Carla Em Hollak

List of Publications by Year
in descending order

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	Clinical impact of the worldwide shortage of verteporfin (VisudyneÂ®) on ophthalmic care. Acta Ophthalmologica, 2022, 100, .	1.1	42
2	A randomized, placebo-controlled clinical trial evaluating olipudase alfa enzyme replacement therapy for chronic acid sphingomyelinase deficiency (ASMD) in adults: One-year results. Genetics in Medicine, 2022, 24, 1425-1436.	2.4	30
3	Independent Registries Are Cost-Effective Tools to Provide Mandatory Postauthorization Surveillance for Orphan Medicinal Products. Value in Health, 2021, 24, 268-273.	0.3	7
4	Influence of sex and phenotype on cardiac outcomes in patients with Fabry disease. Heart, 2021, 107, 1889-1897.	2.9	8
5	Expanding the clinical spectrum of cerebrotendinous xanthomatosis: Implications for newborn screening, followâ€up and treatment. Journal of Internal Medicine, 2021, 290, 942-943.	6.0	4
6	Cost-Based Price Calculation of Mexiletine for Nondystrophic Myotonia. Value in Health, 2021, 24, 925-929.	0.3	11
7	Author Reply. Value in Health, 2021, 25, 158-159.	0.3	0
8	Drug Repurposing for Rare Diseases: A Role for Academia. Frontiers in Pharmacology, 2021, 12, 746987.	3.5	18
9	Bone mineral density is within normal range in most adult phenylketonuria patients. Journal of Inherited Metabolic Disease, 2020, 43, 251-258.	3.6	8
10	Gray and white matter are both affected in classical galactosemia: An explorative study on the association between neuroimaging and clinical outcome. Molecular Genetics and Metabolism, 2020, 131, 370-379.	1.1	7
11	Registries for orphan drugs: generating evidence or marketing tools?. Orphanet Journal of Rare Diseases, 2020, 15, 235.	2.7	18
12	Kidney and vascular function in adult patients with hereditary fructose intolerance. Molecular Genetics and Metabolism Reports, 2020, 23, 100600.	1.1	7
13	Biochemical and imaging parameters in acid sphingomyelinase deficiency: Potential utility as biomarkers. Molecular Genetics and Metabolism, 2020, 130, 16-26.	1.1	15
14	The Galactose Index measured in fibroblasts of GALT deficient patients distinguishes variant patients detected by newborn screening from patients with classical phenotypes. Molecular Genetics and Metabolism, 2020, 129, 171-176.	1.1	3
15	Liposome-targeted recombinant human acid sphingomyelinase: Production, formulation, and in vitro evaluation. European Journal of Pharmaceutics and Biopharmaceutics, 2019, 137, 185-195.	4.3	12
16	Long-term treatment effect in cerebrotendinous xanthomatosis depends on age at treatment start. Neurology, 2019, 92, e83-e95.	1.1	73
17	Adaptive pathway development for Fabry disease: a clinical approach. Drug Discovery Today, 2018, 23, 1251-1257.	6.4	10
18	Slowly Progressive Psychiatric Symptoms: ThinkÂMetachromatic Leukodystrophy. Journal of the American Academy of Child and Adolescent Psychiatry, 2018, 57, 74-76.	0.5	17

