# Francjan J Van Spronsen

List of Publications by Year
in descending order



| | | | |
|---|---|---|---|
| 102 papers | 4,391 citations | 136950 32 h-index | 118850 62 g-index |
| 105 all docs | 105 docs citations | 105 times ranked | 3569 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Metabolic control during the neonatal period in phenylketonuria: associations with childhood IQ. Pediatric Research, 2022, 91, 874-878. | 2.3 | 5 |
| 2 | Towards Next-Generation Sequencing (NGS)-Based Newborn Screening: A Technical Study to Prepare for the Challenges Ahead. International Journal of Neonatal Screening, 2022, 8, 17. | 3.2 | 15 |
| 3 | Dietary treatment in Dutch children with phenylketonuria: An inventory of associated social restrictions and eating problems. Nutrition, 2022, 97, 111576. | 2.4 | 8 |
| 4 | Caring for Ukrainian refugee children with acute and chronic diseases. Lancet, The, 2022, 399, 1689. | 13.7 | 4 |
| 5 | Age dependency of plasma vitamin B12 status markers in Dutch children and adolescents. Pediatric Research, 2021, 90, 1058-1064. | 2.3 | 7 |
| 6 | A generic emergency protocol for patients with inborn errors of metabolism causing fasting intolerance: A retrospective, singleâ€center study and the generation of www.emergencyprotocol.net. Journal of Inherited Metabolic Disease, 2021, 44, 1124-1135. | 3.6 | 10 |
| 7 | Phenylketonuria. Nature Reviews Disease Primers, 2021, 7, 36. | 30.5 | 174 |
| 8 | Undiagnosed Phenylketonuria Can Exist Everywhere: Results From an International Survey. Journal of Pediatrics, 2021, 239, 231-234.e2. | 1.8 | 9 |
| 9 | Preventive use of nitisinone in alkaptonuria. Orphanet Journal of Rare Diseases, 2021, 16, 343. | 2.7 | 12 |
| 10 | Correlations of blood and brain biochemistry in phenylketonuria: Results from the Pah-enu2 PKU mouse. Molecular Genetics and Metabolism, 2021, 134, 250-256. | 1.1 | 8 |
| 11 | Gut-Microbiome Composition in Response to Phenylketonuria Depends on Dietary Phenylalanine in BTBR Pahenu2 Mice. Frontiers in Nutrition, 2021, 8, 735366. | 3.7 | 4 |
| 12 | Predictability and inconsistencies of cognitive outcome in patients with phenylketonuria and personalised therapy: the challenge for the future guidelines. Journal of Medical Genetics, 2020, 57, 145-150. | 3.2 | 24 |
| 13 | Tetrahydrobiopterin treatment in phenylketonuria: A repurposing approach. Journal of Inherited Metabolic Disease, 2020, 43, 189-199. | 3.6 | 8 |
| 14 | The first European guidelines on phenylketonuria: Usefulness and implications for BH 4 responsiveness testing. Journal of Inherited Metabolic Disease, 2020, 43, 244-250. | 3.6 | 11 |
| 15 | Bone mineral density is within normal range in most adult phenylketonuria patients. Journal of Inherited Metabolic Disease, 2020, 43, 251-258. | 3.6 | 8 |
| 16 | Efficacy and safety of D,L-3-hydroxybutyrate (D,L-3-HB) treatment in multiple acyl-CoA dehydrogenase deficiency. Genetics in Medicine, 2020, 22, 908-916. | 2.4 | 19 |
| 17 | Does the 48-hour BH4 loading test miss responsive PKU patients?. Molecular Genetics and Metabolism, 2020, 129, 186-192. | 1.1 | 6 |
| 18 | Retrospective evaluation of the Dutch preâ€newborn screening cohort for propionic acidemia and isolated methylmalonic acidemia: What to aim, expect, and evaluate from newborn screening?. Journal of Inherited Metabolic Disease, 2020, 43, 424-437. | 3.6 | 18 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Aspartame and Phe-Containing Degradation Products in Soft Drinks across Europe. Nutrients, 2020, 12, 1887. | 4.1 | 7 |
