

Ekram M Fateen

List of Publications by Year in descending order

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Version: 2024-02-01

14
papers

144
citations

1307366

7
h-index

1199470

12
g-index

14
all docs

14
docs citations

14
times ranked

268
citing authors

#	ARTICLE	IF	CITATIONS
1	PEX6 is Expressed in Photoreceptor Cilia and Mutated in Deafblindness with Enamel Dysplasia and Microcephaly. <i>Human Mutation</i> , 2016, 37, 170-174.	1.1	36
2	Dyggveâ€“Melchiorâ€“Clausen syndrome: clinical, genetic, and radiological study of 15 Egyptian patients from nine unrelated families. <i>Journal of Children's Orthopaedics</i> , 2009, 3, 451-458.	0.4	19
3	Outcome of enzyme replacement therapy in children with Gaucher disease: The Egyptian experience. <i>Egyptian Journal of Medical Human Genetics</i> , 2011, 12, 9-14.	0.5	19
4	Molecular characterization of Egyptian patients with glycogen storage disease type IIIa. <i>Journal of Human Genetics</i> , 2005, 50, 538-542.	1.1	17
5	Egyptian glycogen storage disease type III â€“ identification of six novel AGL mutations, including a large 1.5Åkb deletion and a missense mutation p.L620P with subtype III d. <i>Clinical Chemistry and Laboratory Medicine</i> , 2009, 47, 1233-8.	1.4	10
6	Twenty-five years of biochemical diagnosis of Gaucher disease: the Egyptian experience. <i>Heliyon</i> , 2019, 5, e02574.	1.4	10
7	Glycogen storage disease type III in Egyptian children: A single centre clinico-laboratory study. <i>Arab Journal of Gastroenterology</i> , 2014, 15, 63-67.	0.4	9
8	Fifteen years experience: Egyptian metabolic lab. <i>Egyptian Journal of Medical Human Genetics</i> , 2014, 15, 379-385.	0.5	7
9	Prenatal genetic testing, counseling and follow-up of 33 Egyptian pregnant females with history of mucopolysaccharidoses. <i>Egyptian Journal of Medical Human Genetics</i> , 2015, 16, 159-163.	0.5	5
10	Phenotypical variability in glycogen storage disease type III with a recurrent <i>AGL</i> mutation c.750â€“753delAGAC. <i>Muscle and Nerve</i> , 2011, 43, 451-451.	1.0	3
11	Four novel mutations in the N-acetylgalactosamine-6-sulfate sulfatase gene among Egyptian patients with Morquio A disease. <i>Gene</i> , 2017, 600, 48-54.	1.0	3
12	Mucopolysaccharidoses diagnosis in the era of enzyme replacement therapy in Egypt. <i>Heliyon</i> , 2021, 7, e07830.	1.4	3
13	The Effect of Diet on Antioxidant Status in Patients with Galactosemia. <i>Journal of Medical Sciences (Faisalabad, Pakistan)</i> , 2006, 6, 452-457.	0.0	2
14	Clinical, Biochemical, and Molecular Characterization of Metachromatic Leukodystrophy Among Egyptian Pediatric Patients: Expansion of the ARSA Mutational Spectrum. <i>Journal of Molecular Neuroscience</i> , 2021, 71, 1112-1130.	1.1	1