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	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
2	Pathophysiology of propionic and methylmalonic acidemias. Part 1: Complications. Journal of Inherited Metabolic Disease, 2019, 42, 730-744.	3.6	68
3	Identification of human D lactate dehydrogenase deficiency. Nature Communications, 2019, 10, 1477.	12.8	62
4	Survival and Psychomotor Development With Early Betaine Treatment in Patients With Severe Methylenetetrahydrofolate Reductase Deficiency. JAMA Neurology, 2014, 71, 188.	9.0	60
5	Farnesoid X Receptor Activation Promotes Hepatic Amino Acid Catabolism and Ammonium Clearance in Mice. Gastroenterology, 2017, 152, 1462-1476.e10.	1.3	51
6	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
7	Direct Infusion Based Metabolomics Identifies Metabolic Disease in Patientsâ€™ Dried Blood Spots and Plasma. Metabolites, 2019, 9, 12.	2.9	48
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	Bi-allelic GOT2 Mutations Cause a Treatable Malate-Aspartate Shuttle-Related Encephalopathy. American Journal of Human Genetics, 2019, 105, 534-548.	6.2	46
10	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
11	<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
12	Vitamin B6 is essential for serine de novo biosynthesis. Journal of Inherited Metabolic Disease, 2017, 40, 883-891.	3.6	36
13	Quantification of metabolites in dried blood spots by direct infusion high resolution mass spectrometry. Analytica Chimica Acta, 2017, 979, 45-50.	5.4	33
14	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
15	The Intestine Plays a Substantial Role in Human Vitamin B6 Metabolism: A Caco-2 Cell Model. PLoS ONE, 2013, 8, e54113.	2.5	33
16	Aspartylglycosamine is a biomarker for NGLY1-CDDG, a congenital disorder of deglycosylation. Molecular Genetics and Metabolism, 2019, 127, 368-372.	1.1	31
17	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
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	Pathophysiology of propionic and methylmalonic acidemias. Part 2: Treatment strategies. Journal of Inherited Metabolic Disease, 2019, 42, 745-761.	3.6	27
20	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
21	Inborn disorders of the malate aspartate shuttle. Journal of Inherited Metabolic Disease, 2021, 44, 792-808.	3.6	26
22	The Proline/Citrulline Ratio as a Biomarker for OAT Deficiency in Early Infancy. JIMD Reports, 2012, 6, 95-99.	1.5	24
23	Vitamin B-6 vitamers in human plasma and cerebrospinal fluid. American Journal of Clinical Nutrition, 2014, 100, 587-592.	4.7	24
24	Vitamin B6 in Plasma and Cerebrospinal Fluid of Children. PLoS ONE, 2015, 10, e0120972.	2.5	23
25	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
26	GLS hyperactivity causes glutamate excess, infantile cataract and profound developmental delay. Human Molecular Genetics, 2019, 28, 96-104.	2.9	23
27	Cross-Omics: Integrating Genomics with Metabolomics in Clinical Diagnostics. Metabolites, 2020, 10, 206.	2.9	20
28	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
29	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
30	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
31	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
32	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
33	Untargeted metabolic profiling in dried blood spots identifies disease fingerprint for pyruvate kinase deficiency. Haematologica, 2021, 106, 2720-2725.	3.5	14
34	Suitability of methylmalonic acid and total homocysteine analysis in dried bloodspots. Analytica Chimica Acta, 2015, 853, 435-441.	5.4	13
35	Pyridox (am) ine 5'-phosphate oxidase deficiency induces seizures in Drosophila melanogaster. Human Molecular Genetics, 2019, 28, 3126-3136.	2.9	13
36	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

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37	Inborn errors of enzymes in glutamate metabolism. Journal of Inherited Metabolic Disease, 2020, 43, 200-215.	3.6	13
38	The Alkaline Phosphatase (ALPL) Locus Is Associated with B6 Vitamer Levels in CSF and Plasma. Genes, 2019, 10, 8.	2.4	12
39	Assessing the Pre-Analytical Stability of Small-Molecule Metabolites in Cerebrospinal Fluid Using Direct-Infusion Metabolomics. Metabolites, 2019, 9, 236.	2.9	9
40	Metabolomics in diagnostics of inborn metabolic disorders. Current Opinion in Systems Biology, 2022, 29, 100409.	2.6	9
41	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
42	FAIR Genomes metadata schema promoting Next Generation Sequencing data reuse in Dutch healthcare and research. Scientific Data, 2022, 9, 169.	5.3	8
43	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
44	A New Approach for Fast Metabolic Diagnostics in CMAMMA. JIMD Reports, 2016, 30, 15-22.	1.5	7
45	Focal epilepsy with fearâ€related behavior as primary presentation in Boerboel dogs. Journal of Veterinary Internal Medicine, 2019, 33, 694-700.	1.6	7
46	Clinical relevance of testing for metabolic vitamin B12 deficiency in patients with polyneuropathy. Nutritional Neuroscience, 2022, 25, 2536-2546.	3.1	6
47	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
48	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
49	Metabolic fingerprinting reveals extensive consequences of GLS hyperactivity. Biochimica Et Biophysica Acta - General Subjects, 2020, 1864, 129484.	2.4	3
50	Accurate discrimination of Hartnup disorder from other aminoacidurias using a diagnostic ratio. Molecular Genetics and Metabolism Reports, 2020, 22, 100551.	1.1	3
51	Untargeted metabolic analysis in dried blood spots reveals metabolic signature in 22q11.2 deletion syndrome. Translational Psychiatry, 2022, 12, 97.	4.8	3
52	Expanding the clinical phenotype of COG6 deficiency. Journal of Medical Genetics, 2014, 51, 425.1-425.	3.2	2
53	Metabolic Fingerprint in Hereditary Spherocytosis Correlates With Red Blood Cell Characteristics and Clinical Severity. HemaSphere, 2021, 5, e591.	2.7	2