Ida Vanessa D Schwartz

List of Publications by Year
in descending order

237 papers	6,176 citations	87723 38 h-index	95083 68 g-index
280 all docs	280 docs citations	280 times ranked	5525 citing authors

#	Article	IF	Citations
1	Mucopolysaccharidosis I: Management and Treatment Guidelines. Pediatrics, 2009, 123, 19-29.	1.0	400
2	Enzyme replacement therapy for mucopolysaccharidosis VI: A phase 3, randomized, double-blind, placebo-controlled, multinational study of recombinant human N-acetylgalactosamine 4-sulfatase (recombinant human arylsulfatase B or rhASB) and follow-on, open-label extension study. Journal of Pediatrics, 2006, 148, 533-539.e6.	0.9	335
3	Use of Misoprostol during Pregnancy and MÃ¶bius' Syndrome in Infants. New England Journal of Medicine, 1998, 338, 1881-1885.	13.9	245
4	Direct Comparison of Measures of Endurance, Mobility, and Joint Function During Enzyme-Replacement Therapy of Mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome): Results After 48 Weeks in a Phase 2 Open-Label Clinical Study of Recombinant Human N-Acetylgalactosamine 4-Sulfatase. Pediatrics, 2005, 115, e681-e689.	1.0	198
5	Long-term follow-up of endurance and safety outcomes during enzyme replacement therapy for mucopolysaccharidosis VI: Final results of three clinical studies of recombinant human N-acetylgalactosamine 4-sulfatase. Molecular Genetics and Metabolism, 2008, 94, 469-475.	0.5	198
6	Multidisciplinary Management of Hunter Syndrome. Pediatrics, 2009, 124, e1228-e1239.	1.0	159
7	Magnetic resonance imaging findings in Hunter syndrome. Acta Paediatrica, International Journal of Paediatrics, 2008, 97, 61-68.	0.7	152
8	Gender, Race and Parenthood Impact Academic Productivity During the COVID-19 Pandemic: From Survey to Action. Frontiers in Psychology, 2021, 12, 663252.	1.1	152
9	Mucopolysaccharidosis I, II, and VI: brief review and guidelines for treatment. Genetics and Molecular Biology, 2010, 33, 589-604.	0.6	150
10	Impact of COVID-19 on academic mothers. Science, 2020, 368, 724-724.	6.0	131
11	Threshold effect of urinary glycosaminoglycans and the walk test as indicators of disease progression in a survey of subjects with Mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). , 2005, 134A, 144-150.		130
12	A clinical study of 77 patients with mucopolysaccharidosis type II. Acta Paediatrica, International Journal of Paediatrics, 2007, 96, 63-70.	0.7	112
13	Clinical and biochemical study of 28 patients with mucopolysaccharidosis type VI. Clinical Genetics, 2004, 66, 208-213.	1.0	103
14	Heparan sulfate levels in mucopolysaccharidoses and mucolipidoses. Journal of Inherited Metabolic Disease, 2005, 28, 743-757.	1.7	96
15	Enzyme replacement therapy for mucopolysaccharidosis VI: evaluation of longâ€term pulmonary function in patients treated with recombinant human Nâ€acetylgalactosamine 4â€sulfatase. Journal of Inherited Metabolic Disease, 2010, 33, 51-60.	1.7	80
16	Development and Testing of New Screening Method for Keratan Sulfate in Mucopolysaccharidosis IVA. Pediatric Research, 2004, 55, 592-597.	1.1	79
17	Brain MRI in mucopolysaccharidosis. Neurology, 2007, 69, 917-924.	1.5	77
18	Identification and characterization of 13 new mutations in mucopolysaccharidosis type I patients. Molecular Genetics and Metabolism, 2003, 78, 37-43.	0.5	75

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19	Keratan sulphate levels in mucopolysaccharidoses and mucolipidoses. Journal of Inherited Metabolic Disease, 2005, 28, 187-202.	1.7	71
20	CNS involvement in Fabry disease: Clinical and imaging studies before and after 12 months of enzyme replacement therapy. Journal of Inherited Metabolic Disease, 2004, 27, 229-240.	1.7	69
21	Correlation of MR Imaging and MR Spectroscopy Findings with Cognitive Impairment in Mucopolysaccharidosis II. American Journal of Neuroradiology, 2007, 28, 1029-1033.	1.2	65
22	Expression of the disease on female carriers of X-linked lysosomal disorders: a brief review. Orphanet Journal of Rare Diseases, 2010, 5, 14.	1.2	64
23	Evidence that l-Carnitine and Selenium Supplementation Reduces Oxidative Stress in Phenylketonuric Patients. Cellular and Molecular Neurobiology, 2011, 31, 429-436.	1.7	64
24	Mucopolysaccharidoses in Brazil: What happens from birth to biochemical diagnosis?. American Journal of Medical Genetics, Part A, 2008, 146A, 1741-1747.	0.7	63
25	New cases of thalidomide embryopathy in Brazil. Birth Defects Research Part A: Clinical and Molecular Teratology, 2007, 79, 671-672.	1.6	61
26	The germline mutational landscape of BRCA1 and BRCA2 in Brazil. Scientific Reports, 2018, 8, 9188.	1.6	61
27	l-Carnitine Blood Levels and Oxidative Stress in Treated Phenylketonuric Patients. Cellular and Molecular Neurobiology, 2009, 29, 211-218.	1.7	59
28	Enzyme replacement therapy for mucopolysaccharidosis VI: longâ€term cardiac effects of galsulfase (Naglazyme^{Â®}) therapy. Journal of Inherited Metabolic Disease, 2013, 36, 385-394.	1.7	58
29	Enzyme replacement therapy for mucopolysaccharidosis VI: Growth and pubertal development in patients treated with recombinant human N-acetylgalactosamine 4-sulfatase. Journal of Pediatric Rehabilitation Medicine, 2010, 3, 89-100.	0.3	58
30	Phenylketonuria and Gut Microbiota: A Controlled Study Based on Next-Generation Sequencing. PLoS ONE, 2016, 11, e0157513.	1.1	52
31	Exploring the patient journey to diagnosis of Gaucher disease from the perspective of 212 patients with Gaucher disease and 16 Gaucher expert physicians. Molecular Genetics and Metabolism, 2017, 122, 122-129.	0.5	51
32	Oxidative stress in patients with mucopolysaccharidosis type II before and during enzyme replacement therapy. Molecular Genetics and Metabolism, 2011, 103, 121-127.	0.5	48
33	Enzyme replacement therapy for Fabry disease: a systematic review and meta-analysis. Genetics and Molecular Biology, 2012, 35, 947-954.	0.6	45
34	Demographics and patient characteristics of 1209 patients with Gaucher disease: Descriptive analysis from the Gaucher Outcome Survey (GOS). American Journal of Hematology, 2018, 93, 205-212.	2.0	44
35	Evidence that DNA damage is associated to phenylalanine blood levels in leukocytes from phenylketonuric patients. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 2009, 679, 13-16.	0.9	41
36	Identification of a common mutation in mucopolysaccharidosis IVA: correlation among genotype, phenotype, and keratan sulfate. Journal of Human Genetics, 2004, 49, 490-494.	1.1	40

