Francois J M Eyskens

List of Publications by Year
in descending order

57	1,835	304743 22	265206 42
papers	citations	h-index	g-index

58	58	58	2801
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Oral pharmacological chaperone migalastat compared with enzyme replacement therapy in Fabry disease: 18-month results from the randomised phase III ATTRACT study. Journal of Medical Genetics, 2017, 54, 288-296.	3.2	262
2	Respiratory chain complex V deficiency due to a mutation in the assembly gene ATP12. Journal of Medical Genetics, 2004, 41, 120-124.	3.2	175
3	International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and followâ€up. Journal of Inherited Metabolic Disease, 2017, 40, 171-176.	3.6	132
4	Mutation of the iron-sulfur cluster assembly gene IBA57 causes severe myopathy and encephalopathy. Human Molecular Genetics, 2013, 22, 2590-2602.	2.9	103
5	Belgian Fabry Study. Stroke, 2010, 41, 863-868.	2.0	99
6	Deletion of PREPL, a Gene Encoding a Putative Serine Oligopeptidase, in Patients with Hypotonia-Cystinuria Syndrome. American Journal of Human Genetics, 2006, 78, 38-51.	6.2	87
7	Dihydropyrimidinase deficiency: Phenotype, genotype and structural consequences in 17 patients. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 639-648.	3.8	67
8	Middelheim Fabry Study (MiFaS): A retrospective Belgian study on the prevalence of Fabry disease in young patients with cryptogenic stroke. Clinical Neurology and Neurosurgery, 2007, 109, 479-484.	1.4	60
9	Neurocognitive outcome in tyrosinemia type 1 patients compared to healthy controls. Orphanet Journal of Rare Diseases, 2016, 11, 87.	2.7	60
10	Multiplex ligationâ€dependent probe amplification to detect subtelomeric rearrangements in routine diagnostics. Clinical Genetics, 2006, 69, 58-64.	2.0	57
11	Oral Migalastat HCl Leads to Greater Systemic Exposure and Tissue Levels of Active Î±-Galactosidase A in Fabry Patients when Co-Administered with Infused Agalsidase. PLoS ONE, 2015, 10, e0134341.	2.5	50
12	Identifying Nonâ€“Duchenne Muscular Dystrophyâ€“Positive and False Negative Results in Prior Duchenne Muscular Dystrophy Newborn Screening Programs. JAMA Neurology, 2016, 73, 111.	9.0	48
13	Phase I/II Trial of Liverâ€“derived Mesenchymal Stem Cells in Pediatric Liverâ€“based Metabolic Disorders: A Prospective, Open Label, Multicenter, Partially Randomized, Safety Study of One Cycle of Heterologous Human Adult Liverâ€“derived Progenitor Cells (HepaStem) in Urea Cycle Disorders and Crigler-Najjar Syndrome Patients. Transplantation, 2019, 103, 1903-1915.	1.0	47
14	Neonatal Thyroid-Stimulating Hormone Concentrations in Belgium: A Useful Indicator for Detecting Mild Iodine Deficiency?. PLoS ONE, 2012, 7, e47770.	2.5	44
15	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	5.3	42
16	A nonsense mutation in the 3-hydroxy-3-methylglutaryl-CoA lyase gene produces exon skipping in two patients of different origin with 3-hydroxy-3-methylglutaryl-CoA lyase deficiency. Biochemical Journal, 1997, 323, 329-335.	3.7	39
17	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”A successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93-106.	3.6	35
18	Hyperoxaluria with hyperglycoluria not due to alanine:glyoxylate aminotransferase defect: A novel type of primary hyperoxaluria. Kidney International, 1996, 50, 1747-1752.	5.2	32

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19	Phenotypical characterization of Î±-galactosidase A gene mutations identified in a large Fabry disease screening program in stroke in the young. Clinical Neurology and Neurosurgery, 2013, 115, 1088-1093.	1.4	31
20	Efficacy, safety and population pharmacokinetics of sapropterin in PKU patients <4Âyears: results from the SPARK open-label, multicentre, randomized phase IIIb trial. Orphanet Journal of Rare Diseases, 2017, 12, 47.	2.7	26
21	Dietary practices in pyridoxine non-responsive homocystinuria: A European survey. Molecular Genetics and Metabolism, 2013, 110, 454-459.	1.1	23
22	Neurologic sequelae in transient nonketotic hyperglycinemia of the neonate. Journal of Pediatrics, 1992, 121, 620-621.	1.8	22
23	Data mining methods for classification of Medium-Chain Acyl-CoA dehydrogenase deficiency (MCADD) using non-derivatized tandem MS neonatal screening data. Journal of Biomedical Informatics, 2011, 44, 319-325.	4.3	22
24	Long-term follow-up on the effect of combined therapy of bile acids and statins in the treatment of cerebrotendinous xanthomatosis: A case report. Clinical Neurology and Neurosurgery, 2014, 118, 9-11.	1.4	22
25	Bone Health in Classic Galactosemia: Systematic Review and Meta-Analysis. JIMD Reports, 2017, 35, 87-96.	1.5	22
26	Novel Mutations in the PEX2 Gene of Four Unrelated Patients with a Peroxisome Biogenesis Disorder. Pediatric Research, 2004, 55, 431-436.	2.3	21
27	Genotype-phenotype correlation in PEX5-deficient peroxisome biogenesis defective cell lines. Human Mutation, 2009, 30, 93-98.	2.5	21
28	Cytomegalovirus DNA Detection in Guthrie Cards. Otology and Neurotology, 2009, 30, 943-949.	1.3	19
29	Emotional and behavioral problems, quality of life and metabolic control in NTBC-treated Tyrosinemia type 1 patients. Orphanet Journal of Rare Diseases, 2019, 14, 285.	2.7	19
30	Dietary practices in propionic acidemia: A European survey. Molecular Genetics and Metabolism Reports, 2017, 13, 83-89.	1.1	18
31	Mitochondrial mosaics in the liver of 3 infants with mtDNA defects. BMC Clinical Pathology, 2009, 9, 4.	1.8	15
32	Long-term efficacy and safety of sapropterin in patients who initiated sapropterin at <â€‰4Âyears of age with phenylketonuria: results of the 3-year extension of the SPARK open-label, multicentre, randomised phase IIIb trial. Orphanet Journal of Rare Diseases, 2021, 16, 341.	2.7	13
33	Dietary practices in isovaleric acidemia: A European survey. Molecular Genetics and Metabolism Reports, 2017, 12, 16-22.	1.1	12
34	Urinary Excretion of Purine and Pyrimidine Metabolites in the Neonate. Pediatric Research, 1993, 34, 762-766.	2.3	8
35	Cognitive functioning and psychiatric disorders in children with a metabolic disease. European Child and Adolescent Psychiatry, 2006, 15, 207-213.	4.7	8
36	Methylmalonic acidaemia in pregnancy. BMJ Case Reports, 2014, 2014, bcr2014203723-bcr2014203723.	0.5	8

