

# Thierry Levade

## List of Publications by Year in descending order

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Version: 2024-02-01

57  
papers

2,734  
citations

186265  
28  
h-index

189892  
50  
g-index

59  
all docs

59  
docs citations

59  
times ranked

3565  
citing authors

#	ARTICLE	IF	CITATIONS
1	A diagnosis of progressive myoclonic ataxia guided by blood biomarkers. <i>Parkinsonism and Related Disorders</i> , 2022, 94, 124-126.	2.2	0
2	Disorders of Sphingolipid Synthesis, Sphingolipidoses, Niemann-Pick Disease Type C and Neuronal Ceroid Lipofuscinoses. , 2022, , 735-764.		1
3	A case of <sc><i>ASAH1</i></sc>â€related pure SMA evolving into adultâ€onset Farber disease. <i>Clinical Genetics</i> , 2021, 100, 234-235.	2.0	3
4	New Insights into the Role of Sphingolipid Metabolism in Melanoma. <i>Cells</i> , 2020, 9, 1967.	4.1	15
5	Concise asymmetric synthesis of new enantiomeric <i>C</i>-alkyl pyrrolidines acting as pharmacological chaperones against Gaucher disease. <i>Organic and Biomolecular Chemistry</i> , 2020, 18, 7852-7861.	2.8	12
6	Spinal muscular atrophy and Farber disease due to ASAH1 variants: A case report. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2369-2371.	1.2	4
7	Are Glucosylceramide-Related Sphingolipids Involved in the Increased Risk for Cancer in Gaucher Disease Patients? Review and Hypotheses. <i>Cancers</i> , 2020, 12, 475.	3.7	13
8	First Report of a Patient with MPS Type VII, Due to Novel Mutations in GUSB, Who Underwent Enzyme Replacement and Then Hematopoietic Stem Cell Transplantation. <i>International Journal of Molecular Sciences</i> , 2019, 20, 5345.	4.1	10
9	Selective Targeting of the Interconversion between Glucosylceramide and Ceramide by Scaffold Tailoring of Iminosugar Inhibitors. <i>Molecules</i> , 2019, 24, 354.	3.8	5
10	Sphingomyelin Synthase 1 (SMS1) Downregulation Is Associated With Sphingolipid Reprogramming and a Worse Prognosis in Melanoma. <i>Frontiers in Pharmacology</i> , 2019, 10, 443.	3.5	22
11	Inherited monogenic defects of ceramide metabolism: Molecular bases and diagnoses. <i>Clinica Chimica Acta</i> , 2019, 495, 457-466.	1.1	2
12	Allogeneic hematopoietic cell transplantation in Farber disease. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 286-294.	3.6	13
13	Targeting the Sphingosine 1-Phosphate Axis Exerts Potent Antitumor Activity in BRAFi-Resistant Melanomas. <i>Molecular Cancer Therapeutics</i> , 2019, 18, 289-300.	4.1	25
14	Pregnancy outcome in Refsum disease: Affected fetuses and children born to an affected mother. <i>JIMD Reports</i> , 2019, 46, 11-15.	1.5	0
15	Chronic lung injury and impaired pulmonary function in a mouse model of acid ceramidase deficiency. <i>American Journal of Physiology - Lung Cellular and Molecular Physiology</i> , 2018, 314, L406-L420.	2.9	26
16	Acid ceramidase deficiency: Farber disease and SMA-PME. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 121.	2.7	91
17	PNPLA1 defects in patients with autosomal recessive congenital ichthyosis and KO mice sustain PNPLA1 irreplaceable function in epidermal omega-O-acylceramide synthesis and skin permeability barrier. <i>Human Molecular Genetics</i> , 2017, 26, 1787-1800.	2.9	47
18	Acid Ceramidase Deficiency in Mice Results in a Broad Range of Central Nervous System Abnormalities. <i>American Journal of Pathology</i> , 2017, 187, 864-883.	3.8	41

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19	Acid Ceramidase Deficiency is characterized by a unique plasma cytokine and ceramide profile that is altered by therapy. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2017, 1863, 386-394.	3.8	35
20	A Review of Gaucher Disease Pathophysiology, Clinical Presentation and Treatments. <i>International Journal of Molecular Sciences</i> , 2017, 18, 441.	4.1	497
21	Role of Sphingolipids in Death Receptor Signalling. <i>Resistance To Targeted Anti-cancer Therapeutics</i> , 2017, , 229-245.	0.1	0
22	Brief Report: Peripheral Osteolysis in Adults Linked to <i>ASAH1</i> (Acid Ceramidase) Mutations: A New Presentation of Farber's Disease. <i>Arthritis and Rheumatology</i> , 2016, 68, 2323-2327.	5.6	17
23	Glucosylceramidases and malignancies in mammals. <i>Biochimie</i> , 2016, 125, 267-280.	2.6	36
24	Disorders of Sphingolipid Synthesis, Sphingolipidoses, Niemann-Pick Disease Type C and Neuronal Ceroid Lipofuscinoses. , 2016, , 551-575.		3
25	Downregulation of sphingosine kinase-1 induces protective tumor immunity by promoting M1 macrophage response in melanoma. <i>Oncotarget</i> , 2016, 7, 71873-71886.	1.8	35
26	Monogenic neurological disorders of sphingolipid metabolism. <i>Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids</i> , 2015, 1851, 1040-1051.	2.4	25
27	Human genetic disorders of sphingolipid biosynthesis. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 65-76.	3.6	29
28	Natural disease history and characterisation of SUMF1 molecular defects in ten unrelated patients with multiple sulfatase deficiency. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 31.	2.7	33
29	Acid ceramidase deficiency associated with spinal muscular atrophy with progressive myoclonic epilepsy. <i>Neuromuscular Disorders</i> , 2015, 25, 959-963.	0.6	32
30	Basics of Sphingolipid Metabolism and Signalling. , 2015, , 1-20.		4
31	Dysregulation of Sphingolipid Metabolism in Melanoma: Roles in Pigmentation, Cell Survival and Tumor Progression. , 2015, , 123-139.		2
32	Farber lipogranulomatosis with predominant joint involvement mimicking juvenile idiopathic arthritis. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 1079-1080.	3.6	23
33	Molecular basis of acid ceramidase deficiency in a neonatal form of Farber disease: Identification of the first large deletion in <i>ASAH1</i> gene. <i>Molecular Genetics and Metabolism</i> , 2013, 109, 276-281.	1.1	25
34	Systemic ceramide accumulation leads to severe and varied pathological consequences. <i>EMBO Molecular Medicine</i> , 2013, 5, 827-842.	6.9	90
35	The nonlysosomal $\beta$ -glucosidase GBA2 promotes endoplasmic reticulum stress and impairs tumorigenicity of human melanoma cells. <i>FASEB Journal</i> , 2013, 27, 489-498.	0.5	39
36	Spinal Muscular Atrophy Associated with Progressive Myoclonic Epilepsy Is Caused by Mutations in <i>ASAH1</i> . <i>American Journal of Human Genetics</i> , 2012, 91, 5-14.	6.2	250