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37	Successful domino liver transplantation in maple syrup urine disease using a related living donor. Brazilian Journal of Medical and Biological Research, 2014, 47, 522-526.	0.7	40
38	Mucopolysaccharidosis type II: Identification of 30 novel mutations among Latin American patients. Molecular Genetics and Metabolism, 2014, 111, 133-138.	0.5	40
39	Presenting signs and patient coâ€variables in Gaucher disease: outcome of the Gaucher Earlier Diagnosis Consensus (GEDâ€C) Delphi initiative. Internal Medicine Journal, 2019, 49, 578-591.	0.5	39
40	Experimental evidence of oxidative stress in plasma of homocystinuric patients: A possible role for homocysteine. Molecular Genetics and Metabolism, 2011, 104, 112-117.	0.5	38
41	Influence of CYP19A1 polymorphisms on the treatment of breast cancer with aromatase inhibitors: a systematic review and meta-analysis. BMC Medicine, 2015, 13, 139.	2.3	36
42	ALG6â€CDG: a recognizable phenotype with epilepsy, proximal muscle weakness, ataxia and behavioral and limb anomalies. Journal of Inherited Metabolic Disease, 2016, 39, 713-723.	1.7	36
43	The lysosomal storage disorders mucolipidosis type II, type III alpha/beta, and type III gamma: Update onGNPTABandGNPTGmutations. Human Mutation, 2019, 40, 842-864.	1.1	36
44	Genetic studies in a cluster of Mucopolysaccharidosis Type VI patients in Northeast Brazil. Molecular Genetics and Metabolism, 2011, 104, 603-607.	0.5	34
45	Prevalence of Hispanic BRCA1 and BRCA2 mutations among hereditary breast and ovarian cancer patients from Brazil reveals differences among Latin American populations. Cancer Genetics, 2016, 209, 417-422.	0.2	33
46	Universal newborn screening: A roadmap for action. Molecular Genetics and Metabolism, 2018, 124, 177-183.	0.5	33
47	Stearoyl-CoA Desaturase-1: Is It the Link between Sulfur Amino Acids and Lipid Metabolism?. Biology, 2015, 4, 383-396.	1.3	30
48	Sleep abnormalities in untreated patients with mucopolysaccharidosis type VI. American Journal of Medical Genetics, Part A, 2011, 155, 1546-1551.	0.7	29
49	Functional capacity evaluation of patients with mucopolysaccharidosis. Journal of Pediatric Rehabilitation Medicine, 2012, 5, 37-46.	0.3	29
50	BRCA1 and BRCA2 mutational profile and prevalence in hereditary breast and ovarian cancer (HBOC) probands from Southern Brazil: Are international testing criteria appropriate for this specific population?. PLoS ONE, 2017, 12, e0187630.	1.1	29
51	Triagem neonatal de distÃºrbios metabÃ³licos. Ciencia E Saude Coletiva, 2002, 7, 129-137.	0.1	28
52	Nerve conduction studies, electromyography and sympathetic skin response in Fabry's disease. Journal of the Neurological Sciences, 2003, 214, 21-25.	0.3	28
53	Mucopolysaccharidosis type VI: Identification of novel mutations on the arylsulphatase B gene in South American patients. Journal of Inherited Metabolic Disease, 2005, 28, 1027-1034.	1.7	28
54	Mucolipidosis II and III alpha/beta in Brazil: Analysis of the GNPTAB gene. Gene, 2013, 524, 59-64.	1.0	27

