FranÃ§ois Feillet

List of Publications by Year
in descending order

76 papers	3,399 citations	159585 30 h-index	149698 56 g-index
83 all docs	83 docs citations	83 times ranked	3405 citing authors

#	Article	IF	Citations
1	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. Human Genetics, 2022, 141, 1269-1278.	3.8	10
2	Fructoseâ€1,6â€bisphosphatase deficiency causes fatty liver disease and requires longâ€term hepatic followâ€up. Journal of Inherited Metabolic Disease, 2022, 45, 215-222.	3.6	7
3	Sebelipase alfa in children and adults with lysosomal acid lipase deficiency: Final results of the ARISE study. Journal of Hepatology, 2022, 76, 577-587.	3.7	11
4	Importance of the long non-coding RNA (lncRNA) transcript HULC for the regulation of phenylalanine hydroxylase and treatment of phenylketonuria. Molecular Genetics and Metabolism, 2022, 135, 171-178.	1.1	3
5	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B12. Clinical Epigenetics, 2022, 14, 52.	4.1	10
6	Clinical, phenotypic and genetic landscape of case reports with genetically proven inherited disorders of vitamin B12 metabolism: A meta-analysis. Cell Reports Medicine, 2022, 3, 100670.	6.5	5
7	Inherited metabolic disorders beyond the new generation sequencing era: the need for in-depth cellular and molecular phenotyping. Human Genetics, 2022, 141, 1235-1237.	3.8	3
8	Consensus guidelines for the diagnosis and management of pyridoxineâ€dependent epilepsy due to Î±â€aminoadipic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 178-192.	3.6	47
9	A noncoding RNA modulator potentiates phenylalanine metabolism in mice. Science, 2021, 373, 662-673.	12.6	42
10	A biâ€allelic lossâ€ofâ€function SARS1 variant in children with neurodevelopmental delay, deafness, cardiomyopathy, and decompensation during fever. Human Mutation, 2021, 42, 1576-1583.	2.5	6
11	Is the Phenylalanine-Restricted Diet a Risk Factor for Overweight or Obesity in Patients with Phenylketonuria (PKU)? A Systematic Review and Meta-Analysis. Nutrients, 2021, 13, 3443.	4.1	27
12	The diagnostic rate of inherited metabolic disorders by exome sequencing in a cohort of 547 individuals with developmental disorders. Molecular Genetics and Metabolism Reports, 2021, 29, 100812.	1.1	2
13	Mutations in MTHFR and POLG impaired activity of the mitochondrial respiratory chain in 46-year-old twins with spastic paraparesis. Journal of Human Genetics, 2020, 65, 91-98.	2.3	5
14	The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209.	2.4	14
15	Neurological manifestations in adults with phenylketonuria: new cases and review of the literature. Journal of Neurology, 2020, 267, 531-542.	3.6	21
16	Bone mineral density is within normal range in most adult phenylketonuria patients. Journal of Inherited Metabolic Disease, 2020, 43, 251-258.	3.6	8
17	Population and evolutionary genetics of the PAH locus to uncover overdominance and adaptive mechanisms in phenylketonuria: Results from a multiethnic study. EBioMedicine, 2020, 51, 102623.	6.1	6
18	Dietary practices in methylmalonic acidaemia: a European survey. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 147-155.	0.9	8