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37	Acid Ceramidase Expression Modulates the Sensitivity of A375 Melanoma Cells to Dacarbazine. <i>Journal of Biological Chemistry</i> , 2011, 286, 28200-28209.	3.4	71
38	Danon disease: intrafamilial phenotypic variability related to a novel LAMP-2 mutation. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 515-522.	3.6	12
39	Autologous Transplantation of Lentivector/Acid Ceramidase <sup>+</sup> Transduced Hematopoietic Cells in Nonhuman Primates. <i>Human Gene Therapy</i> , 2011, 22, 679-687.	2.7	34
40	A simple fluorogenic method for determination of acid ceramidase activity and diagnosis of Farber disease. <i>Journal of Lipid Research</i> , 2010, 51, 3542-3547.	4.2	53
41	IL-6 Deficiency Attenuates Murine Diet-Induced Non-Alcoholic Steatohepatitis. <i>PLoS ONE</i> , 2009, 4, e7929.	2.5	75
42	Disruption of Sphingosine 1-Phosphate Lyase Confers Resistance to Chemotherapy and Promotes Oncogenesis through Bcl-2/Bcl-xL Upregulation. <i>Cancer Research</i> , 2009, 69, 9346-9353.	0.9	103
43	The natural marine anhydrophytosphingosine, Jaspine B, induces apoptosis in melanoma cells by interfering with ceramide metabolism. <i>Biochemical Pharmacology</i> , 2009, 78, 477-485.	4.4	99
44	Highly Regioselective Oxirane Ring <sup>+</sup> Opening of a Versatile Epoxyprololidine Precursor of New Imino <sup>+</sup> Sugar <sup>+</sup> Based Sphingolipid Mimics. <i>European Journal of Organic Chemistry</i> , 2009, 2009, 2474-2489.	2.4	15
45	Danon disease: Further clinical and molecular heterogeneity. <i>Muscle and Nerve</i> , 2009, 39, 837-844.	2.2	15
46	Recommendations on the diagnosis and management of Niemann-Pick disease type C. <i>Molecular Genetics and Metabolism</i> , 2009, 98, 152-165.	1.1	210
47	In vivo delivery of human acid ceramidase via cord blood transplantation and direct injection of lentivirus as novel treatment approaches for Farber disease. <i>Molecular Genetics and Metabolism</i> , 2008, 95, 133-141.	1.1	32
48	Functions of sphingolipid metabolism in mammals <sup>+</sup> Lessons from genetic defects. <i>Biochimica Et Biophysica Acta - Molecular and Cell Biology of Lipids</i> , 2008, 1781, 145-183.	2.4	62
49	Synthesis of a Novel Ceramide Analogue and its Use in a High-Throughput Fluorogenic Assay for Ceramidases. <i>ChemBioChem</i> , 2007, 8, 642-648.	2.6	53
50	C-Alkyl 5-membered ring imino sugars as new potent cytotoxic glucosylceramide synthase inhibitors. <i>Organic and Biomolecular Chemistry</i> , 2006, 4, 4437-4439.	2.8	22
51	Sphingolipids as modulators of cancer cell death: Potential therapeutic targets. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , 2006, 1758, 2104-2120.	2.6	116
52	The absence of functional glucosylceramide synthase does not sensitize melanoma cells for anticancer drugs. <i>FASEB Journal</i> , 2003, 17, 1144-1146.	0.5	45
53	Danon's disease (X-linked vacuolar cardiomyopathy and myopathy): a case with a novel Lamp-2 gene mutation. <i>Neuromuscular Disorders</i> , 2002, 12, 882-885.	0.6	41
54	Elevation of glucosylceramide in multidrug-resistant cancer cells and accumulation in cytoplasmic droplets. <i>International Journal of Cancer</i> , 2001, 94, 157-165.	5.1	110

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55	Retrovirus-Mediated Correction of the Metabolic Defect in Cultured Farber Disease Cells. Human Gene Therapy, 1999, 10, 1321-1329.	2.7	30
56	A simple method for screening for Farber disease on cultured skin fibroblasts. Clinica Chimica Acta, 1996, 245, 61-71.	1.1	25
57	Allogeneic hematopoietic cell transplantation in Farber disease. Journal of Inherited Metabolic Disease, 0, , .	3.6	4