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55	Living related versus deceased donor liver transplantation for maple syrup urine disease. Molecular Genetics and Metabolism, 2016, 117, 336-343.	0.5	27
56	Quality of life in caregivers of children and adolescents with Osteogenesis Imperfecta. Health and Quality of Life Outcomes, 2015, 13, 41.	1.0	26
57	Efficacy and safety of intravenous laronidase for mucopolysaccharidosis type I: A systematic review and meta-analysis. PLoS ONE, 2017, 12, e0184065.	1.1	26
58	Analyses of disease-related GNPTAB mutations define a novel GlcNAc-1-phosphotransferase interaction domain and an alternative site-1 protease cleavage site. Human Molecular Genetics, 2015, 24, 3497-3505.	1.4	25
59	DNA damage in leukocytes from pretreatment mucopolysaccharidosis type II patients; protective effect of enzyme replacement therapy. Mutation Research - Genetic Toxicology and Environmental Mutagenesis, 2011, 721, 206-210.	0.9	24
60	Classical homocystinuria: A common inborn error of metabolism? An epidemiological study based on genetic databases. Molecular Genetics & Genomic Medicine, 2020, 8, e1214.	0.6	24
61	Natural history of multiple sulfatase deficiency: Retrospective phenotyping and functional variant analysis to characterize an ultraâ€rare disease. Journal of Inherited Metabolic Disease, 2020, 43, 1298-1309.	1.7	23
62	Brain magnetic resonance imaging findings in patients with mucopolysaccharidosis VI. Journal of Inherited Metabolic Disease, 2013, 36, 357-362.	1.7	22
63	Glycogen storage disease type I: clinical and laboratory profile. Jornal De Pediatria, 2014, 90, 572-579.	0.9	22
64	Healthâ€related quality of life in paediatric patients with intoxicationâ€type inborn errors of metabolism: Analysis of an international data set. Journal of Inherited Metabolic Disease, 2021, 44, 215-225.	1.7	22
65	Serum Markers of Neurodegeneration in Maple Syrup Urine Disease. Molecular Neurobiology, 2017, 54, 5709-5719.	1.9	21
66	Sensitivity, advantages, limitations, and clinical utility of targeted next-generation sequencing panels for the diagnosis of selected lysosomal storage disorders. Genetics and Molecular Biology, 2019, 42, 197-206.	0.6	21
67	Esophageal stenosis in a child presenting a de novo 7q terminal deletion. European Journal of Medical Genetics, 2010, 53, 333-336.	0.7	20
68	Effects of imiglucerase on the growth and metabolism of Gaucher disease type I patients: a systematic review. Nutrition and Metabolism, 2013, 10, 34.	1.3	20
69	Reported outcomes of 453 pregnancies in patients with Gaucher disease: An analysis from the Gaucher outcome survey. Blood Cells, Molecules, and Diseases, 2018, 68, 226-231.	0.6	20
70	Atypical macrocephaly-cutis marmorata telangiectatica congenita with retinoblastoma. Clinical Dysmorphology, 2002, 11, 199-202.	0.1	19
71	Enzyme replacement therapy for Mucopolysaccharidosis Type I among patients followed within the MPS Brazil Network. Genetics and Molecular Biology, 2014, 37, 23-29.	0.6	19
72	Biotinidase deficiency: Genotype-biochemical phenotype association in Brazilian patients. PLoS ONE, 2017, 12, e0177503.	1.1	19

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73	Clinical and biochemical study of 29 Brazilian patients with metachromatic leukodystrophy. Journal of Inherited Metabolic Disease, 2010, 33, 257-262.	1.7	18
74	Maple syrup urine disease in Brazil: a panorama of the last two decades. Jornal De Pediatria, 2015, 91, 292-298.	0.9	18
75	<scp>CBS</scp> mutations are good predictors for B6â€responsiveness: A study based on the analysis of 35 Brazilian Classical Homocystinuria patients. Molecular Genetics & Genomic Medicine, 2018, 6, 160-170.	0.6	18
76	Characteristics of 26 patients with type 3 Gaucher disease: A descriptive analysis from the Gaucher Outcome Survey. Molecular Genetics and Metabolism Reports, 2018, 14, 73-79.	0.4	18
77	Health-related quality of life of children and adolescents with osteogenesis imperfecta: a cross-sectional study using PedsQLâ„¢. BMC Pediatrics, 2018, 18, 95.	0.7	18
78	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome): assessment of joint mobility and grip and pinch strength. Jornal De Pediatria, 2008, 84, 130-5.	0.9	18
79	Quality of Life of Brazilian Patients with Gaucher Disease and Fabry Disease. JIMD Reports, 2012, 7, 31-37.	0.7	17
80	In vitro digestion of starches in a dynamic gastrointestinal model: an innovative study to optimize dietary management of patients with hepatic glycogen storage diseases. Journal of Inherited Metabolic Disease, 2015, 38, 529-536.	1.7	17
81	Quality of life and adherence to treatment in early-treated Brazilian phenylketonuria pediatric patients. Brazilian Journal of Medical and Biological Research, 2018, 51, e6709.	0.7	17
82	Hepatic glycogen storage diseases are associated to microbial dysbiosis. PLoS ONE, 2019, 14, e0214582.	1.1	17
83	Multiple sulfatase deficiency: clinical report and description of two novel mutations in a Brazilian patient. Metabolic Brain Disease, 2009, 24, 493-500.	1.4	16
84	Severe phenotype in MPS II patients associated with a large deletion including contiguous genes. American Journal of Medical Genetics, Part A, 2012, 158A, 1055-1059.	0.7	16
85	Biotinidase deficiency: clinical and genetic studies of 38 Brazilian patients. BMC Medical Genetics, 2014, 15, 96.	2.1	16
86	Rare disease landscape in Brazil: report of a successful experience in inborn errors of metabolism. Orphanet Journal of Rare Diseases, 2016, 11, 76.	1.2	16
87	Analysis of body composition and nutritional status in Brazilian phenylketonuria patients. Molecular Genetics and Metabolism Reports, 2016, 6, 16-20.	0.4	16
88	Placenta analysis of prenatally diagnosed patients reveals early GAG storage in mucopolysaccharidoses II and VI. Molecular Genetics and Metabolism, 2011, 103, 197-198.	0.5	15
89	Origin and spread of a common deletion causing mucolipidosis type II: insights from patterns of haplotypic diversity. Clinical Genetics, 2011, 80, 273-280.	1.0	15
90	Ethical issues related to the access to orphan drugs in Brazil: the case of mucopolysaccharidosis type I. Journal of Medical Ethics, 2011, 37, 233-239.	1.0	15

