

François Feillet

List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/5656876/publications.pdf>

Version: 2024-02-01

76
papers

3,399
citations

159358

30
h-index

149479

56
g-index

83
all docs

83
docs citations

83
times ranked

3405
citing authors

#	ARTICLE	IF	CITATIONS
1	Efficacy of sapropterin dihydrochloride (tetrahydrobiopterin, 6R-BH4) for reduction of phenylalanine concentration in patients with phenylketonuria: a phase III randomised placebo-controlled study. <i>Lancet, The</i> , 2007, 370, 504-510.	6.3	277
2	Key European guidelines for the diagnosis and management of patients with phenylketonuria. <i>Lancet Diabetes and Endocrinology</i> , 2017, 5, 743-756.	5.5	272
3	Genotypic and phenotypic spectrum of pyridoxine-dependent epilepsy (ALDH7A1 deficiency). <i>Brain</i> , 2010, 133, 2148-2159.	3.7	219
4	A Phase 3 Trial of Sebelipase Alfa in Lysosomal Acid Lipase Deficiency. <i>New England Journal of Medicine</i> , 2015, 373, 1010-1020.	13.9	212
5	Molecular genetics of tetrahydrobiopterin-responsive phenylalanine hydroxylase deficiency. <i>Human Mutation</i> , 2008, 29, 167-175.	1.1	158
6	Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. <i>Brain</i> , 2014, 137, 1350-1360.	3.7	151
7	The Genetic Landscape and Epidemiology of Phenylketonuria. <i>American Journal of Human Genetics</i> , 2020, 107, 234-250.	2.6	138
8	Optimizing the use of sapropterin (BH4) in the management of phenylketonuria. <i>Molecular Genetics and Metabolism</i> , 2009, 96, 158-163.	0.5	121
9	Mutations in SLC13A5 Cause Autosomal-Recessive Epileptic Encephalopathy with Seizure Onset in the First Days of Life. <i>American Journal of Human Genetics</i> , 2014, 95, 113-120.	2.6	112
10	Management of phenylketonuria in Europe: Survey results from 19 countries. <i>Molecular Genetics and Metabolism</i> , 2010, 99, 109-115.	0.5	94
11	Clinical, laboratory and molecular findings and long-term follow-up data in 96 French patients with PMM2-CDC (phosphomannomutase 2-congenital disorder of glycosylation) and review of the literature. <i>Journal of Medical Genetics</i> , 2017, 54, 843-851.	1.5	88
12	Challenges and Pitfalls in the Management of Phenylketonuria. <i>Pediatrics</i> , 2010, 126, 333-341.	1.0	72
13	Fluctuations in phenylalanine concentrations in phenylketonuria: A review of possible relationships with outcomes. <i>Molecular Genetics and Metabolism</i> , 2013, 110, 418-423.	0.5	69
14	Long-term Follow-up and Outcome of Phenylketonuria Patients on Sapropterin: A Retrospective Study. <i>Pediatrics</i> , 2013, 131, e1881-e1888.	1.0	68
15	A PRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. <i>Nature Communications</i> , 2018, 9, 67.	5.8	64
16	Nutritional issues in treating phenylketonuria. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 659-664.	1.7	58
17	Vitamin D-Dependent Rickets Type 1B (25-Hydroxylase Deficiency): A Rare Condition or a Misdiagnosed Condition?. <i>Journal of Bone and Mineral Research</i> , 2017, 32, 1893-1899.	3.1	57
18	Consensus guidelines for the diagnosis and management of pyridoxine-dependent epilepsy due to Î±-aminoadipic semialdehyde dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 178-192.	1.7	47