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19	The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250.	6.2	138
20	Prolonged 25-OH Vitamin D Deficiency Does Not Impair Bone Mineral Density in Adult Patients With Vitamin D 25-Hydroxylase Deficiency (CYP2R1). Calcified Tissue International, 2020, 107, 191-194.	3.1	4
21	Health Status of French Young Patients with Inborn Errors of Metabolism with Lifelong Restricted Diet. Journal of Pediatrics, 2020, 220, 184-192.e6.	1.8	9
22	Analysis of fibroblasts from patients with cblC and cblG genetic defects of cobalamin metabolism reveals global dysregulation of alternative splicing. Human Molecular Genetics, 2020, 29, 1969-1985.	2.9	7
23	Deciphering exome sequencing data: Bringing mitochondrial DNA variants to light. Human Mutation, 2019, 40, 2430-2443.	2.5	11
24	SIRT1 activation rescues the mislocalization of RNA-binding proteins and cognitive defects induced by inherited cobalamin disorders. Metabolism: Clinical and Experimental, 2019, 101, 153992.	3.4	23
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. Genetics in Medicine, 2019, 21, 2025-2035.	2.4	40
26	Efficacy of low dose nitisinone in the management of alkaptonuria. Molecular Genetics and Metabolism, 2019, 127, 184-190.	1.1	21
27	Hearing impairment as an early sign of alphaâ€mannosidosis in children with a mild phenotype: Report of seven new cases. American Journal of Medical Genetics, Part A, 2019, 179, 1756-1763.	1.2	13
28	Exome sequencing of cases with neural tube defects identifies candidate genes involved in one-carbon/vitamin B12 metabolisms and Sonic Hedgehog pathway. Human Genetics, 2019, 138, 703-713.	3.8	13
29	Neuropsychological Profile of Children with Early and Continuously Treated Phenylketonuria: Systematic Review and Future Approaches. Journal of the International Neuropsychological Society, 2019, 25, 624-643.	1.8	16
30	International best practice for the evaluation of responsiveness to sapropterin dihydrochloride in patients with phenylketonuria. Molecular Genetics and Metabolism, 2019, 127, 1-11.	1.1	44
31	Vitamin B-12 and liver activity and expression of methionine synthase are decreased in fetuses with neural tube defects. American Journal of Clinical Nutrition, 2019, 109, 674-683.	4.7	13
32	A PRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. Nature Communications, 2018, 9, 67.	12.8	64
33	Can untreated PKU patients escape from intellectual disability? A systematic review. Orphanet Journal of Rare Diseases, 2018, 13, 149.	2.7	36
34	Diagnostic and therapeutic recommendations for the treatment of hyperphenylalaninemia in patients 0â€“4Âyears of age. Orphanet Journal of Rare Diseases, 2018, 13, 173.	2.7	12
35	Transition from pediatric to adult care in adolescents with hereditary metabolic diseases: Specific guidelines from the French network for rare inherited metabolic diseases (G2M). Archives De Pediatrie, 2018, 25, 344-349.	1.0	13
36	Key European guidelines for the diagnosis and management of patients with phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 743-756.	11.4	272

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37	Vitamin Dâ€“Dependent Rickets Type 1B (25-Hydroxylase Deficiency): A Rare Condition or a Misdiagnosed Condition?. Journal of Bone and Mineral Research, 2017, 32, 1893-1899.	2.8	57
38	Clinical, laboratory and molecular findings and long-term follow-up data in 96 French patients with PMM2-CDG (phosphomannomutase 2-congenital disorder of glycosylation) and review of the literature. Journal of Medical Genetics, 2017, 54, 843-851.	3.2	88
39	Issues with European guidelines for phenylketonuria â€“ Authors' reply. Lancet Diabetes and Endocrinology,the, 2017, 5, 683-684.	11.4	8
40	Cystathionine Î²-synthase genetic variant rs2124459 is associated with a reduced risk of cleft palate in French and Belgian populations. Journal of Medical Genetics, 2016, 53, 828-834.	3.2	5
41	Diagnostic and management practices for phenylketonuria in 19 countries of the South and Eastern European Region: survey results. European Journal of Pediatrics, 2016, 175, 261-272.	2.7	23
42	Genotype-phenotype associations in French patients with phenylketonuria and importance of genotype for full assessment of tetrahydrobiopterin responsiveness. Orphanet Journal of Rare Diseases, 2015, 10, 158.	2.7	21
43	Tetrahydrobiopterin (BH4) responsiveness in neonates with hyperphenylalaninemia: A semi-mechanistically-based, nonlinear mixed-effect modeling. Molecular Genetics and Metabolism, 2015, 114, 564-569.	1.1	7
44	Management of adult patients with phenylketonuria: survey results from 24 countries. European Journal of Pediatrics, 2015, 174, 119-127.	2.7	32
45	Mapping the functional landscape of frequentphenylalanine hydroxylase(PAH) genotypes promotes personalised medicine in phenylketonuria. Journal of Medical Genetics, 2015, 52, 175-185.	3.2	37
46	Fructose 1,6â€bisphosphatase deficiency: clinical, biochemical and genetic features in French patients. Journal of Inherited Metabolic Disease, 2015, 38, 881-887.	3.6	43
47	The KuvanÂ® Adult Maternal Paediatric European Registry (KAMPER) Multinational Observational Study: Baseline and 1-Year Data in Phenylketonuria Patients Responsive to Sapropterin. JIMD Reports, 2015, 23, 35-43.	1.5	11
48	A Phase 3 Trial of Sebelipase Alfa in Lysosomal Acid Lipase Deficiency. New England Journal of Medicine, 2015, 373, 1010-1020.	27.0	212
49	Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. Brain, 2014, 137, 1350-1360.	7.6	151
50	Mutations in SLC13A5 Cause Autosomal-Recessive Epileptic Encephalopathy with Seizure Onset in the First Days of Life. American Journal of Human Genetics, 2014, 95, 113-120.	6.2	112
51	Use of sapropterin dihydrochloride in maternal phenylketonuria. A European experience of eight cases. Journal of Inherited Metabolic Disease, 2014, 37, 753-762.	3.6	28
52	Gastric intrinsic factor deficiency with combined GIF heterozygous mutations and FUT2 secretor variant. Biochimie, 2013, 95, 995-1001.	2.6	23
53	Long-term Follow-up and Outcome of Phenylketonuria Patients on Sapropterin: A Retrospective Study. Pediatrics, 2013, 131, e1881-e1888.	2.1	68
54	Fluctuations in phenylalanine concentrations in phenylketonuria: A review of possible relationships with outcomes. Molecular Genetics and Metabolism, 2013, 110, 418-423.	1.1	69

