Pramod K Mistry

List of Publications by Year
in descending order

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112 papers	6,944 citations	57631 44 h-index	62479 80 g-index
118 all docs	118 docs citations	118 times ranked	4473 citing authors

#	Article	IF	Citations
1	The clinical spectrum of SARS-CoV-2 infection in Gaucher disease: Effect of both a pandemic and a rare disease that disrupts the immune system. Molecular Genetics and Metabolism, 2022, 135, 115-121.	0.5	3
2	Transjugular Intrahepatic Portosystemic Shunt for Refractory Ascites in Gaucher Disease. Cureus, 2022, 14, e23941.	0.2	0
3	Transformative effect of a Humanitarian Program for individuals affected by rare diseases: building support systems and creating local expertise. Orphanet Journal of Rare Diseases, 2022, 17, 87.	1.2	2
4	Accuracy of chitotriosidase activity and CCL18 concentration in assessing type I Gaucher disease severity. A systematic review with meta-analysis of individual participant data. Haematologica, 2021, 106, 437-445.	1.7	18
5	Gaucher disease and SARS-CoV-2 infection: Experience from 181 patients in New York. Molecular Genetics and Metabolism, 2021, 132, 44-48.	0.5	23
6	Gaucher disease: Basic and translational science needs for more complete therapy and management. Molecular Genetics and Metabolism, 2021, 132, 59-75.	0.5	28
7	Gaucher disease type 1 patients from the ICGG Gaucher Registry sustain initial clinical improvements during twenty years of imiglucerase treatment. Molecular Genetics and Metabolism, 2021, 132, 100-111.	0.5	28
8	Miglustat Therapy for SCARB2 -Associated Action Myoclonusâ€“Renal Failure Syndrome. Neurology: Genetics, 2021, 7, e614.	0.9	3
9	Clinical outcomes after 4.5â€‰years of eliglustat therapy for <scp>Gaucher</scp> disease type 1: Phase 3 <scp>ENGAGE</scp> trial final results. American Journal of Hematology, 2021, 96, 1156-1165.	2.0	22
10	Incremental biomarker and clinical outcomes after switch from enzyme therapy to eliglustat substrate reduction therapy in Gaucher disease. Molecular Genetics and Metabolism Reports, 2021, 29, 100798.	0.4	7
11	Long-read single molecule real-time (SMRT) sequencing of GBA1 locus in Gaucher disease national cohort from Argentina reveals high frequency of complex allele underlying severe skeletal phenotypes: Collaborative study from the Argentine Group for Diagnosis and Treatment of Gaucher Disease. Molecular Genetics and Metabolism Reports, 2021, 29, 100820.	0.4	3
12	Etiology of cirrhosis in the young. Human Pathology, 2020, 96, 96-103.	1.1	9
13	Organic Solute Transporter Alpha Deficiency: A Disorder With Cholestasis, Liver Fibrosis, and Congenital Diarrhea. Hepatology, 2020, 71, 1879-1882.	3.6	19
14	The road to biosimilars in rare diseases â€•ongoing lessons from Gaucher disease. American Journal of Hematology, 2020, 95, 233-237.	2.0	9
15	Gaucher disease and SARS-CoV-2 infection: Emerging management challenges. Molecular Genetics and Metabolism, 2020, 130, 164-169.	0.5	25
16	Glucosylsphingosine but not Saposin C, is the target antigen in Gaucher disease-associated gammopathy. Molecular Genetics and Metabolism, 2020, 129, 286-291.	0.5	28
17	Realâ€‰world effectiveness of eliglustat in treatmentâ€naÃ¯ve and switch patients enrolled in the International Collaborative Gaucher Group Gaucher Registry. American Journal of Hematology, 2020, 95, 1038-1046.	2.0	26
18	Largest Cohort Study of Gaucher Disease Type 3 from a Single Center in Egypt Spanning Two Decades. Blood, 2020, 136, 21-22.	0.6	0

