Causes Autosomal-Recessive Isolated Ectopia Lentis. American Journal of Human Genetics, 2009, 84, 274-278.	6.2	122
14	Homozygosity Mapping Identifies an Additional Locus for Wolfram Syndrome on Chromosome 4q. American Journal of Human Genetics, 2000, 66, 1229-1236.	6.2	116
15	Homozygosity Mapping Places the Acrodermatitis Enteropathica Gene on Chromosomal Region 8q24.3. American Journal of Human Genetics, 2001, 68, 1055-1060.	6.2	108
16	The syndrome of chronic recurrent multifocal osteomyelitis and congenital dyserythropoietic anaemia. Report of a new family and a review. European Journal of Pediatrics, 2001, 160, 705-710.	2.7	108
17	A splice site mutation confirms the role ofLPIN2 in Majeed syndrome. Arthritis and Rheumatism, 2007, 56, 960-964.	6.7	102
18	A novel mutation of IL1RN in the deficiency of interleukin-1 receptor antagonist syndrome: Description of two unrelated cases from Brazil. Arthritis and Rheumatism, 2011, 63, 4007-4017.	6.7	96

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19	Neutrophil dysfunction in a family with a SAPHO syndromeâ€“like phenotype. Arthritis and Rheumatism, 2008, 58, 3264-3269.	6.7	88
20	Familial Mediterranean fever in children: the expanded clinical profile. QJM - Monthly Journal of the Association of Physicians, 1999, 92, 309-318.	0.5	84
21	Multiplex epithelium dysfunction due to CLDN10 mutation: the HELIX syndrome. Genetics in Medicine, 2018, 20, 190-201.	2.4	75
22	Disruption of the non-canonical Wnt gene PRICKLE2 leads to autism-like behaviors with evidence for hippocampal synaptic dysfunction. Molecular Psychiatry, 2013, 18, 1077-1089.	7.9	74
23	The Spectrum of Familial Mediterranean Fever Gene Mutations in Arabs: Report of a Large Series. Seminars in Arthritis and Rheumatism, 2005, 34, 813-818.	3.4	71
24	Seizures Are Regulated by Ubiquitin-specific Peptidase 9 X-linked (USP9X), a De-Ubiquitinase. PLoS Genetics, 2015, 11, e1005022.	3.5	66
25	Genotype/phenotype correlations in Arab patients with familial Mediterranean fever. Seminars in Arthritis and Rheumatism, 2002, 31, 371-376.	3.4	59
26	Progressive pseudorheumatoid dysplasia: report of a family and review.. Journal of Medical Genetics, 1997, 34, 559-563.	3.2	54
27	Henoch-Schonlein Purpura: clinical experience and contemplations on a streptococcal association. Journal of Tropical Pediatrics, 1996, 42, 200-203.	1.5	39
28	PRICKLE1 Interaction with SYNAPSIN I Reveals a Role in Autism Spectrum Disorders. PLoS ONE, 2013, 8, e80737.	2.5	39
29	Epidermal Nevus Syndrome: Subgroup With Neuronal Migration Defects. Journal of Child Neurology, 1992, 7, 29-34.	1.4	36
30	Consanguinity: implications for practice, research, and policy. Lancet, The, 2006, 367, 970-971.	13.7	35
31	Interleukin-1 receptor antagonist deficiency with a novel mutation; late onset and successful treatment with canakinumab: a case report. Journal of Medical Case Reports, 2015, 9, 145.	0.8	35
32	Henoch-Schonlein purpura and streptococcal infection: a prospective case-control study. Annals of Tropical Paediatrics, 1999, 19, 253-255.	1.0	34
33	Bleeding tendency in Wolfram syndrome: a newly identified feature with phenotype genotype correlation. European Journal of Pediatrics, 2001, 160, 243-246.	2.7	34
34	Wolfram syndrome: Identification of a phenotypic and genotypic variant from Jordan. American Journal of Medical Genetics Part A, 2002, 115, 61-65.	2.4	33
35	Risk factors for childhood epilepsy: a case-control study from Irbid, Jordan. Seizure: the Journal of the British Epilepsy Association, 2003, 12, 171-174.	2.0	33
36	The clinical spectrum of Henoch-SchÃ¶nlein purpura in infants and young children. European Journal of Pediatrics, 1995, 154, 969-972.	2.7	29

