

Carla Em Hollak

List of Publications by Year in descending order

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93
papers

5,049
citations

76326

40
h-index

91884

69
g-index

100
all docs

100
docs citations

100
times ranked

4333
citing authors

#	ARTICLE	IF	CITATIONS
1	Novel oral treatment of Gaucher's disease with N-butyldeoxynojirimycin (OGT 918) to decrease substrate biosynthesis. <i>Lancet, The</i> , 2000, 355, 1481-1485.	13.7	700
2	Recommendations for initiation and cessation of enzyme replacement therapy in patients with Fabry disease: the European Fabry Working Group consensus document. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 36.	2.7	239
3	Enzyme therapy for Fabry disease: Neutralizing antibodies toward agalsidase alpha and beta. <i>Kidney International</i> , 2004, 66, 1589-1595.	5.2	232
4	Gaucher disease type 1: Revised recommendations on evaluations and monitoring for adult patients. <i>Seminars in Hematology</i> , 2004, 41, 15-22.	3.4	215
5	Treatment of Fabry Disease: Outcome of a Comparative Trial with Agalsidase Alfa or Beta at a Dose of 0.2 mg/kg. <i>PLoS ONE</i> , 2007, 2, e598.	2.5	164
6	Elevated Levels of M-CSF, sCD14 and IL8 in Type 1 Gaucher Disease. <i>Blood Cells, Molecules, and Diseases</i> , 1997, 23, 201-212.	1.4	129
7	4 Plasma and metabolic abnormalities in Gaucher's disease. <i>Best Practice and Research: Clinical Haematology</i> , 1997, 10, 691-709.	1.1	128
8	Treatment of Fabry disease with different dosing regimens of agalsidase: Effects on antibody formation and GL-3. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 319-325.	1.1	118
9	The Morquio A Clinical Assessment Program: Baseline results illustrating progressive, multisystemic clinical impairments in Morquio A subjects. <i>Molecular Genetics and Metabolism</i> , 2013, 109, 54-61.	1.1	117
10	Long-term clinical outcomes in type 1 Gaucher disease following 10 years of imiglucerase treatment. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 543-553.	3.6	104
11	Plasma level of the macrophage-derived soluble CD163 is increased and positively correlates with severity in Gaucher's disease. <i>European Journal of Haematology</i> , 2004, 72, 135-139.	2.2	99
12	Increased plasma macrophage inflammatory protein (MIP)-1 α and MIP-1 β levels in type 1 Gaucher disease. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2007, 1772, 788-796.	3.8	96
13	HPLC for Simultaneous Quantification of Total Ceramide, Glucosylceramide, and Ceramide Trihexoside Concentrations in Plasma. <i>Clinical Chemistry</i> , 2007, 53, 742-747.	3.2	90
14	Force Majeure: Therapeutic measures in response to restricted supply of imiglucerase (Cerezyme) for patients with Gaucher disease. <i>Blood Cells, Molecules, and Diseases</i> , 2010, 44, 41-47.	1.4	88
15	Recommendations for the management of the haematological and onco-haematological aspects of Gaucher disease. <i>British Journal of Haematology</i> , 2007, 138, 676-686.	2.5	81
16	Plasma tumor necrosis factor- α (TNF- α) levels in Gaucher disease. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 1996, 1317, 219-222.	3.8	79
17	Small fiber neuropathy in Fabry disease. <i>Molecular Genetics and Metabolism</i> , 2012, 106, 135-141.	1.1	79
18	Long-term treatment effect in cerebrotendinous xanthomatosis depends on age at treatment start. <i>Neurology</i> , 2019, 92, e83-e95.	1.1	73