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19	Enzyme replacement therapy for mucopolysaccharidosis type IV (Morquio syndrome). The Cochrane Library, 2018, , .	2.8	1
20	Classifying the additional morbidities of Gaucher disease. Blood Cells, Molecules, and Diseases, 2018, 68, 209-210.	1.4	2
21	Editorial. Journal of Inherited Metabolic Disease, 2018, 41, 899-900.	3.6	0
22	Development and clinical consequences of white matter lesions in Fabry disease: a systematic review. Molecular Genetics and Metabolism, 2018, 125, 205-216.	1.1	23
23	The impact of metabolic control and tetrahydrobiopterin treatment on health related quality of life of patients with early-treated phenylketonuria: A PKU-COBESO study. Molecular Genetics and Metabolism, 2018, 125, 96-103.	1.1	16
24	Favourable effect of early versus late start of enzyme replacement therapy on plasma globotriaosylsphingosine levels in men with classical Fabry disease. Molecular Genetics and Metabolism, 2017, 121, 157-161.	1.1	64
25	Long-Term Dose-Dependent Agalsidase Effects on Kidney Histology in Fabry Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2017, 12, 1470-1479.	4.5	42
26	Micronutrients, Essential Fatty Acids and Bone Health in Phenylketonuria. Annals of Nutrition and Metabolism, 2017, 70, 111-121.	1.9	23
27	Iron storage in liver, bone marrow and splenic Gaucheroma reflects residual disease in type 1 Gaucher disease patients on treatment. British Journal of Haematology, 2017, 179, 635-647.	2.5	21
28	Fertility in adult women with classic galactosemia and primary ovarian insufficiency. Fertility and Sterility, 2017, 108, 168-174.	1.0	42
29	The impact of altered carnitine availability on acylcarnitine metabolism, energy expenditure and glucose tolerance in diet-induced obese mice. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2016, 1862, 1375-1382.	3.8	15
30	Imaging characteristics of focal splenic and hepatic lesions in type 1 Gaucher disease. Blood Cells, Molecules, and Diseases, 2016, 60, 49-57.	1.4	24
31	Assessment of plasma acylcarnitines before and after weight loss in obese subjects. Archives of Biochemistry and Biophysics, 2016, 606, 73-80.	3.0	25
32	Hyperferritinemia and iron metabolism in Gaucher disease: Potential pathophysiological implications. Blood Reviews, 2016, 30, 431-437.	5.7	22
33	Gallbladder and the risk of polyps and carcinoma in metachromatic leukodystrophy. Neurology, 2016, 87, 103-111.	1.1	40
34	Lung Transplantation in Gaucher Disease. Chest, 2016, 149, e1-e5.	0.8	16
35	Enzyme replacement and substrate reduction therapy for Gaucher disease. The Cochrane Library, 2015, 2015, CD010324.	2.8	62
36	Is BRIEF a useful instrument in day to day care of patients with phenylketonuria?. Molecular Genetics and Metabolism, 2015, 114, 425-430.	1.1	9

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37	Biomarker responses correlate with antibody status in mucopolysaccharidosis type I patients on long-term enzyme replacement therapy. Molecular Genetics and Metabolism, 2015, 114, 129-137.	1.1	49
38	Recommendations for initiation and cessation of enzyme replacement therapy in patients with Fabry disease: the European Fabry Working Group consensus document. Orphanet Journal of Rare Diseases, 2015, 10, 36.	2.7	239
39	The attenuated/late onset lysosomal storage disorders: Therapeutic goals and indications for enzyme replacement treatment in Gaucher and Fabry disease. Best Practice and Research in Clinical Endocrinology and Metabolism, 2015, 29, 205-218.	4.7	15
40	The clinical spectrum and pathophysiology of skeletal complications in lysosomal storage disorders. Best Practice and Research in Clinical Endocrinology and Metabolism, 2015, 29, 219-235.	4.7	51
41	Parkinsonism in Phenylketonuria: A Consequence of Dopamine Depletion?. JIMD Reports, 2014, 20, 35-38.	1.5	14
42	Establishing 3-nitrotyrosine as a biomarker for the vasculopathy of Fabry disease. Kidney International, 2014, 86, 58-66.	5.2	71
43	Uncertain Diagnosis of Fabry Disease in Patients with Neuropathic Pain, Angiokeratoma or Cornea Verticillata: Consensus on the Approach to Diagnosis and Follow-Up. JIMD Reports, 2014, 17, 83-90.	1.5	42
44	A systematic review on effectiveness and safety of eliglustat for type 1 Gaucher disease. Expert Opinion on Orphan Drugs, 2014, 2, 523-529.	0.8	10
45	Modelling Gaucher disease progression: long-term enzyme replacement therapy reduces the incidence of splenectomy and bone complications. Orphanet Journal of Rare Diseases, 2014, 9, 112.	2.7	34
46	Cost-effectiveness of enzyme replacement therapy for type 1 Gaucher disease. Orphanet Journal of Rare Diseases, 2014, 9, 51.	2.7	43
47	Tetrahydrobiopterin responsiveness in phenylketonuria: prediction with the 48-hour loading test and genotype. Orphanet Journal of Rare Diseases, 2013, 8, 103.	2.7	33
48	Clinical pathways for inborn errors of metabolism: warranted and feasible. Orphanet Journal of Rare Diseases, 2013, 8, 37.	2.7	6
49	Cost-effectiveness of enzyme replacement therapy for Fabry disease. Orphanet Journal of Rare Diseases, 2013, 8, 29.	2.7	58
50	Fabry patients' experiences with the timing of diagnosis relevant for the discussion on newborn screening. Molecular Genetics and Metabolism, 2013, 109, 201-207.	1.1	25
51	Mental health and social functioning in early treated Phenylketonuria: The PKU-COBESO study. Molecular Genetics and Metabolism, 2013, 110, S57-S61.	1.1	32
52	Longâ€term clinical outcomes in type 1 Gaucher disease following 10 years of imiglucerase treatment. Journal of Inherited Metabolic Disease, 2013, 36, 543-553.	3.6	104
53	Miglustat therapy in type 1 Gaucher disease: Clinical and safety outcomes in a multicenter retrospective cohort study. Blood Cells, Molecules, and Diseases, 2013, 51, 116-124.	1.4	60
54	The Morquio A Clinical Assessment Program: Baseline results illustrating progressive, multisystemic clinical impairments in Morquio A subjects. Molecular Genetics and Metabolism, 2013, 109, 54-61.	1.1	117