| 20 | The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250. | 6.2 | 138 |
| 21 | Evaluation of 11â€‰years of newborn screening for maple syrup urine disease in the Netherlands and a systematic review of the literature: Strategies for optimization. JIMD Reports, 2020, 54, 68-78. | 1.5 | 12 |
| 22 | A Microbial Community Ecology Perspective on the Gut-Microbiome-Brain Axis. Frontiers in Endocrinology, 2020, 11, 611. | 3.5 | 7 |
| 23 | Treating neutropenia and neutrophil dysfunction in glycogen storage disease type Ib with an SGLT2 inhibitor. Blood, 2020, 136, 1033-1043. | 1.4 | 90 |
| 24 | Dried blood spot versus venous blood sampling for phenylalanine and tyrosine. Orphanet Journal of Rare Diseases, 2020, 15, 82. | 2.7 | 20 |
| 25 | Hippocampal microglia modifications in C57Bl/6 Pah and BTBR Pah phenylketonuria (PKU) mice depend on the genetic background, irrespective of disturbed sleep patterns. Neurobiology of Learning and Memory, 2019, 160, 139-143. | 1.9 | 8 |
| 26 | Changes in pediatric plasma acylcarnitines upon fasting for refined interpretation of metabolic stress. Molecular Genetics and Metabolism, 2019, 127, 327-335. | 1.1 | 5 |
| 27 | Blood and Brain Biochemistry and Behaviour in NTBC and Dietary Treated Tyrosinemia Type 1 Mice. Nutrients, 2019, 11, 2486. | 4.1 | 6 |
| 28 | Long-Term Outcomes and Practical Considerations in the Pharmacological Management of Tyrosinemia Type 1. Paediatric Drugs, 2019, 21, 413-426. | 3.1 | 31 |
| 29 | Biomarkers of Micronutrients in Regular Follow-Up for Tyrosinemia Type 1 and Phenylketonuria Patients. Nutrients, 2019, 11, 2011. | 4.1 | 5 |
| 30 | The Benefit of Large Neutral Amino Acid Supplementation to a Liberalized Phenylalanine-Restricted Diet in Adult Phenylketonuria Patients: Evidence from Adult Pah-Enu2 Mice. Nutrients, 2019, 11, 2252. | 4.1 | 8 |
| 31 | The neurological and psychological phenotype of adult patients with earlyâ€treated phenylketonuria: A systematic review. Journal of Inherited Metabolic Disease, 2019, 42, 209-219. | 3.6 | 42 |
| 32 | Over Restriction of Dietary Protein Allowance: The Importance of Ongoing Reassessment of Natural Protein Tolerance in Phenylketonuria. Nutrients, 2019, 11, 995. | 4.1 | 13 |
| 33 | A nationwide retrospective observational study of population newborn screening for mediumâ€chain acylâ€CoA dehydrogenase (MCAD) deficiency in the Netherlands. Journal of Inherited Metabolic Disease, 2019, 42, 890-897. | 3.6 | 21 |
| 34 | 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 |
| 35 | Long-term dietary intervention with low Phe and/or a specific nutrient combination improve certain aspects of brain functioning in phenylketonuria (PKU). PLoS ONE, 2019, 14, e0213391. | 2.5 | 6 |
| 36 | The Effect of Glycomacropeptide versus Amino Acids on Phenylalanine and Tyrosine Variability over 24 Hours in Children with PKU: A Randomized Controlled Trial. Nutrients, 2019, 11, 520. | 4.1 | 18 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Untreated PKU Patients without Intellectual Disability: What Do They Teach Us?. Nutrients, 2019, 11, 2572. | 4.1 | 16 |
| 38 | The Effect of Various Doses of Phenylalanine Supplementation on Blood Phenylalanine and Tyrosine Concentrations in Tyrosinemia Type 1 Patients. Nutrients, 2019, 11, 2816. | 4.1 | 6 |
| 39 | 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 |