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19	Lessons from lung transplantation: Cause for redefining the pathophysiology of pulmonary hypertension in gaucher disease. Respiratory Medicine Case Reports, 2019, 28, 100893.	0.2	4
20	Aberrant progranulin, YKL-40, cathepsin D and cathepsin S in Gaucher disease. Molecular Genetics and Metabolism, 2019, 128, 62-67.	0.5	13
21	Reply to: â€œWhole exome sequencing for personalized hepatology: Expanding applications in adults and challengesâ€. Journal of Hepatology, 2019, 71, 850-851.	1.8	2
22	Exome Sequencing in Clinical Hepatology. Hepatology, 2019, 70, 2185-2192.	3.6	19
23	Addendum to Letter to the Editor: Safety, efficacy, and authorization of eliglustat as a first-line therapy in Gaucher disease type 1. Blood Cells, Molecules, and Diseases, 2019, 77, 101-102.	0.6	1
24	Clinical utility of genomic analysis in adults with idiopathic liver disease. Journal of Hepatology, 2019, 70, 1214-1221.	1.8	47
25	Recommendations for clinical monitoring of patients with acid sphingomyelinase deficiency (ASMD). Molecular Genetics and Metabolism, 2019, 126, 98-105.	0.5	56
26	Gaucher disease in Montenegro - genotype/phenotype correlations: Five cases report. World Journal of Clinical Cases, 2019, 7, 1475-1482.	0.3	3
27	MGUS, lymphoplasmacytic malignancies, and Gaucher disease: the significance of the clinical association. Blood, 2018, 131, 2500-2501.	0.6	16
28	Hepatocellular carcinoma in Gaucher disease: an international case series. Journal of Inherited Metabolic Disease, 2018, 41, 819-827.	1.7	37
29	Safety, efficacy, and authorization of eliglustat as a first-line therapy in Gaucher disease type 1. Blood Cells, Molecules, and Diseases, 2018, 71, 71-74.	0.6	27
30	Validating glycoprotein non-metastatic melanoma B (gpNMB, osteoactivin), a new biomarker of Gaucher disease. Blood Cells, Molecules, and Diseases, 2018, 68, 47-53.	0.6	45
31	Therapeutic position of eliglustat. Blood Cells, Molecules, and Diseases, 2018, 69, 117-118.	0.6	3
32	Diagnosis and Management of Gaucher Disease in India â€“ Consensus Guidelines of the Gaucher Disease Task Force of the Society for Indian Academy of Medical Genetics and the Indian Academy of Pediatrics. Indian Pediatrics, 2018, 55, 143-153.	0.2	19
33	Antigen-mediated regulation in monoclonal gammopathies and myeloma. JCI Insight, 2018, 3, .	2.3	43
34	Diagnosis and Management of Gaucher Disease in India - Consensus Guidelines of the Gaucher Disease Task Force of the Society for Indian Academy of Medical Genetics and the Indian Academy of Pediatrics. Indian Pediatrics, 2018, 55, 143-153.	0.2	6
35	Long-term hematological, visceral, and growth outcomes in children with Gaucher disease type 3 treated with imiglucerase in the International Collaborative Gaucher Group Gaucher Registry. Molecular Genetics and Metabolism, 2017, 120, 47-56.	0.5	45
36	Roscoe Owen Brady, MD: Remembrances of co-investigators and colleagues. Molecular Genetics and Metabolism, 2017, 120, 1-7.	0.5	3

