# FranÃ§ois Feillet

List of Publications by Year
in descending order



| | | | |
|---|---|---|---|
| 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 | 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. | 1.8 | 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. | 1.7 | 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. | 1.8 | 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. | 0.5 | 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. | 1.8 | 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. | 3.3 | 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. | 1.8 | 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. | 1.7 | 47 |
| 9 | A noncoding RNA modulator potentiates phenylalanine metabolism in mice. Science, 2021, 373, 662-673. | 6.0 | 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. | 1.1 | 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. | 1.7 | 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. | 0.4 | 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. | 1.1 | 5 |
| 14 | The natural history of infantile mitochondrial DNA depletion syndrome due to RRM2B deficiency. Genetics in Medicine, 2020, 22, 199-209. | 1.1 | 14 |
| 15 | Neurological manifestations in adults with phenylketonuria: new cases and review of the literature. Journal of Neurology, 2020, 267, 531-542. | 1.8 | 21 |
| 16 | Bone mineral density is within normal range in most adult phenylketonuria patients. Journal of Inherited Metabolic Disease, 2020, 43, 251-258. | 1.7 | 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. | 2.7 | 6 |
| 18 | Dietary practices in methylmalonic acidaemia: a European survey. Journal of Pediatric Endocrinology and Metabolism, 2020, 33, 147-155. | 0.4 | 8 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250. | 2.6 | 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. | 1.5 | 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. | 0.9 | 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. | 1.4 | 7 |
| 23 | Deciphering exome sequencing data: Bringing mitochondrial DNA variants to light. Human Mutation, 2019, 40, 2430-2443. | 1.1 | 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. | 1.5 | 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. | 1.1 | 40 |
| 26 | Efficacy of low dose nitisinone in the management of alkaptonuria. Molecular Genetics and Metabolism, 2019, 127, 184-190. | 0.5 | 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. | 0.7 | 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. | 1.8 | 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. | 1.2 | 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. | 0.5 | 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. | 2.2 | 13 |
| 32 | A PRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. Nature Communications, 2018, 9, 67. | 5.8 | 64 |
| 33 | Can untreated PKU patients escape from intellectual disability? A systematic review. Orphanet Journal of Rare Diseases, 2018, 13, 149. | 1.2 | 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. | 1.2 | 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. | 0.4 | 13 |
| 36 | Key European guidelines for the diagnosis and management of patients with phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 743-756. | 5.5 | 272 |

| # | Article | IF | Citations |
|---|---|---|---|
| 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. | 3.1 | 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. | 1.5 | 88 |
| 39 | Issues with European guidelines for phenylketonuria â€“ Authors' reply. Lancet Diabetes and Endocrinology,the, 2017, 5, 683-684. | 5.5 | 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. | 1.5 | 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. | 1.3 | 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. | 1.2 | 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. | 0.5 | 7 |
| 44 | Management of adult patients with phenylketonuria: survey results from 24 countries. European Journal of Pediatrics, 2015, 174, 119-127. | 1.3 | 32 |
| 45 | Mapping the functional landscape of frequentphenylalanine hydroxylase(PAH) genotypes promotes personalised medicine in phenylketonuria. Journal of Medical Genetics, 2015, 52, 175-185. | 1.5 | 37 |
| 46 | Fructose 1,6â€bisphosphatase deficiency: clinical, biochemical and genetic features in French patients. Journal of Inherited Metabolic Disease, 2015, 38, 881-887. | 1.7 | 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. | 0.7 | 11 |
| 48 | A Phase 3 Trial of Sebelipase Alfa in Lysosomal Acid Lipase Deficiency. New England Journal of Medicine, 2015, 373, 1010-1020. | 13.9 | 212 |
| 49 | Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. Brain, 2014, 137, 1350-1360. | 3.7 | 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. | 2.6 | 112 |
| 51 | Use of sapropterin dihydrochloride in maternal phenylketonuria. A European experience of eight cases. Journal of Inherited Metabolic Disease, 2014, 37, 753-762. | 1.7 | 28 |
| 52 | Gastric intrinsic factor deficiency with combined GIF heterozygous mutations and FUT2 secretor variant. Biochimie, 2013, 95, 995-1001. | 1.3 | 23 |
| 53 | Long-term Follow-up and Outcome of Phenylketonuria Patients on Sapropterin: A Retrospective Study. Pediatrics, 2013, 131, e1881-e1888. | 1.0 | 68 |
| 54 | Fluctuations in phenylalanine concentrations in phenylketonuria: A review of possible relationships with outcomes. Molecular Genetics and Metabolism, 2013, 110, 418-423. | 0.5 | 69 |

| # | Article | IF | Citations |
|---|---|---|---|
| 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. | 1.4 | 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. | 1.7 | 38 |
| 57 | Maternal phenylketonuria: low phenylalaninemia might increase the risk of intra uterine growth retardation. Journal of Inherited Metabolic Disease, 2012, 35, 993-999. | 1.7 | 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. | 1.7 | 24 |
| 59 | Nutritional issues in treating phenylketonuria. Journal of Inherited Metabolic Disease, 2010, 33, 659-664. | 1.7 | 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. | 3.7 | 219 |
| 62 | Rapid identification of HEXA mutations in Tay-Sachs patients. Biochemical and Biophysical Research Communications, 2010, 392, 599-602. | 1.0 | 6 |
| 63 | Management of phenylketonuria in Europe: Survey results from 19 countries. Molecular Genetics and Metabolism, 2010, 99, 109-115. | 0.5 | 94 |
| 64 | Outcomes beyond phenylalanine: An international perspectiveâ˜†. Molecular Genetics and Metabolism, 2010, 99, S79-S85. | 0.5 | 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. | 0.5 | 25 |
| 66 | Challenges and Pitfalls in the Management of Phenylketonuria. Pediatrics, 2010, 126, 333-341. | 1.0 | 72 |
| 67 | Optimizing the use of sapropterin (BH4) in the management of phenylketonuria. Molecular Genetics and Metabolism, 2009, 96, 158-163. | 0.5 | 121 |
| 68 | Molecular genetics of tetrahydrobiopterin-responsive phenylalanine hydroxylase deficiency. Human Mutation, 2008, 29, 167-175. | 1.1 | 158 |
| 69 | Evaluation of neonatal BH4 loading test in neonates screened for hyperphenylalaninemia. Early Human Development, 2008, 84, 561-567. | 0.8 | 13 |
| 70 | Pharmacokinetics of Sapropterin in Patients with Phenylketonuria. Clinical Pharmacokinetics, 2008, 47, 817-825. | 1.6 | 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. | 6.3 | 277 |
| 72 | Management of Phenylketonuria and Hyperphenylalaninemia. Journal of Nutrition, 2007, 137, 1561S-1563S. | 1.3 | 43 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Prise en charge nutritionnelle des troubles du comportement alimentaire chez l'adolescent. Nutrition Clinique Et Metabolisme, 2005, 19, 247-253. | 0.2 | 1 |
| 74 | Maternal phenylketonuria: the French survey. European Journal of Pediatrics, 2004, 163, 540-546. | 1.3 | 20 |
| 75 | Neonatal screening and long-term follow-up of phenylketonuria: the French database. Early Human Development, 2001, 65, 149-158. | 0.8 | 27 |
| 76 | Plasma cholesterol and endogenous cholesterol synthesis during refeeding in anorexia nervosa. Clinica Chimica Acta, 2000, 294, 45-56. | 0.5 | 37 |