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91	Body composition in patients with classical homocystinuria: body mass relates to homocysteine and choline metabolism. Gene, 2014, 546, 443-447.	1.0	15
92	Evaluation of plasma biomarkers of inflammation in patients with maple syrup urine disease. Journal of Inherited Metabolic Disease, 2018, 41, 631-640.	1.7	15
93	Evaluation of the frequency of non-motor symptoms of Parkinsonâ€™s disease in adult patients with Gaucher disease type 1. Orphanet Journal of Rare Diseases, 2019, 14, 103.	1.2	15
94	<scp>SARSâ€CoV</scp>â€2 pandemic in the Brazilian community of rare diseases: A patient reported survey. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 301-311.	0.7	15
95	Unique frequency of known mutations in Brazilian MPS I patients. , 2000, 90, 108-109.		14
96	The microbiome and inborn errors of metabolism: Why we should look carefully at their interplay?. Genetics and Molecular Biology, 2018, 41, 515-532.	0.6	14
97	Domino Liver Transplant in Maple Syrup Urine Disease: Technical Details of Cases in Which the First Surgery Involved a Living Donor. Transplantation, 2019, 103, 536-543.	0.5	14
98	Liver manifestations in a cohort of 39 patients with congenital disorders of glycosylation: pin-pointing the characteristics of liver injury and proposing recommendations for follow-up. Orphanet Journal of Rare Diseases, 2021, 16, 20.	1.2	14
99	Evaluation of orofacial motricity in patients with mucopolysaccharidosis: a cross-sectional study. Jornal De Pediatria, 2009, 85, 254-260.	0.9	14
100	Access to treatment for phenylketonuria by judicial means in Rio Grande do Sul, Brazil. Ciencia E Saude Coletiva, 2015, 20, 1607-1616.	0.1	13
101	Determination of amylose/amylopectin ratio of starches. Journal of Inherited Metabolic Disease, 2015, 38, 985-986.	1.7	13
102	Enigmatic in vivo GlcNAc-1-phosphotransferase (GNPTG) transcript correction to wild type in two mucolipidosis III gamma siblings homozygous for nonsense mutations. Journal of Human Genetics, 2016, 61, 555-560.	1.1	13
103	A convenient approach to facilitate monitoring Gaucher disease progression and therapeutic response. Analyst, The, 2017, 142, 3380-3387.	1.7	13
104	Glycogen storage diseases: Twentyâ€seven new variants in a cohort of 125 patients. Molecular Genetics & Genomic Medicine, 2019, 7, e877.	0.6	13
105	Clinical and biochemical studies in mucopolysaccharidosis type II carriers. Journal of Inherited Metabolic Disease, 2009, 32, 732-738.	1.7	12
106	Gaucher disease type I: Assessment of basal metabolic rate in patients from southern Brazil. Blood Cells, Molecules, and Diseases, 2011, 46, 42-46.	0.6	12
107	Are MPS II heterozygotes actually asymptomatic? A study based on clinical and biochemical data, Xâ€inactivation analysis and imaging evaluations. American Journal of Medical Genetics, Part A, 2011, 155, 50-57.	0.7	12
108	Dentomaxillofacial manifestations of mucopolysaccharidosis VI: clinical and imaging findings from two cases, with an emphasis on the temporomandibular joint. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2013, 116, e141-e148.	0.2	12

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109	Feeding Difficulties and Orofacial Myofunctional Disorder in Patients with Hepatic Glycogen Storage Diseases. JIMD Reports, 2018, 45, 21-27.	0.7	12
110	Prevalence and causes of congenital microcephaly in the absence of a Zika virus outbreak in southern Brazil. Jornal De Pediatria, 2019, 95, 600-606.	0.9	12
111	Ocular manifestations in classic homocystinuria. Ophthalmic Genetics, 2021, 42, 71-74.	0.5	12
112	Epidemiological aspects of hereditary fructose intolerance: A database study. Human Mutation, 2021, 42, 1548-1566.	1.1	12
113	Food Neophobia in Patients With Phenylketonuria. Journal of Endocrinology and Metabolism, 2019, 9, 108-112.	0.1	12
114	Mucopolysaccharidosis VII: clinical, biochemical and molecular investigation of a Brazilian family. Clinical Genetics, 2003, 64, 172-175.	1.0	11
115	Mucopolysaccharidosis. FIRE Forum for International Research in Education, 2015, 3, 232640981561380.	0.7	11
116	Acute exercise in treated phenylketonuria patients: Physical activity and biochemical response. Molecular Genetics and Metabolism Reports, 2015, 5, 55-59.	0.4	11
117	Brain-derived neurotrophic factor expression increases after enzyme replacement therapy in Gaucher disease. Journal of Neuroimmunology, 2015, 278, 190-193.	1.1	11
118	GNPTAB missense mutations cause loss of GlcNAc-1-phosphotransferase activity in mucolipidosis type II through distinct mechanisms. International Journal of Biochemistry and Cell Biology, 2017, 92, 90-94.	1.2	11
119	Efficacy of speech therapy in post-intubation patients with oropharyngeal dysphagia: a randomized controlled trial. CoDAS, 2021, 33, e20190246.	0.2	11
120	Three Main Causes of Homocystinuria: CBS, cblC and MTHFR Deficiency. What do they Have in Common?. Journal of Inborn Errors of Metabolism and Screening, 0, 7, .	0.3	11
121	A Systematic Review and Meta-Analysis of Enzyme Replacement Therapy in Late-Onset Pompe Disease. Journal of Clinical Medicine, 2021, 10, 4828.	1.0	11
122	Epidemiology of rare diseases in Brazil: protocol of the Brazilian Rare Diseases Network (RARAS-BRDN). Orphanet Journal of Rare Diseases, 2022, 17, 84.	1.2	11
123	Acromegaloid facial appearance and hypertrichosis: a case suggesting autosomal recessive inheritance. Clinical Dysmorphology, 2004, 13, 49-50.	0.1	10
124	The prognostic value of the serum ferritin in a southern Brazilian cohort of patients with Gaucher disease. Genetics and Molecular Biology, 2016, 39, 30-34.	0.6	10
125	Is the gut microbiota dysbiotic in patients with classical homocystinuria?. Biochimie, 2020, 173, 3-11.	1.3	10
126	Prospective study of 11 Brazilian patients with mucopolysaccharidosis II. Jornal De Pediatria, 2006, 82, 273-278.	0.9	10