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19	Monitoring of Gaucher patients with a novel chitotriosidase assay. <i>Clinica Chimica Acta</i> , 2007, 381, 136-139.	1.1	72
20	Dixon Quantitative Chemical Shift Imaging Is a Sensitive Tool for the Evaluation of Bone Marrow Responses to Individualized Doses of Enzyme Supplementation Therapy in Type 1 Gaucher Disease. <i>Blood Cells, Molecules, and Diseases</i> , 2001, 27, 1005-1012.	1.4	71
21	Establishing 3-nitrotyrosine as a biomarker for the vasculopathy of Fabry disease. <i>Kidney International</i> , 2014, 86, 58-66.	5.2	71
22	Biochemistry of glycosphingolipid storage disorders: implications for therapeutic intervention. <i>Philosophical Transactions of the Royal Society B: Biological Sciences</i> , 2003, 358, 905-914.	4.0	67
23	Prominent increase in plasma ganglioside GM3 is associated with clinical manifestations of type I Gaucher disease. <i>Clinica Chimica Acta</i> , 2008, 389, 109-113.	1.1	65
24	Plasma chitotriosidase and CCL18 as surrogate markers for granulomatous macrophages in sarcoidosis. <i>Clinica Chimica Acta</i> , 2010, 411, 31-36.	1.1	65
25	Favourable effect of early versus late start of enzyme replacement therapy on plasma globotriaosylsphingosine levels in men with classical Fabry disease. <i>Molecular Genetics and Metabolism</i> , 2017, 121, 157-161.	1.1	64
26	Enzyme replacement and substrate reduction therapy for Gaucher disease. <i>The Cochrane Library</i> , 2015, 2015, CD010324.	2.8	62
27	Miglustat therapy in type 1 Gaucher disease: Clinical and safety outcomes in a multicenter retrospective cohort study. <i>Blood Cells, Molecules, and Diseases</i> , 2013, 51, 116-124.	1.4	60
28	Limitations of drug registries to evaluate orphan medicinal products for the treatment of lysosomal storage disorders. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 16.	2.7	58
29	Consequences of a global enzyme shortage of agalsidase beta in adult Dutch Fabry patients. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 69.	2.7	58
30	Cost-effectiveness of enzyme replacement therapy for Fabry disease. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 29.	2.7	58
31	The clinical spectrum and pathophysiology of skeletal complications in lysosomal storage disorders. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2015, 29, 219-235.	4.7	51
32	Biomarker responses correlate with antibody status in mucopolysaccharidosis type I patients on long-term enzyme replacement therapy. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 129-137.	1.1	49
33	Response of women with Fabry disease to enzyme replacement therapy: Comparison with men, using data from FOS—the Fabry Outcome Survey. <i>Molecular Genetics and Metabolism</i> , 2011, 103, 207-214.	1.1	48
34	The cytosolic β -glucosidase GBA3 does not influence type 1 Gaucher disease manifestation. <i>Blood Cells, Molecules, and Diseases</i> , 2011, 46, 19-26.	1.4	45
35	Vascular Aspects of Fabry Disease in Relation to Clinical Manifestations and Elevations in Plasma Globotriaosylsphingosine. <i>Hypertension</i> , 2012, 60, 998-1005.	2.7	45
36	Evaluation of miglustat as maintenance therapy after enzyme therapy in adults with stable type 1 Gaucher disease: a prospective, open-label non-inferiority study. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 102.	2.7	44

