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	Disentangling molecular and clinical stratification patterns in beta-galactosidase deficiency. Journal of Medical Genetics, 2022, 59, 377-384.	3.2	2
2	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
3	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
4	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
5	Cardiomyopathy in Vici syndrome. Egyptian Journal of Medical Human Genetics, 2018, 19, 49-50.	1.0	0
6	Are we missing fucosidosis?. Egyptian Journal of Medical Human Genetics, 2018, 19, 151-152.	1.0	1
7	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
8	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
9	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
10	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
11	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
12	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
13	Neurofibromatosis type 1 and multiple sclerosis: Genetically related diseases. Egyptian Journal of Medical Human Genetics, 2017, 18, 295-298.	1.0	5
14	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
15	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
16	A case of infantile osteopetrosis: The radioclinical features with literature update. Bone Reports, 2016, 4, 11-16.	0.4	10
17	Treatment options for patients with Gaucher disease. Egyptian Journal of Medical Human Genetics, 2016, 17, 281-285.	1.0	12
18	Treatment-induced copper deficiency in two patients with Wilsonâ€™s disease. Egyptian Liver Journal, 2016, 6, 16-20.	0.6	1

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19	BH4 deficiency with unusual presentations: Challenges and lessons. Egyptian Journal of Medical Human Genetics, 2016, 17, 241-242.	1.0	2
20	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
21	Wolman disease in patients with familial hemophagocytic lymphohistiocytosis (FHL) negative mutations. Egyptian Journal of Medical Human Genetics, 2016, 17, 277-280.	1.0	7
22	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
23	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
24	Orthopaedic manifestations of Proteus syndrome in a child with literature update. Bone Reports, 2015, 3, 104-108.	0.4	9
25	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
26	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
27	Structural chromosomal abnormalities in couples with recurrent abortion in Egypt. Turkish Journal of Medical Sciences, 2015, 45, 208-213.	0.9	17
28	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
29	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
30	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
31	A severe form of cholestasis lymphoedema syndrome (Aagenaes syndrome) with progressive arthritis. Egyptian Liver Journal, 2014, 4, 30-32.	0.6	0
32	Non-deletion mutations in Egyptian patients with Duchenne muscular dystrophy. Egyptian Journal of Medical Human Genetics, 2014, 15, 235-240.	1.0	4
33	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
34	The blessing effect of an extra copy of chromosome 21. Egyptian Journal of Medical Human Genetics, 2014, 15, 209-210.	1.0	0
35	Prominent extensor truncal dystonia in egyptian patients with Wilson's disease. Movement Disorders, 2014, 29, 151-153.	3.9	2
36	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

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37	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
38	Consanguinity and its relevance to clinical genetics. Egyptian Journal of Medical Human Genetics, 2013, 14, 157-164.	1.0	66
39	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
40	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
41	TNF-Î±-308 G/A Polymorphism in Egyptian Budd-Chiari Syndrome Patients. Turkish Journal of Haematology, 2012, 29, 420-421.	0.2	2
42	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
43	Inherited Thrombophilia in Pediatric Ischemic Stroke: An Egyptian Study. Pediatric Neurology, 2012, 47, 114-118.	2.1	10
44	Connexin 26 (GJB2) mutation in KID syndrome: An Egyptian patient. Egyptian Journal of Medical Human Genetics, 2011, 12, 91-93.	1.0	5
45	Colchicine resistant FMF is not always true resistance. Egyptian Journal of Medical Human Genetics, 2011, 12, 99-101.	1.0	2
46	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
47	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
48	Consanguineous matings among Egyptian population. Egyptian Journal of Medical Human Genetics, 2011, 12, 157-163.	1.0	78
49	Mutations in KIF7 link Joubert syndrome with Sonic Hedgehog signaling and microtubule dynamics. Journal of Clinical Investigation, 2011, 121, 2662-2667.	8.2	173
50	Phenotypic and Genetic Characterization of a Cohort of Pediatric Wilson Disease Patients. BMC Pediatrics, 2011, 11, 56.	1.7	45
51	Cholestasis in patients with Cockayne syndrome and suggested modified criteria for clinical diagnosis. Orphanet Journal of Rare Diseases, 2011, 6, 13.	2.7	8
52	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
53	Ribosomal protein S19 - 631 insertion is an African-originated mutation. Turkish Journal of Haematology, 2010, 27, 123-124.	0.5	0
54	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

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55	Phenotype of apoptotic lymphocytes in children with Down syndrome. Immunity and Ageing, 2009, 6, 2.	4.2	26
56	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
57	DOUBLE HOMOZYGOSITY FOR MUTATIONS OF AGL AND SCN9A MIMICKING NEUROHEPATOPATHY SYNDROME. Neurology, 2008, 70, 2343-2344.	1.1	9
58	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
59	Evidence for single origins of 35delG and delE120 mutations in the GJB2 gene in Anatolia. Clinical Genetics, 2004, 67, 31-37.	2.0	46
60	Dorfman-Chanarin syndrome in Egypt. American Journal of Medical Genetics Part A, 2003, 121A, 75-78.	2.4	9