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55	Vascular Aspects of Fabry Disease in Relation to Clinical Manifestations and Elevations in Plasma Globotriaosylsphingosine. Hypertension, 2012, 60, 998-1005.	2.7	45
56	Small fiber neuropathy in Fabry disease. Molecular Genetics and Metabolism, 2012, 106, 135-141.	1.1	79
57	Evaluation of miglustat as maintenance therapy after enzyme therapy in adults with stable type 1 Gaucher disease: a prospective, open-label non-inferiority study. Orphanet Journal of Rare Diseases, 2012, 7, 102.	2.7	44
58	An evidence-based review of the potential benefits of taliglucerase alfa in the treatment of patients with Gaucher disease. Core Evidence, 2012, 7, 15.	4.7	24
59	Short-term withdrawal from imiglucerase: What can we learn from it?. Blood Cells, Molecules, and Diseases, 2011, 46, 105-106.	1.4	5
60	The cytosolic Î²-glucosidase GBA3 does not influence type 1 Gaucher disease manifestation. Blood Cells, Molecules, and Diseases, 2011, 46, 19-26.	1.4	45
61	Expert opinion on temporary treatment recommendations for Fabry disease during the shortage of enzyme replacement therapy (ERT). Molecular Genetics and Metabolism, 2011, 102, 99-102.	1.1	43
62	Response of women with Fabry disease to enzyme replacement therapy: Comparison with men, using data from FOSâ€"the Fabry Outcome Survey. Molecular Genetics and Metabolism, 2011, 103, 207-214.	1.1	48
63	Impact of growing up with Fabry disease on achievement of psychosocial milestones and quality of life. Molecular Genetics and Metabolism, 2011, 104, 308-313.	1.1	16
64	Limitations of drug registries to evaluate orphan medicinal products for the treatment of lysosomal storage disorders. Orphanet Journal of Rare Diseases, 2011, 6, 16.	2.7	58
65	"MY PKU": increasing self-management in patients with phenylketonuria. A randomized controlled trial. Orphanet Journal of Rare Diseases, 2011, 6, 48.	2.7	12
66	Consequences of a global enzyme shortage of agalsidase beta in adult Dutch Fabry patients. Orphanet Journal of Rare Diseases, 2011, 6, 69.	2.7	58
67	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
68	Autonomic neuropathy in Fabry disease: a prospective study using the Autonomic Symptom Profile and cardiovascular autonomic function tests. BMC Neurology, 2010, 10, 38.	1.8	34
69	Force Majeure: Therapeutic measures in response to restricted supply of imiglucerase (Cerezyme) for patients with Gaucher disease. Blood Cells, Molecules, and Diseases, 2010, 44, 41-47.	1.4	88
70	Guidelines for the restart of imiglucerase in patients with Gaucher disease: Recommendations from the European Working Group on Gaucher disease. Blood Cells, Molecules, and Diseases, 2010, 44, 86-87.	1.4	5
71	Spontaneous regression of disease manifestations can occur in type 1 Gaucher disease; results of a retrospective cohort study. Blood Cells, Molecules, and Diseases, 2010, 44, 181-187.	1.4	14
72	Plasma chitotriosidase and CCL18 as surrogate markers for granulomatous macrophages in sarcoidosis. Clinica Chimica Acta, 2010, 411, 31-36.	1.1	65