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37	Expert opinion on temporary treatment recommendations for Fabry disease during the shortage of enzyme replacement therapy (ERT). <i>Molecular Genetics and Metabolism</i> , 2011, 102, 99-102.	1.1	43
38	Cost-effectiveness of enzyme replacement therapy for type 1 Gaucher disease. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 51.	2.7	43
39	Uncertain Diagnosis of Fabry Disease in Patients with Neuropathic Pain, Angiokeratoma or Cornea Verticillata: Consensus on the Approach to Diagnosis and Follow-Up. <i>JIMD Reports</i> , 2014, 17, 83-90.	1.5	42
40	Long-Term Dose-Dependent Agalsidase Effects on Kidney Histology in Fabry Disease. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2017, 12, 1470-1479.	4.5	42
41	Fertility in adult women with classic galactosemia and primary ovarian insufficiency. <i>Fertility and Sterility</i> , 2017, 108, 168-174.	1.0	42
42	Clinical impact of the worldwide shortage of verteporfin (Visudyne®) on ophthalmic care. <i>Acta Ophthalmologica</i> , 2022, 100, .	1.1	42
43	Gallbladder and the risk of polyps and carcinoma in metachromatic leukodystrophy. <i>Neurology</i> , 2016, 87, 103-111.	1.1	40
44	Autonomic neuropathy in Fabry disease: a prospective study using the Autonomic Symptom Profile and cardiovascular autonomic function tests. <i>BMC Neurology</i> , 2010, 10, 38.	1.8	34
45	Modelling Gaucher disease progression: long-term enzyme replacement therapy reduces the incidence of splenectomy and bone complications. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 112.	2.7	34
46	Tetrahydrobiopterin responsiveness in phenylketonuria: prediction with the 48-hour loading test and genotype. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 103.	2.7	33
47	Mental health and social functioning in early treated Phenylketonuria: The PKU-COBESO study. <i>Molecular Genetics and Metabolism</i> , 2013, 110, S57-S61.	1.1	32
48	Gaucher disease: a model disorder for biomarker discovery. <i>Expert Review of Proteomics</i> , 2009, 6, 411-419.	3.0	31
49	A randomized, placebo-controlled clinical trial evaluating olipudase alfa enzyme replacement therapy for chronic acid sphingomyelinase deficiency (ASMD) in adults: One-year results. <i>Genetics in Medicine</i> , 2022, 24, 1425-1436.	2.4	30
50	Fabry patients' experiences with the timing of diagnosis relevant for the discussion on newborn screening. <i>Molecular Genetics and Metabolism</i> , 2013, 109, 201-207.	1.1	25
51	Assessment of plasma acylcarnitines before and after weight loss in obese subjects. <i>Archives of Biochemistry and Biophysics</i> , 2016, 606, 73-80.	3.0	25
52	An evidence-based review of the potential benefits of taliglucerase alfa in the treatment of patients with Gaucher disease. <i>Core Evidence</i> , 2012, 7, 15.	4.7	24
53	Imaging characteristics of focal splenic and hepatic lesions in type 1 Gaucher disease. <i>Blood Cells, Molecules, and Diseases</i> , 2016, 60, 49-57.	1.4	24
54	Detection of Mutated Angiotensin I-Converting Enzyme by Serum/Plasma Analysis Using a Pair of Monoclonal Antibodies. <i>Clinical Chemistry</i> , 2005, 51, 1040-1043.	3.2	23

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55	Micronutrients, Essential Fatty Acids and Bone Health in Phenylketonuria. <i>Annals of Nutrition and Metabolism</i> , 2017, 70, 111-121.	1.9	23
56	Development and clinical consequences of white matter lesions in Fabry disease: a systematic review. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 205-216.	1.1	23
57	Enzyme therapy for the treatment of type 1 Gaucher disease: clinical outcomes and dose " response relationships. <i>Expert Opinion on Pharmacotherapy</i> , 2009, 10, 2641-2652.	1.8	22
58	Hyperferritinemia and iron metabolism in Gaucher disease: Potential pathophysiological implications. <i>Blood Reviews</i> , 2016, 30, 431-437.	5.7	22
59	Iron storage in liver, bone marrow and splenic Gaucheroma reflects residual disease in type 1 Gaucher disease patients on treatment. <i>British Journal of Haematology</i> , 2017, 179, 635-647.	2.5	21
60	Registries for orphan drugs: generating evidence or marketing tools?. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 235.	2.7	18
61	Drug Repurposing for Rare Diseases: A Role for Academia. <i>Frontiers in Pharmacology</i> , 2021, 12, 746987.	3.5	18
62	Slowly Progressive Psychiatric Symptoms: Think Metachromatic Leukodystrophy. <i>Journal of the American Academy of Child and Adolescent Psychiatry</i> , 2018, 57, 74-76.	0.5	17
63	Impact of growing up with Fabry disease on achievement of psychosocial milestones and quality of life. <i>Molecular Genetics and Metabolism</i> , 2011, 104, 308-313.	1.1	16
64	Lung Transplantation in Gaucher Disease. <i>Chest</i> , 2016, 149, e1-e5.	0.8	16
65	The impact of metabolic control and tetrahydrobiopterin treatment on health related quality of life of patients with early-treated phenylketonuria: A PKU-COBESO study. <i>Molecular Genetics and Metabolism</i> , 2018, 125, 96-103.	1.1	16
66	The attenuated/late onset lysosomal storage disorders: Therapeutic goals and indications for enzyme replacement treatment in Gaucher and Fabry disease. <i>Best Practice and Research in Clinical Endocrinology and Metabolism</i> , 2015, 29, 205-218.	4.7	15
67	The impact of altered carnitine availability on acylcarnitine metabolism, energy expenditure and glucose tolerance in diet-induced obese mice. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2016, 1862, 1375-1382.	3.8	15
68	Biochemical and imaging parameters in acid sphingomyelinase deficiency: Potential utility as biomarkers. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 16-26.	1.1	15
69	Spontaneous regression of disease manifestations can occur in type 1 Gaucher disease; results of a retrospective cohort study. <i>Blood Cells, Molecules, and Diseases</i> , 2010, 44, 181-187.	1.4	14
70	Parkinsonism in Phenylketonuria: A Consequence of Dopamine Depletion?. <i>JIMD Reports</i> , 2014, 20, 35-38.	1.5	14
71	Novel therapeutic targets for the treatment of Fabry disease. <i>Expert Opinion on Therapeutic Targets</i> , 2007, 11, 821-833.	3.4	13
72	"MY PKU": increasing self-management in patients with phenylketonuria. A randomized controlled trial. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 48.	2.7	12

