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	Phenylketonuria. Lancet, The, 2010, 376, 1417-1427.	13.7	854
2	Key European guidelines for the diagnosis and management of patients with phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 743-756.	11.4	272
3	Phenylketonuria. Nature Reviews Disease Primers, 2021, 7, 36.	30.5	174
4	Hereditary tyrosinemia type I: A new clinical classification with difference in prognosis on dietary treatment. Hepatology, 1994, 20, 1187-1191.	7.3	172
5	The Genetic Landscape and Epidemiology of Phenylketonuria. American Journal of Human Genetics, 2020, 107, 234-250.	6.2	138
6	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
7	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
8	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
9	Management of phenylketonuria in Europe: Survey results from 19 countries. Molecular Genetics and Metabolism, 2010, 99, 109-115.	1.1	94
10	Treating neutropenia and neutrophil dysfunction in glycogen storage disease type Ib with an SGLT2 inhibitor. Blood, 2020, 136, 1033-1043.	1.4	90
11	Renal thrombotic microangiopathy in patients with cblC defect: review of an under-recognized entity. Pediatric Nephrology, 2017, 32, 733-741.	1.7	76
12	Impaired Cognitive Functioning in Patients with Tyrosinemia Type I Receiving Nitisinone. Journal of Pediatrics, 2014, 164, 398-401.	1.8	71
13	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
14	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
15	Fluctuations in phenylalanine concentrations in phenylketonuria: A review of possible relationships with outcomes. Molecular Genetics and Metabolism, 2013, 110, 418-423.	1.1	69
16	Long-term Follow-up and Outcome of Phenylketonuria Patients on Sapropterin: A Retrospective Study. Pediatrics, 2013, 131, e1881-e1888.	2.1	68
17	Neurocognitive Evidence for Revision of Treatment Targets and Guidelines for Phenylketonuria. Journal of Pediatrics, 2014, 164, 895-899.e2.	1.8	64
18	Neurocognitive outcome in tyrosinemia type 1 patients compared to healthy controls. Orphanet Journal of Rare Diseases, 2016, 11, 87.	2.7	60

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19	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
20	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
21	Cognitive profile and mental health in adult phenylketonuria: A PKU-COBESO study.. Neuropsychology, 2017, 31, 437-447.	1.3	46
22	Pathogenic variants in glutamyl-tRNAGln amidotransferase subunits cause a lethal mitochondrial cardiomyopathy disorder. Nature Communications, 2018, 9, 4065.	12.8	44
23	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
24	Optimising amino acid absorption: essential to improve nitrogen balance and metabolic control in phenylketonuria. Nutrition Research Reviews, 2019, 32, 70-78.	4.1	44
25	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
26	Allelic Expression Imbalance Promoting a Mutant PEX6 Allele Causes Zellweger Spectrum Disorder. American Journal of Human Genetics, 2017, 101, 965-976.	6.2	41
27	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
28	Can untreated PKU patients escape from intellectual disability? A systematic review. Orphanet Journal of Rare Diseases, 2018, 13, 149.	2.7	36
29	Phenylketonuria: reduced tyrosine brain influx relates to reduced cerebral protein synthesis. Orphanet Journal of Rare Diseases, 2013, 8, 133.	2.7	35
30	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
31	Tetrahydrobiopterin responsiveness in phenylketonuria: prediction with the 48-hour loading test and genotype. Orphanet Journal of Rare Diseases, 2013, 8, 103.	2.7	33
32	Infants with Tyrosinemia Type 1: Should phenylalanine be supplemented?. JIMD Reports, 2014, 18, 117-124.	1.5	33
33	Mental health and social functioning in early treated Phenylketonuria: The PKU-COBESO study. Molecular Genetics and Metabolism, 2013, 110, S57-S61.	1.1	32
34	Neurological and Neuropsychological Problems in Tyrosinemia Type I Patients. Advances in Experimental Medicine and Biology, 2017, 959, 111-122.	1.6	32
35	Liver Cancer in Tyrosinemia Type 1. Advances in Experimental Medicine and Biology, 2017, 959, 101-109.	1.6	32
36	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

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37	Long-Term Outcomes and Practical Considerations in the Pharmacological Management of Tyrosinemia Type 1. Paediatric Drugs, 2019, 21, 413-426.	3.1	31
38	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
39	Hepatocellular Carcinoma in Tyrosinemia Type 1 Without Clear Increase of AFP. Pediatrics, 2015, 135, e749-e752.	2.1	25
40	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
41	Single amino acid supplementation in aminoacidopathies: a systematic review. Orphanet Journal of Rare Diseases, 2014, 9, 7.	2.7	23
42	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
43	Occurrence of subdural hematomas in Dutch glutaric aciduria type 1 patients. European Journal of Pediatrics, 2016, 175, 1001-1006.	2.7	21
44	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
45	Dried blood spot versus venous blood sampling for phenylalanine and tyrosine. Orphanet Journal of Rare Diseases, 2020, 15, 82.	2.7	20
46	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
47	Autism spectrum disorder: an early and frequent feature in cerebrotendinous xanthomatosis. Journal of Inherited Metabolic Disease, 2018, 41, 641-646.	3.6	19
48	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
49	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
50	Dietary Considerations in Tyrosinemia Type I. Advances in Experimental Medicine and Biology, 2017, 959, 197-204.	1.6	18
51	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
52	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
53	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
54	Neonatal screening for profound biotinidase deficiency in the Netherlands: consequences and considerations. European Journal of Human Genetics, 2016, 24, 1424-1429.	2.8	17

