Solaf Mohamed Elsayed

List of Publications by Year
in descending order

60	1,277	471477	361001
papers	citations	17 h-index	35 g-index

64	64	64	2464
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	WNT1 Mutations in Families Affected by Moderately Severe and Progressive Recessive Osteogenesis Imperfecta. American Journal of Human Genetics, 2013, 92, 590-597.	6.2	179
2	Mutations in KIF7 link Joubert syndrome with Sonic Hedgehog signaling and microtubule dynamics. Journal of Clinical Investigation, 2011, 121, 2662-2667.	8.2	173
3	The Cockayne Syndrome Natural History (CoSyNH) study: clinical findings in 102 individuals and recommendations for care. Genetics in Medicine, 2016, 18, 483-493.	2.4	127
4	Mutations in CDC45 , Encoding an Essential Component of the Pre-initiation Complex, Cause Meier-Gorlin Syndrome and Craniosynostosis. American Journal of Human Genetics, 2016, 99, 125-138.	6.2	92
5	Consanguineous matings among Egyptian population. Egyptian Journal of Medical Human Genetics, 2011, 12, 157-163.	1.0	78
6	Consanguinity and its relevance to clinical genetics. Egyptian Journal of Medical Human Genetics, 2013, 14, 157-164.	1.0	66
7	Next-generation sequencing reveals the mutational landscape of clinically diagnosed Usher syndrome: copy number variations, phenocopies, a predominant target for translational read-through, andPEX26mutated in Heimler syndrome. Molecular Genetics & Genomic Medicine, 2017, 5, 531-552.	1.2	55
8	Clinical and genetic characterization of chanarin-dorfman syndrome patients: first report of large deletions in the ABHD5 gene. Orphanet Journal of Rare Diseases, 2010, 5, 33.	2.7	47
9	Evidence for single origins of 35delG and delE120 mutations in the GJB2 gene in Anatolia. Clinical Genetics, 2004, 67, 31-37.	2.0	46
10	Phenotypic and Genetic Characterization of a Cohort of Pediatric Wilson Disease Patients. BMC Pediatrics, 2011, 11, 56.	1.7	45
11	Mutational analysis of ATP7B gene in Egyptian children with Wilson disease: 12 novel mutations. Journal of Human Genetics, 2008, 53, 681-687.	2.3	37
12	Autosomal dominant SCA5 and autosomal recessive infantile SCA are allelic conditions resulting from SPTBN2 mutations. European Journal of Human Genetics, 2014, 22, 286-288.	2.8	37
13	Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene. Human Molecular Genetics, 2015, 24, 2594-2603.	2.9	32
14	Phenotype of apoptotic lymphocytes in children with Down syndrome. Immunity and Ageing, 2009, 6, 2.	4.2	26
15	Outcome of enzyme replacement therapy in children with Gaucher disease: The Egyptian experience. Egyptian Journal of Medical Human Genetics, 2011, 12, 9-14.	1.0	19
16	A novel mutation in BAP/SIL1 gene causes Marinesco-SjÃ¶gren syndrome in an extended pedigree. Clinical Genetics, 2006, 70, 420-423.	2.0	18
17	Structural chromosomal abnormalities in couples with recurrent abortion in Egypt. Turkish Journal of Medical Sciences, 2015, 45, 208-213.	0.9	17
18	Maladaptative Autophagy Impairs Adipose Function in Congenital Generalized Lipodystrophy due to Cavin-1 Deficiency. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2892-2904.	3.6	17

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19	An integrated multiomic approach as an excellent tool for the diagnosis of metabolic diseases: our first 3720 patients. European Journal of Human Genetics, 2022, 30, 1029-1035.	2.8	13
20	Treatment options for patients with Gaucher disease. Egyptian Journal of Medical Human Genetics, 2016, 17, 281-285.	1.0	12
21	Role of Plasma Amino Acids and Urinary Organic Acids in Diagnosis of Mitochondrial Diseases in Children. Pediatric Neurology, 2014, 51, 820-825.	2.1	11
22	Maternal MTHFR C677T genotype and septal defects in offspring with Down syndrome: A pilot study. Egyptian Journal of Medical Human Genetics, 2014, 15, 39-44.	1.0	11
23	Inherited Thrombophilia in Pediatric Ischemic Stroke: An Egyptian Study. Pediatric Neurology, 2012, 47, 114-118.	2.1	10
24	A case of infantile osteopetrosis: The radioclinical features with literature update. Bone Reports, 2016, 4, 11-16.	0.4	10
25	Dorfman-Chanarin syndrome in Egypt. American Journal of Medical Genetics Part A, 2003, 121A, 75-78.	2.4	9
26	DOUBLE HOMOZYGOSITY FOR MUTATIONS OF AGL AND SCN9A MIMICKING NEUROHEPATOPATHY SYNDROME. Neurology, 2008, 70, 2343-2344.	1.1	9
27	Orthopaedic manifestations of Proteus syndrome in a child with literature update. Bone Reports, 2015, 3, 104-108.	0.4	9
28	Cholestasis in patients with Cockayne syndrome and suggested modified criteria for clinical diagnosis. Orphanet Journal of Rare Diseases, 2011, 6, 13.	2.7	8
29	Wolman disease in patients with familial hemophagocytic lymphohistiocytosis (FHL) negative mutations. Egyptian Journal of Medical Human Genetics, 2016, 17, 277-280.	1.0	7
30	Factor V G1691A (Leiden) is a major etiological factor in Egyptian Budd-Chiari syndrome patients. Turkish Journal of Haematology, 2011, 2011, 299-305.	0.5	6
31	Connexin 26 (GJB2) mutation in KID syndrome: An Egyptian patient. Egyptian Journal of Medical Human Genetics, 2011, 12, 91-93.	1.0	5
32	Oral-facial-digital syndrome type II: Transitional type between Mohr and Varadi. Egyptian Journal of Medical Human Genetics, 2013, 14, 311-315.	1.0	5
33	Neurofibromatosis type 1 and multiple sclerosis: Genetically related diseases. Egyptian Journal of Medical Human Genetics, 2017, 18, 295-298.	1.0	5
34	Frequency of inborn errors of metabolism screening for children with unexplained acute encephalopathy at an emergency department. Neuropsychiatric Disease and Treatment, 2018, Volume 14, 1715-1720.	2.2	5
35	Multicentric Osteolysis, Nodulosis, and Arthropathy in two unrelated children with matrix metalloproteinase 2 variants: Genetic-skeletal correlations. Bone Reports, 2021, 15, 101106.	0.4	5
36	Non-deletion mutations in Egyptian patients with Duchenne muscular dystrophy. Egyptian Journal of Medical Human Genetics, 2014, 15, 235-240.	1.0	4

