Eileen P Treacy

List of Publications by Citations

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	Outcomes of siblings with classical galactosemia. *Journal of Pediatrics*, **2009**, 154, 721-6	3.6	96
37	International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up. *Journal of Inherited Metabolic Disease*, **2017**, 40, 171-176	5.4	83
36	Galactosemia, a single gene disorder with epigenetic consequences. *Pediatric Research*, **2010**, 67, 286-92	3.2	55
35	N-glycan abnormalities in children with galactosemia. *Journal of Proteome Research*, **2014**, 13, 385-94	5.6	45
34	In vivo variability of TMA oxidation is partially mediated by polymorphisms of the FMO3 gene. *Molecular Genetics and Metabolism*, **2001**, 73, 224-9	3.7	41
33	Classical galactosaemia: novel insights in IgG N-glycosylation and N-glycan biosynthesis. *European Journal of Human Genetics*, **2016**, 24, 976-84	5.3	35
32	Clinical and genetic characterisation of infantile liver failure syndrome type 1, due to recessive mutations in LARS. *Journal of Inherited Metabolic Disease*, **2015**, 38, 1085-92	5.4	33
31	Fertility preservation in female classic galactosemia patients. *Orphanet Journal of Rare Diseases*, **2013**, 8, 107	4.2	29
30	Effects of temporary low-dose galactose supplements in children aged 5-12 y with classical galactosemia: a pilot study. *Pediatric Research*, **2015**, 78, 272-9	3.2	23
29	Clinical utility gene card for: Trimethylaminuria - update 2014. *European Journal of Human Genetics*, **2015**, 23,	5.3	20
28	Atypical Alstrom syndrome with novel ALMS1 mutations precluded by current diagnostic criteria. *European Journal of Medical Genetics*, **2014**, 57, 55-9	2.6	17
27	Polymorphisms of the Flavin containing monooxygenase 3 (FMO3) gene do not predispose to essential hypertension in Caucasians. *BMC Medical Genetics*, **2005**, 6, 41	2.1	16
26	Challenges in diagnosing and managing adult patients with urea cycle disorders. *Journal of Inherited Metabolic Disease*, **2019**, 42, 1136-1146	5.4	15
25	Classical Galactosaemia and CDG, the N-Glycosylation Interface. A Review. *JIMD Reports*, **2017**, 34, 33-42	1.9	14
24	Bone Health in Classic Galactosemia: Systematic Review and Meta-Analysis. *JIMD Reports*, **2017**, 35, 87-96	1.9	13
23	Validation of an automated ultraperformance liquid chromatography IgG N-glycan analytical method applicable to classical galactosaemia. *Annals of Clinical Biochemistry*, **2018**, 55, 593-603	2.2	13
22	Systematic Review and Meta-analysis of Intelligence Quotient in Early-Treated Individuals with Classical Galactosemia. *JIMD Reports*, **2017**, 37, 115-123	1.9	12

21	Leptin levels in children and adults with classic galactosaemia. *JIMD Reports*, **2013**, 9, 125-131	1.9	10
20	Expanding the genetic and phenotypic spectrum of branched-chain amino acid transferase 2 deficiency. *Journal of Inherited Metabolic Disease*, **2019**, 42, 809-817	5.4	8
19	Fertility in classical galactosaemia, a study of N-glycan, hormonal and inflammatory gene interactions. *Orphanet Journal of Rare Diseases*, **2018**, 13, 164	4.2	8
18	IgG N-Glycosylation Galactose Incorporation Ratios for the Monitoring of Classical Galactosaemia. *JIMD Reports*, **2016**, 27, 47-53	1.9	7
17	Galactokinase deficiency: lessons from the GalNet registry. *Genetics in Medicine*, **2021**, 23, 202-210	8.1	7
16	Deep phenotyping classical galactosemia: clinical outcomes and biochemical markers. *Brain Communications*, **2020**, 2, fcaa006	4.5	6
15	Two Uneventful Pregnancies in a Woman with Glutaric Aciduria Type 1. *JIMD Reports*, **2018**, 41, 29-36	1.9	5
14	Insights into the Pathophysiology of Infertility in Females with Classical Galactosaemia. *International Journal of Molecular Sciences*, **2019**, 20,	6.3	5
13	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. *Journal of Inherited Metabolic Disease*, **2021**, 44, 639-655	5.4	5
12	The genetic and biochemical basis of trimethylaminuria in an Irish cohort. *JIMD Reports*, **2019**, 47, 35-40	1.9	4
11	The role of primary care in management of rare diseases in Ireland. *Irish Journal of Medical Science*, **2020**, 189, 771-776	1.9	3
10	Profiling of intracellular metabolites produced from galactose and its potential for galactosemia research. *Orphanet Journal of Rare Diseases*, **2018**, 13, 146	4.2	2
9	Rare Disease Research Partnership (RAinDRoP): a collaborative approach to identify the top 15 research priorities for rare diseases. *HRB Open Research*, **2020**, 3, 13	1.2	2
8	A retrospective review of the contribution of rare diseases to paediatric mortality in Ireland. *Orphanet Journal of Rare Diseases*, **2020**, 15, 311	4.2	2
7	Rare Disease Research Partnership (RAinDRoP): a collaborative approach to identify research priorities for rare diseases in Ireland. *HRB Open Research*,3, 13	1.2	1
6	Impact of trimethylaminuria on daily psychosocial functioning. *JIMD Reports*, **2021**, 57, 67-75	1.9	1
5	Neurocognitive assessments and long-term outcome in an adult with 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency. *Molecular Genetics and Metabolism Reports*, **2018**, 16, 31-35	1.8	1
4	The Galactose Index measured in fibroblasts of GALT deficient patients distinguishes variant patients detected by newborn screening from patients with classical phenotypes. *Molecular Genetics and Metabolism*, **2020**, 129, 171-176	3.7	0

3	Abnormal -glycan fucosylation, galactosylation, and sialylation of IgG in adults with classical galactosemia, influence of dietary galactose intake. *JIMD Reports*, **2021**, 61, 76-88	1.9	0
2	Management of pregnancy in a patient with long-chain 3-hydroxyacyl CoA dehydrogenase deficiency. *JIMD Reports*,	1.9	
1	Designing rare disease care pathways in the Republic of Ireland: a co-operative model.. *Orphanet Journal of Rare Diseases*, **2022**, 17, 162	4.2	