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37	Galactosidase Alpha p.A143T Variant Fabry Disease May Result in a Phenotype With Multifocal Microvascular Cerebral Involvement at a Young Age. Frontiers in Neurology, 2018, 9, 336.	2.4	8
38	Dietary practices in methylmalonic acidaemia: a European survey. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 147-155.	0.9	8
39	Peroxisome Mosaics. Advances in Experimental Medicine and Biology, 2003, 544, 97-106.	1.6	8
40	Unusual enzyme findings in five patients with metabolic profiles suggestive of succinic semialdehyde dehydrogenase deficiency (4-hydroxybutyric aciduria). Journal of Inherited Metabolic Disease, 1998, 21, 255-261.	3.6	7
41	Exclusion ofOGDH andBMP4 as candidate genes in two siblings with autosomal recessive DOOR syndrome. American Journal of Medical Genetics, Part A, 2007, 143A, 763-767.	1.2	7
42	Newborn Screening for Lysosomal Storage Disorders in Belgium. FIRE Forum for International Research in Education, 2017, 5, 232640981774423.	0.7	7
43	Once every 4 weeks - 2 mg/kg of pegunigalsidase alfa for treating Fabry disease Preliminary results of a phase 3 study. Molecular Genetics and Metabolism, 2019, 126, S73.	1.1	5
44	Raising Awareness of False Positive Newborn Screening Results Arising from Pivalate-Containing Creams and Antibiotics in Europe When Screening for Isovaleric Acidaemia. International Journal of Neonatal Screening, 2018, 4, 8.	3.2	4
45	RARE INBORN ERRORS OF METABOLISM IN ADULTS: THE LYSOSOMAL STORAGE DISORDERS. Acta Clinica Belgica, 2009, 64, 534-539.	1.2	3
46	A phase 2a study to investigate the effect of a single dose of migalastat HCl, a pharmacological chaperone, on agalsidase activity in subjects with Fabry disease. Molecular Genetics and Metabolism, 2013, 108, S96.	1.1	3
47	Response to Letter Regarding Belgian Fabry Study: Prevalence of Fabry Disease in a Cohort of 1000 Young Patients With Cerebrovascular Disease. Stroke, 2011, 42, .	2.0	2
48	Carnitine Deficiency and Pregnancy. Case Reports in Obstetrics and Gynecology, 2015, 2015, 1-4.	0.3	2
49	A marked difference between two populations under mass screening of neonatal TSH and biotinidase activity. Accreditation and Quality Assurance, 2002, 7, 498-506.	0.8	1
50	Girl With Tyrosinemia Type 1 and Executive Dysfunctions Treated With Methylphenidate. FIRE Forum for International Research in Education, 2018, 6, 232640981878539.	0.7	1
51	Prenatal exclusion of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency by direct detection of the mutation with PCR. Prenatal Diagnosis, 1992, 12, 74-76.	2.3	0
52	The use of split-sample design for performance evaluation of screening kits. Accreditation and Quality Assurance, 2004, 9, 164-167.	0.8	0
53	Data Mining Methods for Classification of Medium-Chain ACYL-COA Dehydrogenase Deficiency (MCADD) using Non-Derivatized Tandem Ms Neonatal Screening Data. Pediatric Research, 2011, 70, 576-576.	2.3	0
54	Multiple sclerosis as a misdiagnosis of Fabry disease. Molecular Genetics and Metabolism, 2019, 126, S53.	1.1	0

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55	Recurrent acute coronary syndrome, polymorphic premature ventricular complexes and a son with a (mis)diagnosis of multiple sclerosis. Acta Cardiologica, 2020, 75, 467-468.	0.9	0
56	Vertebral Tongue-Like Deformity in Mucopolysaccharidosis VI. Journal of the Belgian Society of Radiology, 2021, 105, 54.	0.3	0
57	Disturbances of Valine Metabolism in Patients with Peroxisomal Biogenesis Disorders. Advances in Experimental Medicine and Biology, 2003, 544, 73-74.	1.6	0