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73	Liposome-targeted recombinant human acid sphingomyelinase: Production, formulation, and in vitro evaluation. <i>European Journal of Pharmaceutics and Biopharmaceutics</i> , 2019, 137, 185-195.	4.3	12
74	Cost-Based Price Calculation of Mexiletine for Nondystrophic Myotonia. <i>Value in Health</i> , 2021, 24, 925-929.	0.3	11
75	A systematic review on effectiveness and safety of eliglustat for type 1 Gaucher disease. <i>Expert Opinion on Orphan Drugs</i> , 2014, 2, 523-529.	0.8	10
76	Adaptive pathway development for Fabry disease: a clinical approach. <i>Drug Discovery Today</i> , 2018, 23, 1251-1257.	6.4	10
77	Is BRIEF a useful instrument in day to day care of patients with phenylketonuria?. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 425-430.	1.1	9
78	Bone mineral density is within normal range in most adult phenylketonuria patients. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 251-258.	3.6	8
79	Influence of sex and phenotype on cardiac outcomes in patients with Fabry disease. <i>Heart</i> , 2021, 107, 1889-1897.	2.9	8
80	Gray and white matter are both affected in classical galactosemia: An explorative study on the association between neuroimaging and clinical outcome. <i>Molecular Genetics and Metabolism</i> , 2020, 131, 370-379.	1.1	7
81	Kidney and vascular function in adult patients with hereditary fructose intolerance. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 23, 100600.	1.1	7
82	Independent Registries Are Cost-Effective Tools to Provide Mandatory Postauthorization Surveillance for Orphan Medicinal Products. <i>Value in Health</i> , 2021, 24, 268-273.	0.3	7
83	Clinical pathways for inborn errors of metabolism: warranted and feasible. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 37.	2.7	6
84	Guidelines for the restart of imiglucerase in patients with Gaucher disease: Recommendations from the European Working Group on Gaucher disease. <i>Blood Cells, Molecules, and Diseases</i> , 2010, 44, 86-87.	1.4	5
85	Short-term withdrawal from imiglucerase: What can we learn from it?. <i>Blood Cells, Molecules, and Diseases</i> , 2011, 46, 105-106.	1.4	5
86	Expanding the clinical spectrum of cerebrotendinous xanthomatosis: Implications for newborn screening, follow-up and treatment. <i>Journal of Internal Medicine</i> , 2021, 290, 942-943.	6.0	4
87	The Galactose Index measured in fibroblasts of GALT deficient patients distinguishes variant patients detected by newborn screening from patients with classical phenotypes. <i>Molecular Genetics and Metabolism</i> , 2020, 129, 171-176.	1.1	3
88	Classifying the additional morbidities of Gaucher disease. <i>Blood Cells, Molecules, and Diseases</i> , 2018, 68, 209-210.	1.4	2
89	Enzyme replacement therapy for mucopolysaccharidosis type IV (Morquio syndrome). <i>The Cochrane Library</i> , 2018, , .	2.8	1
90	Editorial. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 899-900.	3.6	0

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91	Author Reply. Value in Health, 2021, 25, 158-159.	0.3	0
92	Determinants of Persisting Thrombocytopenia In Patients with Type 1 Gaucher Disease Treated with Alglucerase/Imiglucerase for 4â€“5 Years. Blood, 2010, 116, 4719-4719.	1.4	0
93	Miglustat Therapy in Type 1 Gaucher Disease: Long-Term Treatment Experience From a Multicenter, Retrospective Cohort Study,. Blood, 2011, 118, 3207-3207.	1.4	0