Nanda M Verhoeven-Duif

List of Publications by Year
in descending order

53 papers	1,342 citations	304743 22 h-index	395702 33 g-index
54 all docs	54 docs citations	54 times ranked	2496 citing authors

#	Article	IF	Citations
1	Clinical relevance of testing for metabolic vitamin B12 deficiency in patients with polyneuropathy. Nutritional Neuroscience, 2022, 25, 2536-2546.	3.1	6
2	The potential and limitations of intrahepatic cholangiocyte organoids to study inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2022, 45, 353-365.	3.6	4
3	Metabolomics in diagnostics of inborn metabolic disorders. Current Opinion in Systems Biology, 2022, 29, 100409.	2.6	9
4	Untargeted metabolic analysis in dried blood spots reveals metabolic signature in 22q11.2 deletion syndrome. Translational Psychiatry, 2022, 12, 97.	4.8	3
5	FAIR Genomes metadata schema promoting Next Generation Sequencing data reuse in Dutch healthcare and research. Scientific Data, 2022, 9, 169.	5.3	8
6	Recommendations for IVDR compliant in-house software development in clinical practice: a how-to paper with three use cases. Clinical Chemistry and Laboratory Medicine, 2022, 60, 982-988.	2.3	8
7	Biallelic variants in HPDL, encoding 4-hydroxyphenylpyruvate dioxygenase-like protein, lead to an infantile neurodegenerative condition. Genetics in Medicine, 2021, 23, 524-533.	2.4	17
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	Dried blood spot metabolomics reveals a metabolic fingerprint with diagnostic potential for Diamond Blackfan Anaemia. British Journal of Haematology, 2021, 193, 1185-1193.	2.5	4
10	Inborn disorders of the malate aspartate shuttle. Journal of Inherited Metabolic Disease, 2021, 44, 792-808.	3.6	26
11	Metabolic Fingerprint in Hereditary Spherocytosis Correlates With Red Blood Cell Characteristics and Clinical Severity. HemaSphere, 2021, 5, e591.	2.7	2
12	The end of the laboratory developed test as we know it? Recommendations from a national multidisciplinary taskforce of laboratory specialists on the interpretation of the IVDR and its complications. Clinical Chemistry and Laboratory Medicine, 2021, 59, 491-497.	2.3	27
13	Untargeted metabolic profiling in dried blood spots identifies disease fingerprint for pyruvate kinase deficiency. Haematologica, 2021, 106, 2720-2725.	3.5	14
14	Inborn errors of enzymes in glutamate metabolism. Journal of Inherited Metabolic Disease, 2020, 43, 200-215.	3.6	13
15	Metabolic fingerprinting reveals extensive consequences of GLS hyperactivity. Biochimica Et Biophysica Acta - General Subjects, 2020, 1864, 129484.	2.4	3
16	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
17	Pyridox(am)ine 5â€²-phosphate oxidase (PNPO) deficiency in zebrafish results in fatal seizures and metabolic aberrations. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2020, 1866, 165607.	3.8	17
18	NaV1.1 and NaV1.6 selective compounds reduce the behavior phenotype and epileptiform activity in a novel zebrafish model for Dravet Syndrome. PLoS ONE, 2020, 15, e0219106.	2.5	28