| 40 | Optimising amino acid absorption: essential to improve nitrogen balance and metabolic control in phenylketonuria. Nutrition Research Reviews, 2019, 32, 70-78. | 4.1 | 44 |
| 41 | A preliminary study of telemedicine for patients with hepatic glycogen storage disease and their healthcare providers: from bedside to home site monitoring. Journal of Inherited Metabolic Disease, 2018, 41, 929-936. | 3.6 | 8 |
| 42 | Response to the Letter to the Editor Regarding â€œMicronutrients, Essential Fatty Acids and Bone Health in Phenylketonuriaâ€. Annals of Nutrition and Metabolism, 2018, 72, 80-81. | 1.9 | 1 |
| 43 | Daily variation of NTBC and its relation to succinylacetone in tyrosinemia type 1 patients comparing a single dose to two doses a day. Journal of Inherited Metabolic Disease, 2018, 41, 181-186. | 3.6 | 17 |
| 44 | Large neutral amino acid supplementation as an alternative to the phenylalanine-restricted diet in adults with phenylketonuria: evidence from adult Pah-enu2 mice. Journal of Nutritional Biochemistry, 2018, 53, 20-27. | 4.2 | 19 |
| 45 | Autism spectrum disorder: an early and frequent feature in cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2018, 41, 641-646. | 3.6 | 19 |
| 46 | Molybdenum cofactor deficiency type A: Prenatal monitoring using MRI. European Journal of Paediatric Neurology, 2018, 22, 536-540. | 1.6 | 5 |
| 47 | Heterogeneous clinical spectrum of DNAJC12-deficient hyperphenylalaninemia: from attention deficit to severe dystonia and intellectual disability. Journal of Medical Genetics, 2018, 55, 249-253. | 3.2 | 29 |
| 48 | Can untreated PKU patients escape from intellectual disability? A systematic review. Orphanet Journal of Rare Diseases, 2018, 13, 149. | 2.7 | 36 |
| 49 | Pathogenic variants in glutamyl-tRNAGln amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. Nature Communications, 2018, 9, 4065. | 12.8 | 44 |
| 50 | The potential role of gut microbiota and its modulators in the management of propionic and methylmalonic acidemia. Expert Opinion on Orphan Drugs, 2018, 6, 683-692. | 0.8 | 4 |
| 51 | Anthropomorphic measurements and nutritional biomarkers after 5â€¯years of BH 4 treatment in phenylketonuria patients. Molecular Genetics and Metabolism, 2018, 124, 238-242. | 1.1 | 7 |
| 52 | Integrating glycomics and genomics uncovers SLC10A7 as essential factor for bone mineralization by regulating post-Golgi protein transport and glycosylation. Human Molecular Genetics, 2018, 27, 3029-3045. | 2.9 | 37 |
| 53 | Renal thrombotic microangiopathy in patients with cblC defect: review of an under-recognized entity. Pediatric Nephrology, 2017, 32, 733-741. | 1.7 | 76 |
| 54 | Key European guidelines for the diagnosis and management of patients with phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 743-756. | 11.4 | 272 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | What Is the Best Blood Sampling Time for Metabolic Control of Phenylalanine and Tyrosine Concentrations in Tyrosinemia Type 1 Patients?. JIMD Reports, 2017, 36, 49-57. | 1.5 | 11 |
| 56 | Issues with European guidelines for phenylketonuria â€“ Authors' reply. Lancet Diabetes and Endocrinology,the, 2017, 5, 683-684. | 11.4 | 8 |
| 57 | Neurological and Neuropsychological Problems in Tyrosinemia Type I Patients. Advances in Experimental Medicine and Biology, 2017, 959, 111-122. | 1.6 | 32 |
| 58 | Liver Cancer in Tyrosinemia Type 1. Advances in Experimental Medicine and Biology, 2017, 959, 101-109. | 1.6 | 32 |
| 59 | Dietary Considerations in Tyrosinemia Type I. Advances in Experimental Medicine and Biology, 2017, 959, 197-204. | 1.6 | 18 |