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37	Chronic Recurrent Multifocal Osteomyelitis and Deficiency of Interleukin-1â€“receptor Antagonist. Pediatric Infectious Disease Journal, 2013, 32, 94.	2.0	29
38	Raine syndrome: report of a case with hand and foot anomalies. Clinical Dysmorphology, 2001, 10, 227-229.	0.3	25
39	T2DM GWAS in the Lebanese population confirms the role of TCF7L2 and CDKAL1 in disease susceptibility. Scientific Reports, 2014, 4, 7351.	3.3	25
40	Transient improvement of congenital lactic acidosis in a male infant with pyruvate decarboxylase deficiency treated with dichloroacetate. Journal of Pediatrics, 1993, 123, 427-430.	1.8	24
41	Familial disorder of sex determination in seven individuals from three related sibships. European Journal of Pediatrics, 2000, 159, 912-918.	2.7	23
42	Neonatal meningitis in Northern Jordan. Journal of Tropical Pediatrics, 1996, 42, 267-270.	1.5	22
43	Assignment of gene responsible for progressive pseudorheumatoid dysplasia to chromosome 6 and examination of COL10A1 as candidate gene. European Journal of Human Genetics, 1998, 6, 251-256.	2.8	22
44	Spectrum of mutations and carrier frequency of familial Mediterranean fever gene in the Algerian population. Rheumatology, 2011, 50, 2306-2310.	1.9	21
45	Biallelic <scp>SCN</scp>10A mutations in neuromuscular disease and epileptic encephalopathy. Annals of Clinical and Translational Neurology, 2017, 4, 26-35.	3.7	20
46	Type II diabetes mellitus and hyperhomocysteinemia: a complex interaction. Diabetology and Metabolic Syndrome, 2017, 9, 19.	2.7	20
47	Novel sequence variants in the TMIE gene in families with autosomal recessive nonsyndromic hearing impairment. Journal of Molecular Medicine, 2006, 84, 226-231.	3.9	19
48	Majeed Syndrome: A Review of the Clinical, Genetic and Immunologic Features. Biomolecules, 2021, 11, 367.	4.0	19
49	Breakpoint localization using arrayâ€CGH in three siblings with an unbalanced 4q;16q translocation and childhood apraxia of speech (CAS). American Journal of Medical Genetics, Part A, 2008, 146A, 2227-2233.	1.2	18
50	Mutations in zinc finger 407 [ZNF407] cause a unique autosomal recessive cognitive impairment syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 80.	2.7	17
51	Multivariate epidemiologic analysis of type 2 diabetes mellitus risks in the Lebanese population. Diabetology and Metabolic Syndrome, 2014, 6, 89.	2.7	17
52	Caffeine Impact on Metabolic Syndrome Components Is Modulated by a CYP1A2 Variant. Annals of Nutrition and Metabolism, 2016, 68, 1-11.	1.9	16
53	Hypogonadotrophic hypogonadism, short stature, cerebellar ataxia, rod-cone retinal dystrophy, and hypersegmented neutrophils: a novel disorder or a new variant of Boucher-Neuhauser syndrome?. Journal of Medical Genetics, 2003, 40, 2e-2.	3.2	15
54	A distinct autosomal recessive ataxia maps to chromosome 12in an inbred family from Jordan. Brain and Development, 2006, 28, 353-357.	1.1	15

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55	Overlap of Familial Mediterranean Fever and Hyper-IgD Syndrome in an Arabic Kindred. Journal of Clinical Immunology, 2015, 35, 249-253.	3.8	15
56	Biallelic loss of function variants in PPP1R21 cause a neurodevelopmental syndrome with impaired endocytic function. Human Mutation, 2019, 40, 267-280.	2.5	15
57	THE EFFECT OF TRIFLUOPERAZINE ON THE GENOTOXICITY OF BLEOMYCIN IN CULTURED HUMAN LYMPHOCYTES. Drug and Chemical Toxicology, 2000, 23, 361-369.	2.3	14
58	The M694I/M694I genotype: A genetic risk factor of AA-amyloidosis in a group of Algerian patients with familial Mediterranean fever. European Journal of Medical Genetics, 2017, 60, 149-153.	1.3	14