#	ARTICLE	IF	CITATIONS
19	Maternal phenylketonuria: low phenylalaninemia might increase the risk of intra uterine growth retardation. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 993-999.	1.7	44
20	International best practice for the evaluation of responsiveness to sapropterin dihydrochloride in patients with phenylketonuria. <i>Molecular Genetics and Metabolism</i> , 2019, 127, 1-11.	0.5	44
21	Management of Phenylketonuria and Hyperphenylalaninemia. <i>Journal of Nutrition</i> , 2007, 137, 1561S-1563S.	1.3	43
22	Outcomes beyond phenylalanine: An international perspective. <i>Molecular Genetics and Metabolism</i> , 2010, 99, S79-S85.	0.5	43
23	Fructose 1,6-bisphosphatase deficiency: clinical, biochemical and genetic features in French patients. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 881-887.	1.7	43
24	A noncoding RNA modulator potentiates phenylalanine metabolism in mice. <i>Science</i> , 2021, 373, 662-673.	6.0	42
25	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. <i>Genetics in Medicine</i> , 2019, 21, 2025-2035.	1.1	40
26	Efficacy and safety of BH4 before the age of 4 years in patients with mild phenylketonuria. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 975-981.	1.7	38
27	Plasma cholesterol and endogenous cholesterol synthesis during refeeding in anorexia nervosa. <i>Clinica Chimica Acta</i> , 2000, 294, 45-56.	0.5	37
28	Pharmacokinetics of Sapropterin in Patients with Phenylketonuria. <i>Clinical Pharmacokinetics</i> , 2008, 47, 817-825.	1.6	37
29	Mapping the functional landscape of frequent phenylalanine hydroxylase (PAH) genotypes promotes personalised medicine in phenylketonuria. <i>Journal of Medical Genetics</i> , 2015, 52, 175-185.	1.5	37
30	Can untreated PKU patients escape from intellectual disability? A systematic review. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 149.	1.2	36
31	Management of adult patients with phenylketonuria: survey results from 24 countries. <i>European Journal of Pediatrics</i> , 2015, 174, 119-127.	1.3	32
32	Use of sapropterin dihydrochloride in maternal phenylketonuria. A European experience of eight cases. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 753-762.	1.7	28
33	Neonatal screening and long-term follow-up of phenylketonuria: the French database. <i>Early Human Development</i> , 2001, 65, 149-158.	0.8	27
34	Interaction between methionine synthase isoforms and MMACHC: characterization in cblG-variant, cblG and cblC inherited causes of megaloblastic anaemia. <i>Human Molecular Genetics</i> , 2013, 22, 4591-4601.	1.4	27
35	Is the Phenylalanine-Restricted Diet a Risk Factor for Overweight or Obesity in Patients with Phenylketonuria (PKU)? A Systematic Review and Meta-Analysis. <i>Nutrients</i> , 2021, 13, 3443.	1.7	27
36	Life-threatening methylenetetrahydrofolate reductase (MTHFR) deficiency with extremely early onset: Characterization of two novel mutations in compound heterozygous patients. <i>Molecular Genetics and Metabolism</i> , 2010, 100, 143-148.	0.5	25

#	ARTICLE	IF	CITATIONS
37	Luminal expression of cubilin is impaired in Imerslund-Grasbeck syndrome with compound AMN mutations in intron 3 and exon 7. <i>Haematologica</i> , 2011, 96, 1715-1719.	1.7	24
38	Gastric intrinsic factor deficiency with combined GIF heterozygous mutations and FUT2 secretor variant. <i>Biochimie</i> , 2013, 95, 995-1001.	1.3	23
39	Diagnostic and management practices for phenylketonuria in 19 countries of the South and Eastern European Region: survey results. <i>European Journal of Pediatrics</i> , 2016, 175, 261-272.	1.3	23
40	SIRT1 activation rescues the mislocalization of RNA-binding proteins and cognitive defects induced by inherited cobalamin disorders. <i>Metabolism: Clinical and Experimental</i> , 2019, 101, 153992.	1.5	23
41	Genotype-phenotype associations in French patients with phenylketonuria and importance of genotype for full assessment of tetrahydrobiopterin responsiveness. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 158.	1.2	21
42	Efficacy of low dose nitisinone in the management of alkaptonuria. <i>Molecular Genetics and Metabolism</i> , 2019, 127, 184-190.	0.5	21
43	Neurological manifestations in adults with phenylketonuria: new cases and review of the literature. <i>Journal of Neurology</i> , 2020, 267, 531-542.	1.8	21
44	Maternal phenylketonuria: the French survey. <i>European Journal of Pediatrics</i> , 2004, 163, 540-546.	1.3	20
45	Neuropsychological Profile of Children with Early and Continuously Treated Phenylketonuria: Systematic Review and Future Approaches. <i>Journal of the International Neuropsychological Society</i> , 2019, 25, 624-643.	1.2	16
46	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. <i>Genetics in Medicine</i> , 2020, 22, 199-209.	1.1	14
47	Evaluation of neonatal BH4 loading test in neonates screened for hyperphenylalaninemia. <i>Early Human Development</i> , 2008, 84, 561-567.	0.8	13
48	Transition from pediatric to adult care in adolescents with hereditary metabolic diseases: Specific guidelines from the French network for rare inherited metabolic diseases (G2M). <i>Archives De Pédiatrie</i> , 2018, 25, 344-349.	0.4	13
49	Hearing impairment as an early sign of alpha-mannosidosis in children with a mild phenotype: Report of seven new cases. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1756-1763.	0.7	13
50	Exome sequencing of cases with neural tube defects identifies candidate genes involved in one-carbon/vitamin B12 metabolisms and Sonic Hedgehog pathway. <i>Human Genetics</i> , 2019, 138, 703-713.	1.8	13
51	Vitamin B-12 and liver activity and expression of methionine synthase are decreased in fetuses with neural tube defects. <i>American Journal of Clinical Nutrition</i> , 2019, 109, 674-683.	2.2	13
52	Diagnostic and therapeutic recommendations for the treatment of hyperphenylalaninemia in patients 0-4 years of age. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 173.	1.2	12
53	The Kuvan® Adult Maternal Paediatric European Registry (KAMPER) Multinational Observational Study: Baseline and 1-Year Data in Phenylketonuria Patients Responsive to Sapropterin. <i>JIMD Reports</i> , 2015, 23, 35-43.	0.7	11
54	Deciphering exome sequencing data: Bringing mitochondrial DNA variants to light. <i>Human Mutation</i> , 2019, 40, 2430-2443.	1.1	11