| 60 | Long-Term Follow-Up of Cognition and Mental Health in Adult Phenylketonuria: A PKU-COBESO Study. Behavior Genetics, 2017, 47, 486-497. | 2.1 | 31 |
| 61 | Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. American Journal of Human Genetics, 2017, 101, 965-976. | 6.2 | 41 |
| 62 | Sleep Disturbances in Phenylketonuria: An Explorative Study in Men and Mice. Frontiers in Neurology, 2017, 8, 167. | 2.4 | 12 |
| 63 | Cognitive profile and mental health in adult phenylketonuria: A PKU-COBESO study.. Neuropsychology, 2017, 31, 437-447. | 1.3 | 46 |
| 64 | Presumptive brain influx of large neutral amino acids and the effect of phenylalanine supplementation in patients with Tyrosinemia type 1. PLoS ONE, 2017, 12, e0185342. | 2.5 | 8 |
| 65 | A Specific Nutrient Combination Attenuates the Reduced Expression of PSD-95 in the Proximal Dendrites of Hippocampal Cell Body Layers in a Mouse Model of Phenylketonuria. Nutrients, 2016, 8, 185. | 4.1 | 10 |
| 66 | Neurocognitive outcome in tyrosinemia type 1 patients compared to healthy controls. Orphanet Journal of Rare Diseases, 2016, 11, 87. | 2.7 | 60 |
| 67 | Therapeutic brain modulation with targeted large neutral amino acid supplements in the Pah-enu2 phenylketonuria mouse model. American Journal of Clinical Nutrition, 2016, 104, 1292-1300. | 4.7 | 35 |
| 68 | Neonates at risk of medium-chain acyl-CoA dehydrogenase deficiency: a perinatal protocol for use before population neonatal screening test results become available. Genetics in Medicine, 2016, 18, 1322-1323. | 2.4 | 2 |
| 69 | Occurrence of subdural hematomas in Dutch glutaric aciduria type 1 patients. European Journal of Pediatrics, 2016, 175, 1001-1006. | 2.7 | 21 |
| 70 | The neonatal tetrahydrobiopterin loading test in phenylketonuria: what is the predictive value?. Orphanet Journal of Rare Diseases, 2016, 11, 10. | 2.7 | 8 |
| 71 | Neonatal screening for profound biotinidase deficiency in the Netherlands: consequences and considerations. European Journal of Human Genetics, 2016, 24, 1424-1429. | 2.8 | 17 |
| 72 | Socialâ€cognitive functioning and social skills in patients with early treated phenylketonuria: a PKUâ€COBESO study. Journal of Inherited Metabolic Disease, 2016, 39, 355-362. | 3.6 | 57 |

| # | Article | IF | Citations |
|---|---|---|---|
| 73 | Reliable Diagnosis of Carnitine Palmitoyltransferase Type IA Deficiency by Analysis of Plasma Acylcarnitine Profiles. JIMD Reports, 2016, 32, 33-39. | 1.5 | 10 |
| 74 | 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 |
| 75 | Acute exercise in treated phenylketonuria patients: Physical activity and biochemical response. Molecular Genetics and Metabolism Reports, 2015, 5, 55-59. | 1.1 | 11 |
| 76 | Is BRIEF a useful instrument in day to day care of patients with phenylketonuria?. Molecular Genetics and Metabolism, 2015, 114, 425-430. | 1.1 | 9 |
| 77 | BH4 treatment in BH4-responsive PKU patients: preliminary data on blood prolactin concentrations suggest increased cerebral dopamine concentrations. Molecular Genetics and Metabolism, 2015, 114, 29-33. | 1.1 | 17 |
| 78 | 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 |
| 79 | Hepatocellular Carcinoma in Tyrosinemia Type 1 Without Clear Increase of AFP. Pediatrics, 2015, 135, e749-e752. | 2.1 | 25 |
| 80 | Voluntary Exercise Prevents Oxidative Stress in the Brain of Phenylketonuria Mice. JIMD Reports, 2015, 27, 69-77. | 1.5 | 14 |
