

# François Boemer

## List of Publications by Year in descending order

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Version: 2024-02-01

53  
papers

1,636  
citations

331259

21  
h-index

301761

39  
g-index

58  
all docs

58  
docs citations

58  
times ranked

2066  
citing authors

#	ARTICLE	IF	CITATIONS
1	Clinical validation of cutoff target ranges in newborn screening of metabolic disorders by tandem mass spectrometry: A worldwide collaborative project. <i>Genetics in Medicine</i> , 2011, 13, 230-254.	1.1	308
2	Neonatal Screening in Europe Revisited: An ISNS Perspective on the Current State and Developments Since 2010. <i>International Journal of Neonatal Screening</i> , 2021, 7, 15.	1.2	118
3	Enhanced interpretation of newborn screening results without analyte cutoff values. <i>Genetics in Medicine</i> , 2012, 14, 648-655.	1.1	117
4	Newborn Screening for Sickle Cell Disease Using Tandem Mass Spectrometry. <i>Clinical Chemistry</i> , 2008, 54, 2036-2041.	1.5	70
5	Newborn screening for SMA in Southern Belgium. <i>Neuromuscular Disorders</i> , 2019, 29, 343-349.	0.3	65
6	Identification of methylenecyclopropyl acetic acid in serum of European horses with atypical myopathy. <i>Equine Veterinary Journal</i> , 2014, 46, 146-149.	0.9	63
7	Hepatocyte Transplantation Using the Domino Concept in a Child with Tetrabiopterin Nonresponsive Phenylketonuria. <i>Cell Transplantation</i> , 2012, 21, 2765-2770.	1.2	59
8	Outcome of publicly funded nationwide first-tier noninvasive prenatal screening. <i>Genetics in Medicine</i> , 2021, 23, 1137-1142.	1.1	58
9	244th ENMC international workshop: Newborn screening in spinal muscular atrophy May 10-12, 2019, Hoofddorp, The Netherlands. <i>Neuromuscular Disorders</i> , 2020, 30, 93-103.	0.3	55
10	Folinic acid treatment for schizophrenia associated with folate receptor autoantibodies. <i>Molecular Genetics and Metabolism</i> , 2014, 113, 307-314.	0.5	47
11	Consensus guidelines for the diagnosis and management of pyridoxine-dependent epilepsy due to Î±-aminoadipic semialdehyde dehydrogenase deficiency. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 178-192.	1.7	47
12	Neonatal Thyroid-Stimulating Hormone Concentrations in Belgium: A Useful Indicator for Detecting Mild Iodine Deficiency?. <i>PLoS ONE</i> , 2012, 7, e47770.	1.1	44
13	Mutation of the iron-sulfur cluster assembly gene <i>IBA57</i> causes fatal infantile leukodystrophy. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 1147-1153.	1.7	43
14	Samaras and seedlings of <i>Acer pseudoplatanus</i> are potential sources of hypoglycin A intoxication in atypical myopathy without necessarily inducing clinical signs. <i>Equine Veterinary Journal</i> , 2016, 48, 414-417.	0.9	35
15	A next-generation newborn screening pilot study: NGS on dried blood spots detects causal mutations in patients with inherited metabolic diseases. <i>Scientific Reports</i> , 2017, 7, 17641.	1.6	35
16	Three years pilot of spinal muscular atrophy newborn screening turned into official program in Southern Belgium. <i>Scientific Reports</i> , 2021, 11, 19922.	1.6	32
17	Tryptophan catabolism increases in breast cancer patients compared to healthy controls without affecting the cancer outcome or response to chemotherapy. <i>Journal of Translational Medicine</i> , 2019, 17, 239.	1.8	31
18	3-years experience review of neonatal screening for hemoglobin disorders using tandem mass spectrometry. <i>Clinica Chimica Acta</i> , 2011, 412, 1476-1479.	0.5	29