#	ARTICLE	IF	CITATIONS
55	Sebelipase alfa in children and adults with lysosomal acid lipase deficiency: Final results of the ARISE study. <i>Journal of Hepatology</i> , 2022, 76, 577-587.	1.8	11
56	Diagnostic yield of clinical exome sequencing as a first-tier genetic test for the diagnosis of genetic disorders in pediatric patients: results from a referral center study. <i>Human Genetics</i> , 2022, 141, 1269-1278.	1.8	10
57	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B12. <i>Clinical Epigenetics</i> , 2022, 14, 52.	1.8	10
58	Health Status of French Young Patients with Inborn Errors of Metabolism with Lifelong Restricted Diet. <i>Journal of Pediatrics</i> , 2020, 220, 184-192.e6.	0.9	9
59	Issues with European guidelines for phenylketonuria – Authors' reply. <i>Lancet Diabetes and Endocrinology</i> , 2017, 5, 683-684.	5.5	8
60	Bone mineral density is within normal range in most adult phenylketonuria patients. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 251-258.	1.7	8
61	Dietary practices in methylmalonic acidemia: a European survey. <i>Journal of Pediatric Endocrinology and Metabolism</i> , 2020, 33, 147-155.	0.4	8
62	Tetrahydrobiopterin (BH4) responsiveness in neonates with hyperphenylalaninemia: A semi-mechanistically-based, nonlinear mixed-effect modeling. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 564-569.	0.5	7
63	Analysis of fibroblasts from patients with cblC and cblG genetic defects of cobalamin metabolism reveals global dysregulation of alternative splicing. <i>Human Molecular Genetics</i> , 2020, 29, 1969-1985.	1.4	7
64	Fructose 1,6-bisphosphatase deficiency causes fatty liver disease and requires long-term hepatic follow-up. <i>Journal of Inherited Metabolic Disease</i> , 2022, 45, 215-222.	1.7	7
65	Rapid identification of HEXA mutations in Tay-Sachs patients. <i>Biochemical and Biophysical Research Communications</i> , 2010, 392, 599-602.	1.0	6
66	Population and evolutionary genetics of the PAH locus to uncover overdominance and adaptive mechanisms in phenylketonuria: Results from a multiethnic study. <i>EBioMedicine</i> , 2020, 51, 102623.	2.7	6
67	A allelic loss-of-function SARS1 variant in children with neurodevelopmental delay, deafness, cardiomyopathy, and decompensation during fever. <i>Human Mutation</i> , 2021, 42, 1576-1583.	1.1	6
68	Cystathionine β -synthase genetic variant rs2124459 is associated with a reduced risk of cleft palate in French and Belgian populations. <i>Journal of Medical Genetics</i> , 2016, 53, 828-834.	1.5	5
69	Mutations in MTHFR and POLG impaired activity of the mitochondrial respiratory chain in 46-year-old twins with spastic paraparesis. <i>Journal of Human Genetics</i> , 2020, 65, 91-98.	1.1	5
70	Clinical, phenotypic and genetic landscape of case reports with genetically proven inherited disorders of vitamin B12 metabolism: A meta-analysis. <i>Cell Reports Medicine</i> , 2022, 3, 100670.	3.3	5
71	Prolonged 25-OH Vitamin D Deficiency Does Not Impair Bone Mineral Density in Adult Patients With Vitamin D 25-Hydroxylase Deficiency (CYP2R1). <i>Calcified Tissue International</i> , 2020, 107, 191-194.	1.5	4
72	Importance of the long non-coding RNA (lncRNA) transcript HULC for the regulation of phenylalanine hydroxylase and treatment of phenylketonuria. <i>Molecular Genetics and Metabolism</i> , 2022, 135, 171-178.	0.5	3

#	ARTICLE	IF	CITATIONS
73	Inherited metabolic disorders beyond the new generation sequencing era: the need for in-depth cellular and molecular phenotyping. <i>Human Genetics</i> , 2022, 141, 1235-1237.	1.8	3
74	Sapropterin in the Treatment of Phenylketonuria. <i>Clinical Medicine Insights Therapeutics</i> , 2010, 2, CMT.S2721.	0.4	2
75	The diagnostic rate of inherited metabolic disorders by exome sequencing in a cohort of 547 individuals with developmental disorders. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 29, 100812.	0.4	2
76	Prise en charge nutritionnelle des troubles du comportement alimentaire chez l'adolescent. <i>Nutrition Clinique Et Metabolisme</i> , 2005, 19, 247-253.	0.2	1