| 81 | Efficacy and safety of cyclic pyranopterin monophosphate substitution in severe molybdenum cofactor deficiency type A: a prospective cohort study. Lancet, The, 2015, 386, 1955-1963. | 13.7 | 122 |
| 82 | Normoglycemic Ketonemia as Biochemical Presentation in Ketotic Glycogen Storage Disease. JIMD Reports, 2015, 28, 41-47. | 1.5 | 15 |
| 83 | Phenylketonuria: Brain Phenylalanine Concentrations Relate Inversely to Cerebral Protein Synthesis. Journal of Cerebral Blood Flow and Metabolism, 2015, 35, 200-205. | 4.3 | 16 |
| 84 | Large Neutral Amino Acid Supplementation Exerts Its Effect through Three Synergistic Mechanisms: Proof of Principle in Phenylketonuria Mice. PLoS ONE, 2015, 10, e0143833. | 2.5 | 59 |
| 85 | Infants with Tyrosinemia Type 1: Should phenylalanine be supplemented?. JIMD Reports, 2014, 18, 117-124. | 1.5 | 33 |
| 86 | Single amino acid supplementation in aminoacidopathies: a systematic review. Orphanet Journal of Rare Diseases, 2014, 9, 7. | 2.7 | 23 |
| 87 | Cross-sectional study of 168 patients with hepatorenal tyrosinaemia and implications for clinical practice. Orphanet Journal of Rare Diseases, 2014, 9, 107. | 2.7 | 110 |
| 88 | Impaired Cognitive Functioning in Patients with Tyrosinemia Type I Receiving Nitisinone. Journal of Pediatrics, 2014, 164, 398-401. | 1.8 | 71 |
| 89 | Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring. Molecular Genetics and Metabolism, 2014, 111, 16-25. | 1.1 | 111 |
| 90 | Neurocognitive Evidence for Revision of Treatment Targets and Guidelines for Phenylketonuria. Journal of Pediatrics, 2014, 164, 895-899.e2. | 1.8 | 64 |

| # | Article | IF | Citations |
|---|---|---|---|
| 91 | Recombinant phenylalanine ammonia lyase in phenylketonuria. Lancet, The, 2014, 384, 6-8. | 13.7 | 10 |
| 92 | Tetrahydrobiopterin responsiveness in phenylketonuria: prediction with the 48-hour loading test and genotype. Orphanet Journal of Rare Diseases, 2013, 8, 103. | 2.7 | 33 |
| 93 | Long-term Follow-up and Outcome of Phenylketonuria Patients on Sapropterin: A Retrospective Study. Pediatrics, 2013, 131, e1881-e1888. | 2.1 | 68 |
| 94 | Mental health and social functioning in early treated Phenylketonuria: The PKU-COBESO study. Molecular Genetics and Metabolism, 2013, 110, S57-S61. | 1.1 | 32 |
| 95 | Phenylketonuria: reduced tyrosine brain influx relates to reduced cerebral protein synthesis. Orphanet Journal of Rare Diseases, 2013, 8, 133. | 2.7 | 35 |
| 96 | Fluctuations in phenylalanine concentrations in phenylketonuria: A review of possible relationships with outcomes. Molecular Genetics and Metabolism, 2013, 110, 418-423. | 1.1 | 69 |
| 97 | Dietary treatment in phenylketonuria does not lead to increased risk of obesity or metabolic syndrome. Molecular Genetics and Metabolism, 2012, 107, 659-663. | 1.1 | 69 |
| 98 | Large neutral amino acids in the treatment of PKU: from theory to practice. Journal of Inherited Metabolic Disease, 2010, 33, 671-676. | 3.6 | 69 |
| 99 | Management of phenylketonuria in Europe: Survey results from 19 countries. Molecular Genetics and Metabolism, 2010, 99, 109-115. | 1.1 | 94 |
| 100 | Phenylketonuria management from an European perspective: A commentary. Molecular Genetics and Metabolism, 2010, 100, 107-110. | 1.1 | 5 |
| 101 | Phenylketonuria. Lancet, The, 2010, 376, 1417-1427. | 13.7 | 854 |
| 102 | Hereditary tyrosinemia type I: A new clinical classification with difference in prognosis on dietary treatment. Hepatology, 1994, 20, 1187-1191. | 7.3 | 172 |