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19	Diagnostic pitfall in antenatal manifestations of CPT II deficiency. <i>Clinical Genetics</i> , 2016, 89, 193-197.	1.0	29
20	Evaluation of Physiological Amino Acids Profiling by Tandem Mass Spectrometry. <i>JIMD Reports</i> , 2013, 13, 119-128.	0.7	26
21	Surprising causes of C5-carnitine false positive results in newborn screening. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 52-54.	0.5	24
22	Neonatal screening for sickle cell disease in Central Africa: a study of 1825 newborns with a new enzyme-linked immunosorbent assay test. <i>Journal of Medical Screening</i> , 2007, 14, 113-116.	1.1	23
23	Acylcarnitines profile best predicts survival in horses with atypical myopathy. <i>PLoS ONE</i> , 2017, 12, e0182761.	1.1	22
24	Financial cost and quality of life of patients with spinal muscular atrophy identified by symptoms or newborn screening. <i>Developmental Medicine and Child Neurology</i> , 2023, 65, 67-77.	1.1	20
25	Quantification of hypoglycin A in serum using a TRAQ <sup>®</sup> assay. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2015, 997, 75-80.	1.2	19
26	Atypical myopathy in Père David's deer ( <i>Elaphurus davidianus</i> ) associated with ingestion of hypoglycin A. <i>Journal of Animal Science</i> , 2018, 96, 3537-3547.	0.2	19
27	Innovative PCR without DNA extraction for African sickle cell disease diagnosis. <i>Hematology</i> , 2018, 23, 181-186.	0.7	17
28	Detection of hypoglycin A in the seeds of sycamore ( <i>Acer pseudoplatanus</i> ) and box elder ( <i>A. tj EQq0 0 0 rgBT /Overlock 10 Zealand Veterinary Journal</i> , 2016, 64, 182-187.	0.4	16
29	Newborn screening of duchenne muscular dystrophy specifically targeting deletions amenable to exon-skipping therapy. <i>Scientific Reports</i> , 2021, 11, 3011.	1.6	14
30	Analytical validation based on total error measurement and cut-off interpretation of a neonatal screening TSH-immunoassay. <i>Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences</i> , 2009, 877, 2412-2417.	1.2	13
31	Mitochondrial function is altered in horse atypical myopathy. <i>Mitochondrion</i> , 2016, 30, 35-41.	1.6	13
32	Screening for sickle cell disease on dried blood: a new approach evaluated on 27,000 Belgian newborns. <i>Journal of Medical Screening</i> , 2006, 13, 132-136.	1.1	12
33	Mitochondrial encephalomyopathy with cytochrome c oxidase deficiency caused by a novel mutation in the MTCO1 gene. <i>Mitochondrion</i> , 2014, 17, 101-105.	1.6	10
34	Performance and Diagnostic Value of Genome-Wide Noninvasive Prenatal Testing in Multiple Gestations. <i>Obstetrics and Gynecology</i> , 2021, 137, 1102-1108.	1.2	10
35	Newborn screening of neuromuscular diseases. <i>Neuromuscular Disorders</i> , 2021, 31, 1070-1080.	0.3	9
36	Management of sickle cell disease: current practices and challenges in a northeastern region of the Democratic Republic of the Congo. <i>Hematology</i> , 2021, 26, 199-205.	0.7	8

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37	Grazing Mares on Pasture with Sycamore Maples: A Potential Threat to Suckling Foals and Food Safety through Milk Contamination. <i>Animals</i> , 2021, 11, 87.	1.0	8
38	Reader response: Discrepancy in redetermination of <i>SMN2</i> copy numbers in children with SMA. <i>Neurology</i> , 2020, 95, 144-145.	1.5	7
39	Novel Loss of Function Variant in BCKDK Causes a Treatable Developmental and Epileptic Encephalopathy. <i>International Journal of Molecular Sciences</i> , 2022, 23, 2253.	1.8	7
40	Neonatal Screening for Sickle Cell Disease in Belgium for More than 20 Years: An Experience for Comprehensive Care Improvement. <i>International Journal of Neonatal Screening</i> , 2018, 4, 37.	1.2	5
41	Raising Awareness of False Positive Newborn Screening Results Arising from Pivalate-Containing Creams and Antibiotics in Europe When Screening for Isovaleric Acidaemia. <i>International Journal of Neonatal Screening</i> , 2018, 4, 8.	1.2	4
42	Acylcarnitine profile in Alaskan sled dogs during submaximal multiday exercise points out metabolic flexibility and liver role in energy metabolism. <i>PLoS ONE</i> , 2021, 16, e0256009.	1.1	4
43	Systematic Screening of Neonatal Sickle Cell Disease with HemoTypeSC™ Kit-Test: Case Study and Literature Review. <i>Open Journal of Blood Diseases</i> , 2020, 10, 12-21.	0.1	4
44	Altered Serum Acylcarnitines Profile after a Prolonged Stay in Intensive Care. <i>Nutrients</i> , 2022, 14, 1122.	1.7	4
45	Molecular Analysis in Two Siblings African Patients with Severe Form of Hunter Syndrome: Identification of a Novel (p.Y54X) Nonsense Mutation. <i>Journal of Tropical Pediatrics</i> , 2007, 53, 434-437.	0.7	3
46	Metabolomic Signatures Discriminate Horses with Clinical Signs of Atypical Myopathy from Healthy Co-grazing Horses. <i>Journal of Proteome Research</i> , 2021, 20, 4681-4692.	1.8	3
47	Overview of current progress and challenges in diagnosis, and management of pediatric sickle cell disease in Democratic Republic of the Congo. <i>Hematology</i> , 2022, 27, 132-140.	0.7	3
48	Atypical Myopathy in Denmark Confirmed With the aTRAQ Assay. <i>Journal of Equine Veterinary Science</i> , 2016, 47, 77-79.	0.4	2
49	Comorbidity of sickle cell trait and albinism: a cross-sectional survey in the Democratic Republic of the Congo. <i>Pan African Medical Journal</i> , 2020, 35, 127.	0.3	2
50	Correspondence on: "Discrepancy in Spinal Muscular Atrophy Incidence findings in newborn screening programs: the influence of carrier screening?" by Kay et al. <i>Genetics in Medicine</i> , 2020, 22, 1913-1914.	1.1	2
51	Blood groups, hemoglobin phenotypes and clinical disorders of consanguineous Yansi population. <i>World Journal of Hematology</i> , 2013, 2, 109.	0.1	1
52	C5-carnitine false positive results in newborn screening: What is the cause?. <i>Medicina Clínica (English)</i> Tj ETQq0 0 0 r gBT /Oyerlock 10		
53	Belgian rare diseases plan in clinical pathology: identification of key biochemical diagnostic tests and establishment of reference laboratories and financing conditions. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 89.	1.2	0