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55	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
56	Phenylketonuria: Brain Phenylalanine Concentrations Relate Inversely to Cerebral Protein Synthesis. Journal of Cerebral Blood Flow and Metabolism, 2015, 35, 200-205.	4.3	16
57	Untreated PKU Patients without Intellectual Disability: What Do They Teach Us?. Nutrients, 2019, 11, 2572.	4.1	16
58	Normoglycemic Ketonemia as Biochemical Presentation in Ketotic Glycogen Storage Disease. JIMD Reports, 2015, 28, 41-47.	1.5	15
59	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
60	Voluntary Exercise Prevents Oxidative Stress in the Brain of Phenylketonuria Mice. JIMD Reports, 2015, 27, 69-77.	1.5	14
61	Over Restriction of Dietary Protein Allowance: The Importance of Ongoing Reassessment of Natural Protein Tolerance in Phenylketonuria. Nutrients, 2019, 11, 995.	4.1	13
62	Sleep Disturbances in Phenylketonuria: An Explorative Study in Men and Mice. Frontiers in Neurology, 2017, 8, 167.	2.4	12
63	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
64	Preventive use of nitisinone in alkaptonuria. Orphanet Journal of Rare Diseases, 2021, 16, 343.	2.7	12
65	Acute exercise in treated phenylketonuria patients: Physical activity and biochemical response. Molecular Genetics and Metabolism Reports, 2015, 5, 55-59.	1.1	11
66	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
67	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
68	Recombinant phenylalanine ammonia lyase in phenylketonuria. Lancet, The, 2014, 384, 6-8.	13.7	10
69	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
70	Reliable Diagnosis of Carnitine Palmitoyltransferase Type IA Deficiency by Analysis of Plasma Acylcarnitine Profiles. JIMD Reports, 2016, 32, 33-39.	1.5	10
71	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
72	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

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73	Undiagnosed Phenylketonuria Can Exist Everywhere: Results From an International Survey. Journal of Pediatrics, 2021, 239, 231-234.e2.	1.8	9
74	The neonatal tetrahydrobiopterin loading test in phenylketonuria: what is the predictive value?. Orphanet Journal of Rare Diseases, 2016, 11, 10.	2.7	8
75	Issues with European guidelines for phenylketonuria â€“ Authors' reply. Lancet Diabetes and Endocrinology,the, 2017, 5, 683-684.	11.4	8
76	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
77	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
78	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
79	Tetrahydrobiopterin treatment in phenylketonuria: A repurposing approach. Journal of Inherited Metabolic Disease, 2020, 43, 189-199.	3.6	8
80	Bone mineral density is within normal range in most adult phenylketonuria patients. Journal of Inherited Metabolic Disease, 2020, 43, 251-258.	3.6	8
81	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
82	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
83	Dietary treatment in Dutch children with phenylketonuria: An inventory of associated social restrictions and eating problems. Nutrition, 2022, 97, 111576.	2.4	8
84	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
85	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
86	Aspartame and Phe-Containing Degradation Products in Soft Drinks across Europe. Nutrients, 2020, 12, 1887.	4.1	7
87	A Microbial Community Ecology Perspective on the Gut-Microbiome-Brain Axis. Frontiers in Endocrinology, 2020, 11, 611.	3.5	7
88	Age dependency of plasma vitamin B12 status markers in Dutch children and adolescents. Pediatric Research, 2021, 90, 1058-1064.	2.3	7
89	Blood and Brain Biochemistry and Behaviour in NTBC and Dietary Treated Tyrosinemia Type 1 Mice. Nutrients, 2019, 11, 2486.	4.1	6
90	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

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91	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
92	Does the 48-hour BH4 loading test miss responsive PKU patients?. Molecular Genetics and Metabolism, 2020, 129, 186-192.	1.1	6
93	Phenylketonuria management from an European perspective: A commentary. Molecular Genetics and Metabolism, 2010, 100, 107-110.	1.1	5
94	Molybdenum cofactor deficiency type A: Prenatal monitoring using MRI. European Journal of Paediatric Neurology, 2018, 22, 536-540.	1.6	5
95	Changes in pediatric plasma acylcarnitines upon fasting for refined interpretation of metabolic stress. Molecular Genetics and Metabolism, 2019, 127, 327-335.	1.1	5
96	Biomarkers of Micronutrients in Regular Follow-Up for Tyrosinemia Type 1 and Phenylketonuria Patients. Nutrients, 2019, 11, 2011.	4.1	5
97	Metabolic control during the neonatal period in phenylketonuria: associations with childhood IQ. Pediatric Research, 2022, 91, 874-878.	2.3	5
98	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
99	Gut-Microbiome Composition in Response to Phenylketonuria Depends on Dietary Phenylalanine in BTBR Pahenu2 Mice. Frontiers in Nutrition, 2021, 8, 735366.	3.7	4
100	Caring for Ukrainian refugee children with acute and chronic diseases. Lancet, The, 2022, 399, 1689.	13.7	4
101	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
102	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