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127	Development and validation of Gaucher disease type 1 (GD1)-specific patient-reported outcome measures (PROMs) for clinical monitoring and for clinical trials. Orphanet Journal of Rare Diseases, 2022, 17, 9.	1.2	10
128	Mucopolissacaridose tipo VI (sÃ¬ndrome de Maroteaux-Lamy): avaliaÃ§Ã£o da mobilidade articular e das forÃ§as de garra e de pinÃ§a. Jornal De Pediatria, 2008, 84, .	0.9	9
129	Cardiac disease as the presenting feature of mucopolysaccharidosis type IIIA: A case report. Molecular Genetics and Metabolism Reports, 2014, 1, 422-424.	0.4	9
130	A de novo or germline mutation in a family with Mucolipidosis III gamma: Implications for molecular diagnosis and genetic counseling. Molecular Genetics and Metabolism Reports, 2014, 1, 98-102.	0.4	9
131	Osteopontin: a potential biomarker of Gaucher disease. Annals of Hematology, 2015, 94, 1119-1125.	0.8	9
132	Could enzyme replacement therapy promote immune tolerance in Gaucher disease type 1?. Blood Cells, Molecules, and Diseases, 2018, 68, 200-202.	0.6	9
133	Information and Diagnosis Networks â€“ tools to improve diagnosis and treatment for patients with rare genetic diseases. Genetics and Molecular Biology, 2019, 42, 155-164.	0.6	9
134	Genetic analysis of patients with fructose-1,6-bisphosphatase deficiency. Gene, 2019, 699, 102-109.	1.0	9
135	Transferrin isoelectric focusing for the investigation of congenital disorders of glycosylation: analysis of a ten-year experience in a Brazilian center. Jornal De Pediatria, 2020, 96, 710-716.	0.9	9
136	Liver involvement in patients with Gaucher disease types I and III. Molecular Genetics and Metabolism Reports, 2020, 22, 100564.	0.4	9
137	Topiramate is effective for status epilepticus and seizure control in neuraminidase deficiency. Arquivos De Neuro-Psiquiatria, 2011, 69, 565-566.	0.3	9
138	Effects of imilglucerase withdrawal on an adult with gaucher disease. British Journal of Haematology, 2001, 113, 1088-1089.	1.2	8
139	Enzyme Replacement Therapy in a Patient with Gaucher Disease Type III: A Paradigmatic Case Showing Severe Adverse Reactions Started a Long Time After the Beginning of Treatment. JIMD Reports, 2013, 11, 1-6.	0.7	8
140	Ghrelin, leptin and adiponectin levels in Gaucher disease type I patients on enzyme replacement therapy. Clinical Nutrition, 2015, 34, 727-731.	2.3	8
141	Clinical Characterization of Mucolipidoses II and III: A Multicenter Study. Journal of Pediatric Genetics, 2019, 08, 198-204.	0.3	8
142	Time to fight the pandemic setbacks for caregiver academics. Nature Human Behaviour, 2021, 5, 1262-1262.	6.2	8
143	Mucopolysaccharidoses in northern Brazil: Targeted mutation screening and urinary glycosaminoglycan excretion in patients undergoing enzyme replacement therapy. Genetics and Molecular Biology, 2011, 34, 410-415.	0.6	7
144	Extension of the molecular analysis to the promoter region of the iduronate 2-sulfatase gene reveals genomic alterations in mucopolysaccharidosis type II patients with normal coding sequence. Gene, 2013, 526, 150-154.	1.0	7

