

Eileen P Treacy

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

38
papers

638
citations

14
h-index

24
g-index

47
ext. papers

806
ext. citations

3.3
avg, IF

3.37
L-index

#	Paper	IF	Citations
38	Designing rare disease care pathways in the Republic of Ireland: a co-operative model.. <i>Orphanet Journal of Rare Diseases</i> , 2022 , 17, 162	4.2	
37	Abnormal -glycan fucosylation, galactosylation, and sialylation of IgG in adults with classical galactosemia, influence of dietary galactose intake. <i>JIMD Reports</i> , 2021 , 61, 76-88	1.9	0
36	Maple syrup urine disease: Clinical outcomes, metabolic control, and genotypes in a screened population after four decades of newborn bloodspot screening in the Republic of Ireland. <i>Journal of Inherited Metabolic Disease</i> , 2021 , 44, 639-655	5.4	5
35	Impact of trimethylaminuria on daily psychosocial functioning. <i>JIMD Reports</i> , 2021 , 57, 67-75	1.9	1
34	Galactokinase deficiency: lessons from the GalNet registry. <i>Genetics in Medicine</i> , 2021 , 23, 202-210	8.1	7
33	The role of primary care in management of rare diseases in Ireland. <i>Irish Journal of Medical Science</i> , 2020 , 189, 771-776	1.9	3
32	The Galactose Index measured in fibroblasts of GALT deficient patients distinguishes variant patients detected by newborn screening from patients with classical phenotypes. <i>Molecular Genetics and Metabolism</i> , 2020 , 129, 171-176	3.7	0
31	Deep phenotyping classical galactosemia: clinical outcomes and biochemical markers. <i>Brain Communications</i> , 2020 , 2, fcaa006	4.5	6
30	Rare Disease Research Partnership (RAinDRoP): a collaborative approach to identify the top 15 research priorities for rare diseases. <i>HRB Open Research</i> , 2020 , 3, 13	1.2	2
29	A retrospective review of the contribution of rare diseases to paediatric mortality in Ireland. <i>Orphanet Journal of Rare Diseases</i> , 2020 , 15, 311	4.2	2
28	The genetic and biochemical basis of trimethylaminuria in an Irish cohort. <i>JIMD Reports</i> , 2019 , 47, 35-40	1.9	4
27	Expanding the genetic and phenotypic spectrum of branched-chain amino acid transferase 2 deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 809-817	5.4	8
26	Challenges in diagnosing and managing adult patients with urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 1136-1146	5.4	15
25	Insights into the Pathophysiology of Infertility in Females with Classical Galactosaemia. <i>International Journal of Molecular Sciences</i> , 2019 , 20,	6.3	5
24	Two Uneventful Pregnancies in a Woman with Glutaric Aciduria Type 1. <i>JIMD Reports</i> , 2018 , 41, 29-36	1.9	5
23	Validation of an automated ultraperformance liquid chromatography IgG N-glycan analytical method applicable to classical galactosaemia. <i>Annals of Clinical Biochemistry</i> , 2018 , 55, 593-603	2.2	13
22	Profiling of intracellular metabolites produced from galactose and its potential for galactosemia research. <i>Orphanet Journal of Rare Diseases</i> , 2018 , 13, 146	4.2	2

21	Fertility in classical galactosaemia, a study of N-glycan, hormonal and inflammatory gene interactions. <i>Orphanet Journal of Rare Diseases</i> , 2018 , 13, 164	4.2	8
20	Neurocognitive assessments and long-term outcome in an adult with 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency. <i>Molecular Genetics and Metabolism Reports</i> , 2018 , 16, 31-35	1.8	1
19	Systematic Review and Meta-analysis of Intelligence Quotient in Early-Treated Individuals with Classical Galactosemia. <i>JIMD Reports</i> , 2017 , 37, 115-123	1.9	12
18	Bone Health in Classic Galactosemia: Systematic Review and Meta-Analysis. <i>JIMD Reports</i> , 2017 , 35, 87-96	6.9	13
17	Classical Galactosaemia and CDG, the N-Glycosylation Interface. A Review. <i>JIMD Reports</i> , 2017 , 34, 33-42	1.9	14
16	International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up. <i>Journal of Inherited Metabolic Disease</i> , 2017 , 40, 171-176	5.4	83
15	IgG N-Glycosylation Galactose Incorporation Ratios for the Monitoring of Classical Galactosaemia. <i>JIMD Reports</i> , 2016 , 27, 47-53	1.9	7
14	Classical galactosaemia: novel insights in IgG N-glycosylation and N-glycan biosynthesis. <i>European Journal of Human Genetics</i> , 2016 , 24, 976-84	5.3	35
13	Effects of temporary low-dose galactose supplements in children aged 5-12 y with classical galactosemia: a pilot study. <i>Pediatric Research</i> , 2015 , 78, 272-9	3.2	23
12	Clinical and genetic characterisation of infantile liver failure syndrome type 1, due to recessive mutations in LARS. <i>Journal of Inherited Metabolic Disease</i> , 2015 , 38, 1085-92	5.4	33
11	Clinical utility gene card for: Trimethylaminuria - update 2014. <i>European Journal of Human Genetics</i> , 2015 , 23,	5.3	20
10	Atypical Alstrom syndrome with novel ALMS1 mutations precluded by current diagnostic criteria. <i>European Journal of Medical Genetics</i> , 2014 , 57, 55-9	2.6	17
9	N-glycan abnormalities in children with galactosemia. <i>Journal of Proteome Research</i> , 2014 , 13, 385-94	5.6	45
8	Fertility preservation in female classic galactosemia patients. <i>Orphanet Journal of Rare Diseases</i> , 2013 , 8, 107	4.2	29
7	Leptin levels in children and adults with classic galactosaemia. <i>JIMD Reports</i> , 2013 , 9, 125-131	1.9	10
6	Galactosemia, a single gene disorder with epigenetic consequences. <i>Pediatric Research</i> , 2010 , 67, 286-92	3.2	55
5	Outcomes of siblings with classical galactosemia. <i>Journal of Pediatrics</i> , 2009 , 154, 721-6	3.6	96
4	Polymorphisms of the Flavin containing monooxygenase 3 (FMO3) gene do not predispose to essential hypertension in Caucasians. <i>BMC Medical Genetics</i> , 2005 , 6, 41	2.1	16

3	In vivo variability of TMA oxidation is partially mediated by polymorphisms of the FMO3 gene. <i>Molecular Genetics and Metabolism</i> , 2001 , 73, 224-9	3.7	4 ¹
2	Rare Disease Research Partnership (RAinDRoP): a collaborative approach to identify research priorities for rare diseases in Ireland. <i>HRB Open Research</i> , 3, 13	1.2	1
1	Management of pregnancy in a patient with long-chain 3-hydroxyacyl CoA dehydrogenase deficiency. <i>JIMD Reports</i> ,	1.9	