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73	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
74	Enzyme therapy for the treatment of type 1 Gaucher disease: clinical outcomes and dose â€“ response relationships. Expert Opinion on Pharmacotherapy, 2009, 10, 2641-2652.	1.8	22
75	Gaucher disease: a model disorder for biomarker discovery. Expert Review of Proteomics, 2009, 6, 411-419.	3.0	31
76	Treatment of Fabry disease with different dosing regimens of agalsidase: Effects on antibody formation and GL-3. Molecular Genetics and Metabolism, 2008, 94, 319-325.	1.1	118
77	Prominent increase in plasma ganglioside GM3 is associated with clinical manifestations of type I Gaucher disease. Clinica Chimica Acta, 2008, 389, 109-113.	1.1	65
78	HPLC for Simultaneous Quantification of Total Ceramide, Glucosylceramide, and Ceramide Trihexoside Concentrations in Plasma. Clinical Chemistry, 2007, 53, 742-747.	3.2	90
79	Increased plasma macrophage inflammatory protein (MIP)-1Î± and MIP-1Î² levels in type 1 Gaucher disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2007, 1772, 788-796.	3.8	96
80	Monitoring of Gaucher patients with a novel chitotriosidase assay. Clinica Chimica Acta, 2007, 381, 136-139.	1.1	72
81	Novel therapeutic targets for the treatment of Fabry disease. Expert Opinion on Therapeutic Targets, 2007, 11, 821-833.	3.4	13
82	Treatment of Fabry Disease: Outcome of a Comparative Trial with Agalsidase Alfa or Beta at a Dose of 0.2 mg/kg. PLoS ONE, 2007, 2, e598.	2.5	164
83	Recommendations for the management of the haematological and oncoâ€haematological aspects of Gaucher disease¹. British Journal of Haematology, 2007, 138, 676-686.	2.5	81
84	Detection of Mutated Angiotensin I-Converting Enzyme by Serum/Plasma Analysis Using a Pair of Monoclonal Antibodies. Clinical Chemistry, 2005, 51, 1040-1043.	3.2	23
85	Enzyme therapy for Fabry disease: Neutralizing antibodies toward agalsidase alpha and beta. Kidney International, 2004, 66, 1589-1595.	5.2	232
86	Plasma level of the macrophage-derived soluble CD163 is increased and positively correlates with severity in Gaucher's disease. European Journal of Haematology, 2004, 72, 135-139.	2.2	99
87	Gaucher disease type 1: Revised recommendations on evaluations and monitoring for adult patients. Seminars in Hematology, 2004, 41, 15-22.	3.4	215
88	Biochemistry of glycosphingolipid storage disorders: implications for therapeutic intervention. Philosophical Transactions of the Royal Society B: Biological Sciences, 2003, 358, 905-914.	4.0	67
89	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. Blood Cells, Molecules, and Diseases, 2001, 27, 1005-1012.	1.4	71
90	Novel oral treatment of Gaucher's disease with N-butyldeoxynojirimycin (OGT 918) to decrease substrate biosynthesis. Lancet, The, 2000, 355, 1481-1485.	13.7	700

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91	Elevated Levels of M-CSF, sCD14 and IL8 in Type 1 Gaucher Disease. Blood Cells, Molecules, and Diseases, 1997, 23, 201-212.	1.4	129
92	4 Plasma and metabolic abnormalities in Gaucher's disease. Best Practice and Research: Clinical Haematology, 1997, 10, 691-709.	1.1	128
93	Plasma tumor necrosis factor-a (TNF-a) levels in Gaucher disease. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 1996, 1317, 219-222.	3.8	79