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37	Consensus recommendation for a diagnostic guideline for acid sphingomyelinase deficiency. Genetics in Medicine, 2017, 19, 967-974.	1.1	77
38	Transformation in pretreatment manifestations of Gaucher disease type 1 during two decades of alglucerase/imiglucerase enzyme replacement therapy in the International Collaborative Gaucher Group (ICGG) Gaucher Registry. American Journal of Hematology, 2017, 92, 929-939.	2.0	35
39	Clinical implications of Paracoccus yeeii bacteremia in a patient with decompensated cirrhosis. IDCases, 2017, 7, 9-10.	0.4	13
40	Glucosylsphingosine Promotes Î±-Synuclein Pathology in Mutant GBA-Associated Parkinson's Disease. Journal of Neuroscience, 2017, 37, 9617-9631.	1.7	180
41	Outcomes after 18 months of eliglustat therapy in treatmentâ€naÃ¯ve adults with <scp>G</scp>aucher disease type 1: The phase 3 ENGAGE trial. American Journal of Hematology, 2017, 92, 1170-1176.	2.0	77
42	Plasma chitotriosidase activity versus CCL18 level for assessing type I Gaucher disease severity: protocol for a systematic review with meta-analysis of individual participant data. Systematic Reviews, 2017, 6, 87.	2.5	19
43	Gaucher disease: Progress and ongoing challenges. Molecular Genetics and Metabolism, 2017, 120, 8-21.	0.5	112
44	Case series and literature review of skeletal tumors and their incidence in the Gaucher disease population. American Journal of Hematology, 2016, 91, 736-741.	2.0	4
45	Clinical course and prognosis in patients with Gaucher disease and parkinsonism. Neurology: Genetics, 2016, 2, e57.	0.9	29
46	Glucosylsphingosine is a key biomarker of Gaucher disease. American Journal of Hematology, 2016, 91, 1082-1089.	2.0	137
47	Inherited Metabolic Disorders: Efficacy of Enzyme Assays on Dried Blood Spots for the Diagnosis of Lysosomal Storage Disorders. JIMD Reports, 2016, 31, 15-27.	0.7	22
48	Clonal Immunoglobulin against Lysolipids in the Origin of Myeloma. New England Journal of Medicine, 2016, 374, 555-561.	13.9	167
49	Recommendations for the use of eliglustat in the treatment of adults with Gaucher disease type 1 in the United States. Molecular Genetics and Metabolism, 2016, 117, 95-103.	0.5	57
50	Understanding the natural history of <scp>G</scp>aucher disease. American Journal of Hematology, 2015, 90, S6-11.	2.0	187
51	Type II NKT-TFH cells against Gaucher lipids regulate B-cell immunity and inflammation. Blood, 2015, 125, 1256-1271.	0.6	119
52	The Role of ARF6 in Biliary Atresia. PLoS ONE, 2015, 10, e0138381.	1.1	66
53	ENGAGE â€" A phase 3, randomized, double-blind, placebo-controlled, multi-center study to investigate the efficacy and safety of eliglustat in adults with Gaucher disease type 1: Results after 18months. Molecular Genetics and Metabolism, 2015, 114, S81-S82.	0.5	8
54	Effect of Oral Eliglustat on Splenomegaly in Patients With Gaucher Disease Type 1. JAMA - Journal of the American Medical Association, 2015, 313, 695.	3.8	120

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55	Position statement: National Gaucher Foundation Medical Advisory Board, January 7, 2014. American Journal of Hematology, 2014, 89, 457-458.	2.0	6
56	Glucocerebrosidase 2 gene deletion rescues type 1 Gaucher disease. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 4934-4939.	3.3	85
57	Individual exome analysis in diagnosis and management of paediatric liver failure of indeterminate aetiology. Journal of Hepatology, 2014, 61, 1056-1063.	1.8	46
58	Gaucher disease: the metabolic defect, pathophysiology, phenotypes and natural history. Pediatric Endocrinology Reviews, 2014, 12 Suppl 1, 72-81.	1.2	35
59	Mutations in GBA2 Cause Autosomal-Recessive Cerebellar Ataxia with Spasticity. American Journal of Human Genetics, 2013, 92, 245-251.	2.6	120
60	Rare disease clinical research network's urea cycle consortium delivers a successful clinical trial to improve alternate pathway therapy. Hepatology, 2013, 57, 2100-2102.	3.6	5
61	Engage: A Phase 3, Randomized, Double-blind, Placebo-controlled, Multi-center Study To Investigate The Efficacy and Safety Of Eliglustat In Adults With Gaucher Disease Type 1: 9 Month Results. Blood, 2013, 122, 2275-2275.	0.6	1
62	Gaucher Disease and Malignancy: A Model for Cancer Pathogenesis in an Inborn Error of Metabolism. Critical Reviews in Oncogenesis, 2013, 18, 235-246.	0.2	96
63	Risk Factors Associated With Biliary Pancreatitis in Children. Journal of Pediatric Gastroenterology and Nutrition, 2012, 54, 651-656.	0.9	42
64	Phenotype diversity in type 1 Gaucher disease: discovering the genetic basis of Gaucher disease/hematologic malignancy phenotype by individual genome analysis. Blood, 2012, 119, 4731-4740.	0.6	39
65	Disease-drug pairs revealed by computational genomic connectivity mapping on GBA1 deficient, Gaucher disease mice. Biochemical and Biophysical Research Communications, 2012, 422, 573-577.	1.0	12
66	Gaucher disease geneGBAfunctions in immune regulation. Proceedings of the National Academy of Sciences of the United States of America, 2012, 109, 10018-10023.	3.3	70
67	Risk factors for fractures and avascular osteonecrosis in type 1 Gaucher disease: A study from the International Collaborative Gaucher Group (ICGG) Gaucher Registry. Journal of Bone and Mineral Research, 2012, 27, 1839-1848.	3.1	77
68	Genomeâ€wide association study of N370S homozygous Gaucher disease reveals the candidacy of CLN8 gene as a genetic modifier contributing to extreme phenotypic variation. American Journal of Hematology, 2012, 87, 377-383.	2.0	49
69	Disease state awareness in Gaucher disease: a Q&A expert roundtable discussion. Clinical Advances in Hematology and Oncology, 2012, 10, 1-16.	0.3	5
70	The mutation spectrum in Indian patients with Gaucher disease. Genome Biology, 2011, 12, .	13.9	6
71	Osteopenia in Gaucher disease develops early in life: Response to imiglucerase enzyme therapy in children, adolescents and adults. Blood Cells, Molecules, and Diseases, 2011, 46, 66-72.	0.6	95
72	Evaluation of high density lipoprotein as a circulating biomarker of Gaucher disease activity. Journal of Inherited Metabolic Disease, 2011, 34, 429-437.	1.7	24