#	Article	IF	Citations
19	Accurate discrimination of Hartnup disorder from other aminoacidurias using a diagnostic ratio. Molecular Genetics and Metabolism Reports, 2020, 22, 100551.	1.1	3
20	Untargeted Metabolomics for Metabolic Diagnostic Screening with Automated Data Interpretation Using a Knowledge-Based Algorithm. International Journal of Molecular Sciences, 2020, 21, 979.	4.1	16
21	Cross-Omics: Integrating Genomics with Metabolomics in Clinical Diagnostics. Metabolites, 2020, 10, 206.	2.9	20
22	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	6.2	46
23	Aspartylglycosamine is a biomarker for NGLY1-CDDG, a congenital disorder of deglycosylation. Molecular Genetics and Metabolism, 2019, 127, 368-372.	1.1	31
24	Pyridox (am) ine 5'-phosphate oxidase deficiency induces seizures in Drosophila melanogaster. Human Molecular Genetics, 2019, 28, 3126-3136.	2.9	13
25	Assessing the Pre-Analytical Stability of Small-Molecule Metabolites in Cerebrospinal Fluid Using Direct-Infusion Metabolomics. Metabolites, 2019, 9, 236.	2.9	9
26	MDH1 deficiency is a metabolic disorder of the malateâ€“aspartate shuttle associated with early onset severe encephalopathy. Human Genetics, 2019, 138, 1247-1257.	3.8	31
27	Pathophysiology of propionic and methylmalonic acidemias. Part 2: Treatment strategies. Journal of Inherited Metabolic Disease, 2019, 42, 745-761.	3.6	27
28	Pathophysiology of propionic and methylmalonic acidemias. Part 1: Complications. Journal of Inherited Metabolic Disease, 2019, 42, 730-744.	3.6	68
29	Discovery of pyridoxal reductase activity as part of human vitamin B6 metabolism. Biochimica Et Biophysica Acta - General Subjects, 2019, 1863, 1088-1097.	2.4	23
30	Identification of human D lactate dehydrogenase deficiency. Nature Communications, 2019, 10, 1477.	12.8	62
31	Direct-infusion based metabolomics unveils biochemical profiles of inborn errors of metabolism in cerebrospinal fluid. Molecular Genetics and Metabolism, 2019, 127, 51-57.	1.1	13
32	GLS hyperactivity causes glutamate excess, infantile cataract and profound developmental delay. Human Molecular Genetics, 2019, 28, 96-104.	2.9	23
33	Focal epilepsy with fearâ€related behavior as primary presentation in Boerboel dogs. Journal of Veterinary Internal Medicine, 2019, 33, 694-700.	1.6	7
34	Identification of a Loss-of-Function Mutation in the Context of Glutaminase Deficiency and Neonatal Epileptic Encephalopathy. JAMA Neurology, 2019, 76, 342.	9.0	33
35	The Alkaline Phosphatase (ALPL) Locus Is Associated with B6 Vitamer Levels in CSF and Plasma. Genes, 2019, 10, 8.	2.4	12
36	Direct Infusion Based Metabolomics Identifies Metabolic Disease in Patientsâ€™ Dried Blood Spots and Plasma. Metabolites, 2019, 9, 12.	2.9	48

#	Article	IF	Citations
37	Improved cognition, mild anxiety-like behavior and decreased motor performance in pyridoxal phosphatase-deficient mice. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 193-205.	3.8	14
38	Phase I study of combined indomethacin and platinum-based chemotherapy to reduce platinum-induced fatty acids. Cancer Chemotherapy and Pharmacology, 2018, 81, 911-921.	2.3	8
39	Farnesoid X Receptor Activation Promotes Hepatic Amino Acid Catabolism and Ammonium Clearance in Mice. Gastroenterology, 2017, 152, 1462-1476.e10.	1.3	51
40	Quantification of metabolites in dried blood spots by direct infusion high resolution mass spectrometry. Analytica Chimica Acta, 2017, 979, 45-50.	5.4	33
41	Vitamin B6 is essential for serine de novo biosynthesis. Journal of Inherited Metabolic Disease, 2017, 40, 883-891.	3.6	36
42	CSF d-serine concentrations are similar in Alzheimer's disease, other dementias, and elderly controls. Neurobiology of Aging, 2016, 42, 213-216.	3.1	40
43	A New Approach for Fast Metabolic Diagnostics in CMAMMA. JIMD Reports, 2016, 30, 15-22.	1.5	7
44	Effectiveness of whole-exome sequencing and costs of the traditional diagnostic trajectory in children with intellectual disability. Genetics in Medicine, 2016, 18, 949-956.	2.4	148
45	Vitamin B6 in Plasma and Cerebrospinal Fluid of Children. PLoS ONE, 2015, 10, e0120972.	2.5	23
46	<scp>FOXO</scp> s support the metabolic requirements of normal and tumor cells by promoting <scp>IDH</scp> 1 expression. EMBO Reports, 2015, 16, 456-466.	4.5	38
47	Increased Plasma Levels of Chemoresistance-Inducing Fatty Acid 16:4(n-3) After Consumption of Fish and Fish Oil. JAMA Oncology, 2015, 1, 350.	7.1	49
48	Suitability of methylmalonic acid and total homocysteine analysis in dried bloodspots. Analytica Chimica Acta, 2015, 853, 435-441.	5.4	13
49	Survival and Psychomotor Development With Early Betaine Treatment in Patients With Severe Methylenetetrahydrofolate Reductase Deficiency. JAMA Neurology, 2014, 71, 188.	9.0	60
50	Expanding the clinical phenotype of COG6 deficiency. Journal of Medical Genetics, 2014, 51, 425.1-425.	3.2	2
51	Vitamin B-6 vitamers in human plasma and cerebrospinal fluid. American Journal of Clinical Nutrition, 2014, 100, 587-592.	4.7	24
52	The Intestine Plays a Substantial Role in Human Vitamin B6 Metabolism: A Caco-2 Cell Model. PLoS ONE, 2013, 8, e54113.	2.5	33
53	The Proline/Citrulline Ratio as a Biomarker for OAT Deficiency in Early Infancy. JIMD Reports, 2012, 6, 95-99.	1.5	24