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145	Pitfalls in the prenatal diagnosis of mucolipidosis II alpha/beta: A case report. Meta Gene, 2014, 2, 403-406.	0.3	7
146	Exome sequencing for mucolipidosis III: Detection of a novel GNPTAB gene mutation in a patient with a very mild phenotype. Molecular Genetics and Metabolism Reports, 2015, 2, 34-37.	0.4	7
147	Medical Costs Related to Enzyme Replacement Therapy for Mucopolysaccharidosis Types I, II, and VI in Brazil: A Multicenter Study. Value in Health Regional Issues, 2015, 8, 99-106.	0.5	7
148	Maple syrup urine disease in Brazilian patients: variants and clinical phenotype heterogeneity. Orphanet Journal of Rare Diseases, 2020, 15, 309.	1.2	7
149	Maternity in the Brazilian CV Lattes: when will it become a reality?. Anais Da Academia Brasileira De Ciencias, 2021, 93, e20201370.	0.3	7
150	The Management of Gaucher Disease in Developing Countries: A Successful Experience in Southern Brazil. Public Health Genomics, 2010, 13, 27-33.	0.6	6
151	Optimized loading test to evaluate responsiveness to tetrahydrobiopterin (BH4) in Brazilian patients with phenylalanine hydroxylase deficiency. Molecular Genetics and Metabolism, 2011, 104, S80-S85.	0.5	6
152	Should neonatal hyperparathyroidism associated with mucolipidosis II/III be treated pharmacologically?a. Journal of Pediatric Endocrinology and Metabolism, 2013, 26, 1011-3.	0.4	6
153	Leptin concentrations and SCD-1 indices in classical homocystinuria: Evidence for the role of sulfur amino acids in the regulation of lipid metabolism. Clinica Chimica Acta, 2017, 473, 82-88.	0.5	6
154	Effect of BTD gene variants on in vitro biotinidase activity. Molecular Genetics and Metabolism, 2019, 127, 361-367.	0.5	6
155	Current Practices and Challenges in the Diagnosis and Management of PKU in Latin America: A Multicenter Survey. Nutrients, 2021, 13, 2566.	1.7	6
156	Tetrahydrobiopterin responsiveness of patients with phenylalanine hydroxylase deficiency. Jornal De Pediatria, 2011, 87, 245-251.	0.9	6
157	Further cases of â€œneighborâ€•mutations in mucopolysaccharidosis type II. American Journal of Medical Genetics, Part A, 2006, 140A, 1684-1686.	0.7	5
158	A Brazilian galactosialidosis patient given renal transplantation: A case report. Journal of Inherited Metabolic Disease, 2008, 31, 205-208.	1.7	5
159	Prenatal diagnosis of mucopolysaccharidosis VI by enzyme assay in a dried spot of fetal blood: a pioneering case report. Prenatal Diagnosis, 2010, 30, 89-90.	1.1	5
160	Visual Dysfunction of Type I and VI Mucopolysaccharidosis Patients Evaluated with Visual Evoked Cortical Potential. Case Reports in Ophthalmology, 2012, 3, 104-112.	0.3	5
161	Combined in vitro and in silico analyses of missense mutations in GNPTAB provide new insights into the molecular bases of mucolipidosis II and III alpha/beta. Human Mutation, 2020, 41, 133-139.	1.1	5
162	Assessment of cellular cobalamin metabolism in Gaucher disease. BMC Medical Genetics, 2020, 21, 12.	2.1	5

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163	Bone marrow burden score is not useful as a follow-up parameter in stable patients with type 1 Gaucher disease after 5Âyears of treatment. Blood Cells, Molecules, and Diseases, 2021, 90, 102591.	0.6	5
164	Impact of the COVID-19 pandemic on the standard of care for patients with lysosomal storage diseases: A survey of healthcare professionals in the Fabry, Gaucher, and Hunter Outcome Survey registries. Molecular Genetics and Metabolism Reports, 2021, 28, 100788.	0.4	5
165	Phenylketonuria Diagnosis by Massive Parallel Sequencing and Genotype-Phenotype Association in Brazilian Patients. Genes, 2021, 12, 20.	1.0	5
166	KIR genes and HLA class I ligands in Gaucher disease. Gene, 2013, 516, 53-57.	1.0	4
167	Does phase angle correlate with hyperhomocysteinemia? A study of patients with classical homocystinuria. Clinical Nutrition, 2013, 32, 479-480.	2.3	4
168	Putting the child at the centre of interâ€professional cooperation in outâ€ofâ€home care. Child and Family Social Work, 2017, 22, 992-999.	0.6	4
169	Nutritional Status and Body Composition in Patients With Hepatic Glycogen Storage Diseases Treated With Uncooked Cornstarchâ€"A Controlled Study. FIRE Forum for International Research in Education, 2017, 5, 232640981773301.	0.7	4
170	Infant mortality in Brazil attributable to inborn errors of metabolism associated with sudden death: a time-series study (2002â€"2014). BMC Pediatrics, 2019, 19, 52.	0.7	4
171	Hepatocellular carcinoma in Gaucher disease: Reinforcing the proposed guidelines. Blood Cells, Molecules, and Diseases, 2019, 74, 34-36.	0.6	4
172	Imbalanced cellular metabolism compromises cartilage homeostasis and joint function in a mouse model of mucolipidosis type III gamma. DMM Disease Models and Mechanisms, 2020, 13, .	1.2	4
173	Outcomes of screening for gammopathies in children and adults with Gaucher disease type 1 in a cohort from Brazil and the United States. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 1052-1059.	0.7	4
174	A triple-blinded crossover study to evaluate the short-term safety of sweet manioc starch for the treatment of glycogen storage disease type Ia. Orphanet Journal of Rare Diseases, 2021, 16, 254.	1.2	4
175	Cardiovascular findings in classic homocystinuria. Molecular Genetics and Metabolism Reports, 2020, 25, 100693.	0.4	4
176	Development of an inventory to assess perceived barriers related to PKU treatment. Journal of Patient-Reported Outcomes, 2020, 4, 29.	0.9	4
177	Prevalence of the most common pathogenic variants in three genes for inborn errors of metabolism associated with sudden unexpected death in infancy: a population-based study in south Brazil. Genetics and Molecular Biology, 2020, 43, 20190298.	0.6	4
178	X-linked adrenoleukodystrophy: clinical and laboratory findings in 15 Brazilian patients. Genetics and Molecular Biology, 2000, 23, 261-264.	0.6	3
179	Analysis of cDNA Molecules is Not Suitable for the Molecular Diagnosis of Mucopolysaccharidosis Type I. Diagnostic Molecular Pathology, 2012, 21, 53-55.	2.1	3
180	Human leukocyte antigens and Gaucher disease. Blood Cells, Molecules, and Diseases, 2013, 50, 202-205.	0.6	3