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73	Pulmonary vascular disease in Gaucher disease: clinical spectrum, determinants of phenotype and longâ€term outcomes of therapy. Journal of Inherited Metabolic Disease, 2011, 34, 643-650.	1.7	63
74	Reducing selection bias in case-control studies from rare disease registries. Orphanet Journal of Rare Diseases, 2011, 6, 61.	1.2	24
75	A reappraisal of Gaucher diseaseâ€"diagnosis and disease management algorithms. American Journal of Hematology, 2011, 86, 110-115.	2.0	135
76	Elevated plasma glucosylsphingosine in Gaucher disease: relation to phenotype, storage cell markers, and therapeutic response. Blood, 2011, 118, e118-e127.	0.6	224
77	Liver transplantation for inherited metabolic disorders of the liver. Current Opinion in Organ Transplantation, 2010, 15, 269-276.	0.8	52
78	Abnormal nonstoring capillary endothelium: a novel feature of Gaucher disease. Ultrastructural study of dermal capillaries. Journal of Inherited Metabolic Disease, 2010, 33, 69-78.	1.7	9
79	The risk of Parkinson's disease in type 1 Gaucher disease. Journal of Inherited Metabolic Disease, 2010, 33, 167-173.	1.7	182
80	High incidence of cholesterol gallstone disease in type 1 Gaucher disease: characterizing the biliary phenotype of type 1 Gaucher disease. Journal of Inherited Metabolic Disease, 2010, 33, 291-300.	1.7	47
81	Focal splenic lesions in type I Gaucher disease are associated with poor platelet and splenic response to macrophageâ€targeted enzyme replacement therapy. Journal of Inherited Metabolic Disease, 2010, 33, 769-774.	1.7	37
82	Hyperferritinemia and iron overload in type 1 Gaucher disease. American Journal of Hematology, 2010, 85, 472-476.	2.0	69
83	Glucocerebrosidase gene-deficient mouse recapitulates Gaucher disease displaying cellular and molecular dysregulation beyond the macrophage. Proceedings of the National Academy of Sciences of the United States of America, 2010, 107, 19473-19478.	3.3	198
84	Misdiagnosis of Niemannâ€Pick disease type C as Gaucher disease. Journal of Inherited Metabolic Disease, 2010, 33, 429-433.	1.7	32
85	Expanding spectrum of the association between Type 1 Gaucher disease and cancers: A series of patients with up to 3 sequential cancers of multiple typesâ€"Correlation with genotype and phenotype. American Journal of Hematology, 2010, 85, 340-345.	2.0	45
86	Hematologic algorithms in Gaucher disease: Outcomes from an International Meeting. Clinical Therapeutics, 2009, 31, S173-S174.	1.1	0
87	The underrecognized progressive nature of N370S Gaucher disease and assessment of cancer risk in 403 patients. American Journal of Hematology, 2009, 84, 208-214.	2.0	146
88	Gaucher disease: Resetting the clinical and scientific agenda. American Journal of Hematology, 2009, 84, 205-207.	2.0	15
89	Timing of initiation of enzyme replacement therapy after diagnosis of type 1 Gaucher disease: effect on incidence of avascular necrosis. British Journal of Haematology, 2009, 147, 561-570.	1.2	97
90	Dose-response relationships for enzyme replacement therapy with imiglucerase/alglucerase in patients with Gaucher disease type 1. Genetics in Medicine, 2009, 11, 92-100.	1.1	94