59	Heterozygous PDGFRB Mutation in a Three-generation Family with Autosomal Dominant Infantile Myofibromatosis. Acta Dermato-Venereologica, 2017, 97, 858-859.	1.3	14
60	Alopecia universalis congenita, XY gonadal dysgenesis and laryngomalacia: a novel malformation syndrome. European Journal of Pediatrics, 2003, 162, 36-40.	2.7	12
61	A chromosomal microdeletion of 15q in a female patient with epilepsy, <scp>ID</scp>, and autism spectrum disorder: a case report. Clinical Case Reports (discontinued), 2017, 5, 1013-1017.	0.5	11
62	The Impact of Genetic Diseases on Jordanians: Strategies Towards Prevention. Journal of Biomedicine and Biotechnology, 2001, 1, 45-47.	3.0	10
63	Distal trisomy 10q syndrome, report of a patient with duplicated q24.31 â€“ qter, autism spectrum disorder and unusual features. Clinical Case Reports (discontinued), 2014, 2, 201-205.	0.5	10
64	Induction of chromosomal aberrations by the rhodium(III) complex cis-[Rh(biq)2Cl2]Cl in cultured human lymphocytes. Mutagenesis, 2000, 15, 375-378.	2.6	9
65	Family-Based Genome-Wide Association Study of Autism Spectrum Disorder in Middle Eastern Families. Genes, 2021, 12, 761.	2.4	7
66	Effects of short term metformin administration on androgens in diabetic men. Journal of King Abdulaziz University, Islamic Economics, 2004, 25, 75-8.	1.1	6
67	Effect of Desferrioxamine in Acute Haemolytic Anaemia of Glucose-6-Phosphate Dehydrogenase Deficiency. Acta Haematologica, 1999, 101, 145-148.	1.4	4
68	Fine Mapping of Progressive Pseudorheumatoid Dysplasia: A Tool for Heterozygote Identification. Genetic Testing and Molecular Biomarkers, 1999, 3, 329-333.	1.7	4
69	Cerebellar hypoplasia, hypergonadotrophic hypogonadism, retinitis pigmentosa, alopecia, microcephaly, psychomotor retardation, and short stature: â€œD-CHRAMPS syndromeâ€. European Journal of Pediatrics, 2002, 161, 170-172.	2.7	4
70	Mosaic partial pericentromeric trisomy 8 and maternal uniparental disomy in a male patient with autism spectrum disorder. Clinical Case Reports (discontinued), 2016, 4, 1125-1131.	0.5	4
71	Congenital glucoseâ€galactose malabsorption: A case report with a novel SLC5A1 mutation. Clinical Case Reports (discontinued), 2019, 7, 51-53.	0.5	4
72	A Case of Wolfram Syndrome in Triplets: Some Newly Recognized Features. Annals of Saudi Medicine, 1999, 19, 132-134.	1.1	4

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73	Homozygous AMN mutation in hereditary selective intestinal malabsorption of vitamin B12 in Jordan. Journal of King Abdulaziz University, Islamic Economics, 2005, 26, 1061-4.	1.1	4
74	A clinical study of a large inbred kindred with pure familial spastic paraplegia. Brain and Development, 1999, 21, 478-482.	1.1	3
75	Diversity in polyp pathology and distribution of Familial Juvenile Polyposis Syndrome. Journal of King Abdulaziz University, Islamic Economics, 2002, 23, 328-31.	1.1	2
76	The pattern of peripheral blood chromosomal abnormalities in Northern Jordan. Journal of King Abdulaziz University, Islamic Economics, 2002, 23, 1552-4.	1.1	2
77	Further delineation of El-Shanti syndrome. European Journal of Pediatrics, 2004, 163, 761-762.	2.7	1
78	Genetic Disorders in Jordan. , 2010, , 325-352.		1
79	A Novel De Novo Pstpip1 Mutation In A Boy With Pyogenic Arthritis, Pyoderma Gangrenosum, Acne (Papa) Syndrome. , 2013, , .		1
80	Fanconi's Anemia and Primary Hypothyroidism. Annals of Saudi Medicine, 1998, 18, 58-59.	1.1	1
81	Hereditary spastic paraplegia. Handbook of Clinical Neurophysiology, 2004, , 633-653.	0.0	0
82	Familial Mediterranean Fever and Other Autoinflammatory Disorders. , 2010, , 111-143.		0
83	A Novel Syndrome With Short Stature, Mandibular Hypoplasia, and Osteoporosis May Be Associated With a PRRT3 Variant. Journal of the Endocrine Society, 2020, 4, bvaa088.	0.2	0