#	Article	IF	Citations
181	New approaches to the treatment of orphan genetic disorders: Mitigating molecular pathologies using chemicals. Anais Da Academia Brasileira De Ciencias, 2015, 87, 1375-1388.	0.3	3
182	Use of a multiplex ligation-dependent probe amplification method for the detection of deletions/duplications in the GBA1 gene in Gaucher disease patients. Blood Cells, Molecules, and Diseases, 2018, 68, 17-20.	0.6	3
183	Attention-deficit hyperactivity disorder in Brazilian patients with phenylketonuria. Acta Neurologica Belgica, 2020, 120, 893-899.	0.5	3
184	Factors that increase risk for poor adherence to phenylketonuria treatment in Brazilian patients. American Journal of Medical Genetics, Part A, 2021, 185, 1991-2002.	0.7	3
185	Jones-Waldman syndrome: Another report?. American Journal of Medical Genetics Part A, 1994, 51, 83-83.	2.4	2
186	AvaliaÃ§Ã£o da motricidade orofacial em pacientes com mucopolissacaridose: um estudo transversal. Jornal De Pediatria, 2009, 85, 254-260.	0.9	2
187	Is lipid metabolism altered in classical homocystinuria?. Molecular Genetics and Metabolism, 2012, 106, 382-383.	0.5	2
188	Hyperimmunoglobulinemia in pediatric Gaucher patients in Southern Brazil. Pediatric Blood and Cancer, 2012, 59, 339-339.	0.8	2
189	Breastfeeding in Gaucher Disease: Is Enzyme Replacement Therapy Safe?. Clinical Therapeutics, 2014, 36, 990-991.	1.1	2
190	Cytokines levels in late-diagnosed Classical Homocystinuria patients. Molecular Genetics and Metabolism Reports, 2018, 17, 43-44.	0.4	2
191	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: data from the E-HOD registry. Journal of Inherited Metabolic Disease, 2018, , .	1.7	2
192	Diagnosis and Management of Classical Homocystinuria in Brazil. FIRE Forum for International Research in Education, 2018, 6, 232640981878890.	0.7	2
193	Chitotriosidase on treatment-naÃ¯ve patients with Gaucher disease: A genotype vs phenotype study. Clinica Chimica Acta, 2019, 492, 1-6.	0.5	2
194	Rare GBA1 genotype associated with severe bone disease in Gaucher disease type 1. Molecular Genetics and Metabolism Reports, 2019, 21, 100544.	0.4	2
195	KHK inhibition for the treatment of hereditary fructose intolerance and nonalcoholic fatty liver disease: a double-edged sword. Cellular and Molecular Life Sciences, 2020, 77, 3465-3466.	2.4	2
196	Molecular basis of various forms of maple syrup urine disease in Chilean patients. Molecular Genetics & Genomic Medicine, 2021, 9, e1616.	0.6	2
197	Pathogenic variants in GNPTAB and GNPTG encoding distinct subunits of GlcNAc-1-phosphotransferase differentially impact bone resorption in patients with mucolipidosis type II and III. Genetics in Medicine, 2021, 23, 2369-2377.	1.1	2
198	Bone Mineral Density in Patients with Hepatic Glycogen Storage Diseases. Nutrients, 2021, 13, 2987.	1.7	2

#	Article	IF	Citations
199	The fructose-1,6-bisphosphatase deficiency and the p.(Lys204ArgfsTer72) variant. Genetics and Molecular Biology, 2021, 44, e20200281.	0.6	2
200	Monitoring of Phenylalanine Levels in Patients with Phenylketonuria Using Dried Blood Spots: a Comparison of Two Methods. Journal of Inborn Errors of Metabolism and Screening, 0, 8, .	0.3	2
201	Characterization of the 3'UTR of the BTD gene and identification of regulatory elements and microRNAs. Genetics and Molecular Biology, 2022, 45, e20200432.	0.6	2
202	Efficacy and Safety of Taliglucerase Alfa for the Treatment of Gaucher Disease: A 9-Year Experience. Journal of Inborn Errors of Metabolism and Screening, 0, 10, .	0.3	2
203	A patient presenting a 22q13 deletion associated with an apparently balanced translocation t(16;22): an illustrative case in the investigation of patients with low ARSA activity. Genetics and Molecular Biology, 2012, 35, 424-427.	0.6	1
204	Screening of high-risk Gaucher disease patients using dried blood spots techniques. Gene, 2013, 523, 114-115.	1.0	1
205	Maple syrup urine disease in Brazil: a panorama of the last two decades. Jornal De Pediatria (VersÃ£o Em) Tj ETQq1 1 0.784314 rgBT /	0.2	1
206	Does enzyme replacement therapy enhance brain-derived neurotrophic factor expression in Gaucher disease?. Journal of Neuroimmunology, 2015, 283, 63.	1.1	1
207	Next-generation sequencing corroborates a probable de novo GNPTG variation previously detected by Sanger sequencing. Molecular Genetics and Metabolism Reports, 2017, 11, 92-93.	0.4	1
208	Mucopolysaccharidosis VI and effects on growth of the apical bases: a case report. Special Care in Dentistry, 2018, 38, 176-184.	0.4	1
209	Breastfeeding in patients with Gaucher disease: Is taliglucerase alfa safe?. Molecular Genetics and Metabolism Reports, 2019, 18, 30-31.	0.4	1
210	Haplotype analysis and origin of the most common pathogenic mutation causing Mucolipidosis II and III alpha/beta in Brazilian patients. Gene Reports, 2020, 19, 100645.	0.4	1
211	Assessment of quality of life in Gaucher disease: A methodological approach. Molecular Genetics & Genomic Medicine, 2021, 9, e1549.	0.6	1
212	Histomorphometric analysis of liver biopsies of treated patients with Gaucher disease type 1. Autopsy and Case Reports, 2021, 11, e2021306.	0.2	1
213	Cartas ao Editor. Arquivos Brasileiros De Oftalmologia, 2007, 70, 563-564.	0.2	1
214	AnÃ¡lise da densidade mineral Ã³ssea em pacientes com fenilcetonÃºria e sua correlaÃ§Ã£o com parÃ¢metros nutricionais. Clinical and Biomedical Research, 2019, 39, 24-31.	0.1	1
215	Punctate calcifications in lysosomal storage disorders. Clinical Dysmorphology, 2009, 18, 172-177.	0.1	0
216	Assessment of Basal Metabolic Rate and Nutritional Status in Patients with Gaucher Disease Type III. JIMD Reports, 2013, 14, 37-42.	0.7	0

