

Joe Tr Clarke

List of Publications by Year in descending order

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Version: 2024-02-01

25
papers

1,524
citations

430874

18
h-index

677142

22
g-index

27
all docs

27
docs citations

27
times ranked

1824
citing authors

#	ARTICLE	IF	CITATIONS
1	Narrative Review: Fabry Disease. <i>Annals of Internal Medicine</i> , 2007, 146, 425.	3.9	177
2	The Natural History of Juvenile or Subacute GM2 Gangliosidosis: 21 New Cases and Literature Review of 134 Previously Reported. <i>Pediatrics</i> , 2006, 118, e1550-e1562.	2.1	165
3	Mutation analysis of PEX7 in 60 probands with rhizomelic chondrodysplasia punctata and functional correlations of genotype with phenotype. <i>Human Mutation</i> , 2002, 20, 284-297.	2.5	122
4	Early treatment of Menkes disease with parenteral Cooper-Histidine: Long-term follow-up of four treated patients. , 1998, 76, 154-164.		109
5	An overview of L-2-hydroxyglutarate dehydrogenase gene (L2HGDH) variants: a genotype-phenotype study. <i>Human Mutation</i> , 2010, 31, 380-390.	2.5	108
6	Efficient analysis of urinary glycosaminoglycans by LC-MS/MS in mucopolysaccharidoses type I, II and VI. <i>Molecular Genetics and Metabolism</i> , 2011, 102, 49-56.	1.1	96
7	Early copper-histidine treatment for Menkes disease. <i>Nature Genetics</i> , 1996, 12, 11-13.	21.4	94
8	How well does urinary lyso-Gb3 function as a biomarker in Fabry disease?. <i>Clinica Chimica Acta</i> , 2010, 411, 1906-1914.	1.1	94
9	Urinary globotriaosylceramide excretion correlates with the genotype in children and adults with Fabry disease. <i>Molecular Genetics and Metabolism</i> , 2008, 93, 331-340.	1.1	88
10	An improved method for glycosaminoglycan analysis by LC-MS/MS of urine samples collected on filter paper. <i>Clinica Chimica Acta</i> , 2012, 413, 771-778.	1.1	62
11	Urinary biomarker investigation in children with Fabry disease using tandem mass spectrometry. <i>Clinica Chimica Acta</i> , 2015, 438, 195-204.	1.1	62
12	UPLC-MS/MS detection of disaccharides derived from glycosaminoglycans as biomarkers of mucopolysaccharidoses. <i>Analytica Chimica Acta</i> , 2016, 936, 139-148.	5.4	53
13	Late onset Leigh syndrome and ataxia due to a T to C mutation at bp 9,185 of mitochondrial DNA. <i>American Journal of Medical Genetics, Part A</i> , 2007, 143A, 808-816.	1.2	51
14	Novel mutations in dihydrolipoamide dehydrogenase deficiency in two cousins with borderline-normal PDH complex activity. <i>American Journal of Medical Genetics, Part A</i> , 2006, 140A, 1542-1552.	1.2	48
15	Clinical and genetic aspects of trigonocephaly: A study of 25 cases. , 2002, 117A, 127-135.		41
16	Systolic Myocardial Mechanics in Patients with Anderson's Fabry Disease with and without Left Ventricular Hypertrophy and in Comparison to Nonobstructive Hypertrophic Cardiomyopathy. <i>Echocardiography</i> , 2012, 29, 810-817.	0.9	39
17	Organic Acidurias and Related Abnormalities. <i>Critical Reviews in Clinical Laboratory Sciences</i> , 1995, 32, 377-429.	6.1	29
18	A Survey of the Pain Experienced by Males and Females with Fabry Disease. <i>Pain Research and Management</i> , 2006, 11, 185-192.	1.8	21

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19	Changes in plasma and urine globotriaosylceramide levels do not predict Fabry disease progression over 1 year of agalsidase alfa. <i>Genetics in Medicine</i> , 2013, 15, 983-989.	2.4	21
20	The use of agalsidase alfa enzyme replacement therapy in the treatment of Fabry disease. <i>Expert Opinion on Biological Therapy</i> , 2009, 9, 631-639.	3.1	18
21	Interstitial deletion of 1p22.2p31.1 and medium-chain acyl-CoA dehydrogenase deficiency in a patient with global developmental delay. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 1581-1586.	1.2	11
22	Left Ventricular Aneurysm Associated With Mucopolysaccharidosis Type VI Syndrome (Maroteaux-Lamy Syndrome). <i>Circulation</i> , 2007, 115, e60-2.	1.6	8
23	Managing sickle cell carrier results generated through newborn screening in Ontario: a precedent-setting policy story. <i>Genetics in Medicine</i> , 2017, 19, 625-627.	2.4	5
24	Agalsidase alfa therapy for Fabry disease. <i>Expert Review of Endocrinology and Metabolism</i> , 2007, 2, 147-154.	2.4	1
25	Congratulations on passing your specialty examinations. <i>Paediatrics and Child Health</i> , 2010, 15, 137-138.	0.6	0