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91	Avascular Necrosis in Untreated Patients with Type 1 Gaucher Disease.. Blood, 2009, 114, 1353-1353.	0.6	3
92	Life expectancy in Gaucher disease type 1. American Journal of Hematology, 2008, 83, 896-900.	2.0	72
93	Prevalence of Type 1 Gaucher Disease in the United States. Archives of Internal Medicine, 2008, 168, 326.	4.3	18
94	Correlation of MRI-Based Bone Marrow Burden Score with Genotype and Spleen Status in Gaucher's Disease. American Journal of Roentgenology, 2008, 191, 115-123.	1.0	47
95	Inherited metabolic disease of the liver. Current Opinion in Gastroenterology, 2008, 24, 278-286.	1.0	29
96	Diagnostic and Disease Management Algorithms for Gaucher Disease: A Guide for Haematologists. Blood, 2008, 112, 4648-4648.	0.6	0
97	Comprehensive Disease Management Model in Heterogeneous Progressive Disease, Exemplified by Gaucher Disease. Clinical Therapeutics, 2007, 29, S79-S80.	1.1	0
98	Consequences of diagnostic delays in type 1 Gaucher disease: The need for greater awareness among Hematologistsâ€“Oncologists and an opportunity for early diagnosis and intervention. American Journal of Hematology, 2007, 82, 697-701.	2.0	129
99	Consequences of Diagnostic Delays in Type 1 Gaucher Disease: A Unique Opportunity among Hematologists/Oncologists for Early Diagnosis and Intervention.. Blood, 2006, 108, 3308-3308.	0.6	0
100	Guidance on the use of miglustat for treating patients with type 1 Gaucher disease. American Journal of Hematology, 2005, 80, 223-229.	2.0	90
101	Individualization of long-term enzyme replacement therapy for Gaucher disease. Genetics in Medicine, 2005, 7, 105-110.	1.1	109
102	Hepatocellular Carcinoma in Type 1 Gaucher Disease: A Case Report with Review of the Literature. Seminars in Liver Disease, 2005, 25, 226-229.	1.8	31
103	Enzyme replacement therapy and monitoring for children with type 1 Gaucher disease: consensus recommendations. Journal of Pediatrics, 2004, 144, 112-120.	0.9	107
104	Therapeutic goals in the treatment of Gaucher disease. Seminars in Hematology, 2004, 41, 4-14.	1.8	418
105	Gaucher disease type 1: Revised recommendations on evaluations and monitoring for adult patients. Seminars in Hematology, 2004, 41, 15-22.	1.8	215
106	Effectiveness of enzyme replacement therapy in 1028 patients with type 1 Gaucher disease after 2 to 5 years of treatment: a report from the Gaucher Registry. American Journal of Medicine, 2002, 113, 112-119.	0.6	491
107	Pulmonary hypertension in type 1 Gaucherâ€™s disease: genetic and epigenetic determinants of phenotype and response to therapy. Molecular Genetics and Metabolism, 2002, 77, 91-98.	0.5	154
108	Molecular Diagnosis of Wilson Disease. Molecular Genetics and Metabolism, 2001, 72, 223-230.	0.5	41

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109	The Gaucher Registry. Archives of Internal Medicine, 2000, 160, 2835.	4.3	451
110	MYCOPHENOLATE MOFETIL MONOTHERAPY IN STABLE LIVER TRANSPLANT PATIENTS WITH CYCLOSPORINE-INDUCED RENAL IMPAIRMENT. Transplantation, 1999, 68, 155-157.	0.5	62
111	12 A practical approach to diagnosis and management of Gaucher's disease. Best Practice and Research: Clinical Haematology, 1997, 10, 817-838.	1.1	28
112	Therapeutic delivery of proteins to macrophages: implications for treatment of Gaucher's disease. Lancet, The, 1996, 348, 1555-1559.	6.3	92