#	Article	IF	Citations
217	Serum Î²2-microglobulin is frequently elevated in type 1 Gaucher patients. Molecular Genetics and Metabolism Reports, 2015, 2, 38-40.	0.4	0
218	Inflammasome during pregnancy in a Gaucher disease patient. Molecular Genetics and Metabolism, 2015, 114, S120.	0.5	0
219	Reply to Letter to the Editor â€“ Leptin levels in Gaucher disease type I patients: A methodological approach. Clinical Nutrition, 2015, 34, 329.	2.3	0
220	Relation between homocysteine and vitamin B12 levels in Brazilian patients with Gaucher disease. Molecular Genetics and Metabolism, 2017, 120, S24-S25.	0.5	0
221	Reported outcomes of 453 pregnancies in patients with Gaucher disease: an analysis from the Gaucher Outcome Survey. Molecular Genetics and Metabolism, 2017, 120, S80.	0.5	0
222	Taliglucerase alfa and type 1 Gaucher disease: a south Brazilian experience. Molecular Genetics and Metabolism, 2017, 120, S105-S106.	0.5	0
223	Gaucheroma mimicking hepatocellular carcinoma in a cirrhotic type I Gaucher disease patient. Molecular Genetics and Metabolism, 2018, 123, S135.	0.5	0
224	Rareâ€¯GBA1â€¯genotype in two siblings with a severe bone phenotype of type 1 Gaucher disease. Molecular Genetics and Metabolism, 2019, 126, S115.	0.5	0
225	Humoral immune response in adult Brazilian patients with Mucolipidosis III gamma. Genetics and Molecular Biology, 2019, 42, 571-573.	0.6	0
226	Liver biopsy findings in patients with Gaucher disease: Experience of the reference center of Rio Grande do Sul, Brazil. Molecular Genetics and Metabolism, 2019, 126, S137-S138.	0.5	0
227	The rs2229611 (G6PC:c.*23ÂT>C) is associated with glycogen storage disease type Ia in Brazilian patients. Molecular Genetics and Metabolism Reports, 2020, 25, 100659.	0.4	0
228	Value of CSF Biomarkers in Predicting Risk of Progression from aMCI to ADD in a 5-Year Follow-Up Cohort. SN Comprehensive Clinical Medicine, 2020, 2, 1543-1550.	0.3	0
229	Concerning 'Liver steatosis is highly prevalent and is associated with metabolic risk factors and liver fibrosis in adult patients with type 1 Gaucher disease' by Nascimbeni et al.. Liver International, 2021, 41, 226-226.	1.9	0
230	Website www.emergencyprotocol.net to Support Prevention of Metabolic Emergencies in Patients with Hepatic Glycogen Storage Diseases and Fatty Acid Oxidation Disorders. Journal of Inborn Errors of Metabolism and Screening, 0, 9, .	0.3	0
231	Elevated holoâ€•transcobalamin in Gaucher disease type II : AÂcase report. American Journal of Medical Genetics, Part A, 2021, 185, 2471-2476.	0.7	0
232	HipoventilaÃ§Ã£o relacionada ao sono de origem central secundÃ¡ria Ã deficiÃªncia de biotinidase: relato de caso. Medicina, 2021, 54, e166390.	0.0	0
233	A decade of molecular diagnosis of Mucolipidosis II and III in Brazil: a pooled analysis of 32 patients. Journal of Inborn Errors of Metabolism and Screening, 0, 9, .	0.3	0
234	Prevalence of thrombophilia and thrombotic events inpatients with Fabry disease in a reference center forlysosomal disorders in Southern Brazil. Clinical and Biomedical Research, 2016, 36, 23-26.	0.1	0

#	Article	IF	Citations
235	Introduction to the special issue on Clinical Genetics in Latin America. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 873-875.	0.7	0
236	Body composition in patients with hepatic glycogen storage diseases. Nutrition, 2022, , 111763.	1.1	0
237	Medium-chain acyl-CoA dehydrogenase deficiency: prevalence of ACADM pathogenic variants c.985A>G and c.199T>C in a healthy population in Rio Grande do Sul, Brazil. Reproductive and Developmental Medicine, 2022, 6, 92-97.	0.2	0