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55	Interaction between methionine synthase isoforms and MMACHC: characterization in cblG-variant, cblG and cblC inherited causes of megaloblastic anaemia. Human Molecular Genetics, 2013, 22, 4591-4601.	2.9	27
56	Efficacy and safety of BH4 before the age of 4 years in patients with mild phenylketonuria. Journal of Inherited Metabolic Disease, 2012, 35, 975-981.	3.6	38
57	Maternal phenylketonuria: low phenylalaninemia might increase the risk of intra uterine growth retardation. Journal of Inherited Metabolic Disease, 2012, 35, 993-999.	3.6	44
58	Luminal expression of cubilin is impaired in Imerslund-Grasbeck syndrome with compound AMN mutations in intron 3 and exon 7. Haematologica, 2011, 96, 1715-1719.	3.5	24
59	Nutritional issues in treating phenylketonuria. Journal of Inherited Metabolic Disease, 2010, 33, 659-664.	3.6	58
60	Sapropterin in the Treatment of Phenylketonuria. Clinical Medicine Insights Therapeutics, 2010, 2, CMT.S2721.	0.4	2
61	Genotypic and phenotypic spectrum of pyridoxine-dependent epilepsy (ALDH7A1 deficiency). Brain, 2010, 133, 2148-2159.	7.6	219
62	Rapid identification of HEXA mutations in Tay-Sachs patients. Biochemical and Biophysical Research Communications, 2010, 392, 599-602.	2.1	6
63	Management of phenylketonuria in Europe: Survey results from 19 countries. Molecular Genetics and Metabolism, 2010, 99, 109-115.	1.1	94
64	Outcomes beyond phenylalanine: An international perspectiveâ˜†. Molecular Genetics and Metabolism, 2010, 99, S79-S85.	1.1	43
65	Life-threatening methylenetetrahydrofolate reductase (MTHFR) deficiency with extremely early onset: Characterization of two novel mutations in compound heterozygous patients. Molecular Genetics and Metabolism, 2010, 100, 143-148.	1.1	25
66	Challenges and Pitfalls in the Management of Phenylketonuria. Pediatrics, 2010, 126, 333-341.	2.1	72
67	Optimizing the use of sapropterin (BH4) in the management of phenylketonuria. Molecular Genetics and Metabolism, 2009, 96, 158-163.	1.1	121
68	Molecular genetics of tetrahydrobiopterin-responsive phenylalanine hydroxylase deficiency. Human Mutation, 2008, 29, 167-175.	2.5	158
69	Evaluation of neonatal BH4 loading test in neonates screened for hyperphenylalaninemia. Early Human Development, 2008, 84, 561-567.	1.8	13
70	Pharmacokinetics of Sapropterin in Patients with Phenylketonuria. Clinical Pharmacokinetics, 2008, 47, 817-825.	3.5	37
71	Efficacy of sapropterin dihydrochloride (tetrahydrobiopterin, 6R-BH4) for reduction of phenylalanine concentration in patients with phenylketonuria: a phase III randomised placebo-controlled study. Lancet, The, 2007, 370, 504-510.	13.7	277
72	Management of Phenylketonuria and Hyperphenylalaninemia. Journal of Nutrition, 2007, 137, 1561S-1563S.	2.9	43

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73	Prise en charge nutritionnelle des troubles du comportement alimentaire chez l'adolescent. Nutrition Clinique Et Metabolisme, 2005, 19, 247-253.	0.5	1
74	Maternal phenylketonuria: the French survey. European Journal of Pediatrics, 2004, 163, 540-546.	2.7	20
75	Neonatal screening and long-term follow-up of phenylketonuria: the French database. Early Human Development, 2001, 65, 149-158.	1.8	27
76	Plasma cholesterol and endogenous cholesterol synthesis during refeeding in anorexia nervosa. Clinica Chimica Acta, 2000, 294, 45-56.	1.1	37