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37	Colchicine resistant FMF is not always true resistance. Egyptian Journal of Medical Human Genetics, 2011, 12, 99-101.	1.0	2
38	TNF-Î±-308 G/A Polymorphism in Egyptian Budd-Chiari Syndrome Patients. Turkish Journal of Haematology, 2012, 29, 420-421.	0.2	2
39	Familial Peters Plus syndrome with absent anal canal, sacral agenesis and sensorineural hearing loss: Expanding the clinical spectrum. Egyptian Journal of Medical Human Genetics, 2013, 14, 423-428.	1.0	2
40	Prominent extensor truncal dystonia in egyptian patients with Wilson's disease. Movement Disorders, 2014, 29, 151-153.	3.9	2
41	BH4 deficiency with unusual presentations: Challenges and lessons. Egyptian Journal of Medical Human Genetics, 2016, 17, 241-242.	1.0	2
42	Pseudoachondroplasia in a child: The role of anthropometric measurements and skeletal imaging in differential diagnosis. Egyptian Journal of Radiology and Nuclear Medicine, 2017, 48, 245-250.	0.6	2
43	Reversal of skeletal radiographic pathology in a case of malignant infantile osteopetrosis following hematopoietic stem cell transplantation. Egyptian Journal of Radiology and Nuclear Medicine, 2017, 48, 237-243.	0.6	2
44	Disentangling molecular and clinical stratification patterns in beta-galactosidase deficiency. Journal of Medical Genetics, 2022, 59, 377-384.	3.2	2
45	Screening for subtle chromosomal rearrangements in an Egyptian sample of children with unexplained mental retardation. Egyptian Journal of Medical Human Genetics, 2011, 12, 63-68.	1.0	1
46	Treatment-induced copper deficiency in two patients with Wilsonâ€™s disease. Egyptian Liver Journal, 2016, 6, 16-20.	0.6	1
47	Are we missing fucosidosis?. Egyptian Journal of Medical Human Genetics, 2018, 19, 151-152.	1.0	1
48	Ribosomal protein S19 - 631 insertion is an African-originated mutation. Turkish Journal of Haematology, 2010, 27, 123-124.	0.5	0
49	Multiple pterygium syndrome with marked pterygia of the fingers and MRI changes in the spine. Egyptian Journal of Medical Human Genetics, 2012, 13, 107-113.	1.0	0
50	Hypothyroidism could be the only manifestation of mitochondrial T8993C mutation in Leigh syndrome. Egyptian Journal of Medical Human Genetics, 2013, 14, 201-203.	1.0	0
51	A severe form of cholestasis lymphoedema syndrome (Aagenaes syndrome) with progressive arthritis. Egyptian Liver Journal, 2014, 4, 30-32.	0.6	0
52	The blessing effect of an extra copy of chromosome 21. Egyptian Journal of Medical Human Genetics, 2014, 15, 209-210.	1.0	0
53	Corrigendum to Role of Plasma Amino Acids and Urinary Organic Acids in Diagnosis of Mitochondrial Diseases in Children. Pediatric Neurology, 2015, 52, 470.	2.1	0
54	A Novel Frameshift Mutation in SLC2A1 Associated with a Mild form of Glucose Transporter Type 1-Related Movement Disorder. Journal of Pediatric Neurology, 2015, 13, 088-091.	0.2	0

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55	Autosomal recessive ichthyosis with limb reduction defect: A simple association and not CHILD syndrome. Egyptian Journal of Medical Human Genetics, 2016, 17, 255-258.	1.0	0
56	Challenges in diagnosis and counseling of a family with two recessive neurometabolic disorders. Egyptian Journal of Medical Human Genetics, 2016, 17, 247-250.	1.0	0
57	REMOVED: An Egyptian patient with Schwartz-Jampel syndrome type I and new ocular findings. Egyptian Journal of Medical Human Genetics, 2017, 18, 393-396.	1.0	0
58	Cardiomyopathy in Vici syndrome. Egyptian Journal of Medical Human Genetics, 2018, 19, 49-50.	1.0	0
59	Removal notice to â€œAn Egyptian patient with Schwartz-Jampel syndrome type I and new ocular findingsâ€•[Egypt J Med Hum Genet 18 (2017) 393â€“396]. Egyptian Journal of Medical Human Genetics, 2018, 19, 443.	1.0	0
60	Skeletal radiology following hematopoietic stem cell transplantation in infantile osteopetrosis: an overlooked assessment tool. BoletÃn MÃ©dico Del Hospital Infantil De MÃ©xico, 2021, 78, 495-496.	0.3	0