Roberto Giugliani

List of Publications by Citations

441 papers	12,981 citations	60 h-index	93 g-index
516 ext. papers	15,015 ext. citations	3.5 avg, IF	6.06 L-index

#	Paper	IF	Citations
441	A phase II/III clinical study of enzyme replacement therapy with idursulfase in mucopolysaccharidosis II (Hunter syndrome). *Genetics in Medicine*, **2006**, 8, 465-73	8.1	416
440	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	3.6	297
439	Treatment of Fabry Disease with the Pharmacologic Chaperone Migalastat. *New England Journal of Medicine*, **2016**, 375, 545-55	59.2	254
438	Enzyme replacement therapy with agalsidase alfa in patients with Fabry disease: an analysis of registry data. *Lancet, The*, **2009**, 374, 1986-96	40	212
437	Recognition and diagnosis of mucopolysaccharidosis II (Hunter syndrome). *Pediatrics*, **2008**, 121, e377-86	7.4	211
436	Natural course of Fabry disease: changing pattern of causes of death in FOS - Fabry Outcome Survey. *Journal of Medical Genetics*, **2009**, 46, 548-52	5.8	206
435	Management guidelines for mucopolysaccharidosis VI. *Pediatrics*, **2007**, 120, 405-18	7.4	182
434	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	3.7	181
433	Cardiac disease in patients with mucopolysaccharidosis: presentation, diagnosis and management. *Journal of Inherited Metabolic Disease*, **2011**, 34, 1183-97	5.4	175
432	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, e681-9	7.4	172
431	Long-term, open-labeled extension study of idursulfase in the treatment of Hunter syndrome. *Genetics in Medicine*, **2011**, 13, 95-101	8.1	156
430	Efficacy and safety of enzyme replacement therapy with BMN 110 (elosulfase alfa) for Morquio A syndrome (mucopolysaccharidosis IVA): a phase 3 randomised placebo-controlled study. *Journal of Inherited Metabolic Disease*, **2014**, 37, 979-90	5.4	152
429	Magnetic resonance imaging findings in Hunter syndrome. *Acta Paediatrica, International Journal of Paediatrics*, **2008**, 97, 61-8	3.1	147
428	Primary renal hypoplasia in humans and mice with PAX2 mutations: evidence of increased apoptosis in fetal kidneys of Pax2(1Neu) +/- mutant mice. *Human Molecular Genetics*, **2000**, 9, 1-11	5.6	147
427	The TP53 mutation, R337H, is associated with Li-Fraumeni and Li-Fraumeni-like syndromes in Brazilian families. *Cancer Letters*, **2007**, 245, 96-102	9.9	144
426	Multidisciplinary management of Hunter syndrome. *Pediatrics*, **2009**, 124, e1228-39	7.4	137
425	Neurologic findings in Machado-Joseph disease: relation with disease duration, subtypes, and (CAG)n. *Archives of Neurology*, **2001**, 58, 899-904		132

424	Initial report from the Hunter Outcome Survey. *Genetics in Medicine*, **2008**, 10, 508-16	8.1	131
423	Epidemiology of mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2017**, 121, 227-240	3.7	128
422	The natural history of juvenile or subacute GM2 gangliosidosis: 21 new cases and literature review of 134 previously reported. *Pediatrics*, **2006**, 118, e1550-62	7.4	126
421	Mucopolysaccharidosis I, II, and VI: Brief review and guidelines for treatment. *Genetics and Molecular Biology*, **2010**, 33, 589-604	2	117
420	A prospective, cross-sectional survey study of the natural history of Niemann-Pick disease type B. *Pediatrics*, **2008**, 122, e341-9	7.4	114
419	Congenital adrenal hyperplasia due to 21-hydroxylase deficiency: Newborn screening and its relationship to the diagnosis and treatment of the disorder. *Screening: Journal of the International Society of Neonatal Screening*, **1993**, 2, 105-139		112
418	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, 144-50		111
417	The validation of pharmacogenetics for the identification of Fabry patients to be treated with migalastat. *Genetics in Medicine*, **2017**, 19, 430-438	8.1	110
416	Novel mutations of ND genes in complex I deficiency associated with mitochondrial encephalopathy. *Brain*, **2007**, 130, 1894-904	11.2	109
415	Intrathecal enzyme replacement therapy in a patient with mucopolysaccharidosis type I and symptomatic spinal cord compression. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 2538-44	2.5	104
414	Clinical course of sly syndrome (mucopolysaccharidosis type VII). *Journal of Medical Genetics*, **2016**, 53, 403-18	5.8	103
413	The natural history of MPS I: global perspectives from the MPS I Registry. *Genetics in Medicine*, **2014** , 16, 759-65	8.1	102
412	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	3.7	102
411	Respiratory and sleep disorders in mucopolysaccharidosis. *Journal of Inherited Metabolic Disease*, **2013**, 36, 201-10	5.4	98
410	Mutational analysis of 105 mucopolysaccharidosis type VI patients. *Human Mutation*, **2007**, 28, 897-903	4.7	98
409	A clinical study of 77 patients with mucopolysaccharidosis type II. *Acta Paediatrica, International Journal of Paediatrics*, **2007**, 96, 63-70	3.1	97
408	Clinical and biochemical study of 28 patients with mucopolysaccharidosis type VI. *Clinical Genetics*, **2004**, 66, 208-13	4	97
407	Twelve different enzyme assays on dried-blood filter paper samples for detection of patients with selected inherited lysosomal storage diseases. *Clinica Chimica Acta*, **2006**, 372, 98-102	6.2	92

406	International guidelines for the management and treatment of Morquio A syndrome. *American Journal of Medical Genetics, Part A*, **2015**, 167A, 11-25	2.5	86
405	Type B Niemann-Pick disease: findings at chest radiography, thin-section CT, and pulmonary function testing. *Radiology*, **2006**, 238, 339-45	20.5	86
404	Safety and pharmacodynamic effects of a pharmacological chaperone on α-galactosidase A activity and globotriaosylceramide clearance in Fabry disease: report from two phase 2 clinical studies. *Orphanet Journal of Rare Diseases*, **2012**, 7, 91	4.2	84
403	Globotriaosylceramide is correlated with oxidative stress and inflammation in Fabry patients treated with enzyme replacement therapy. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2012**, 1822, 226-32	6.9	82
402	A survey of spinocerebellar ataxia in South Brazil - 66 new cases with Machado-Joseph disease, SCA7, SCA8, or unidentified disease-causing mutations. *Journal of Neurology*, **2001**, 248, 870-6	5.5	79
401	Detection of R337H, a germline TP53 mutation predisposing to multiple cancers, in asymptomatic women participating in a breast cancer screening program in Southern Brazil. *Cancer Letters*, **2008**, 261, 21-5	9.9	78
400	Long-term effectiveness of agalsidase alfa enzyme replacement in Fabry disease: A Fabry Outcome Survey analysis. *Molecular Genetics and Metabolism Reports*, **2015**, 3, 21-7	1.8	77
399	Investigation of lysosomal storage diseases in nonimmune hydrops fetalis. *Prenatal Diagnosis*, **2004** , 24, 653-7	3.2	76
398	Development of a bile acid-based newborn screen for Niemann-Pick disease type C. *Science Translational Medicine*, **2016**, 8, 337ra63	17.5	75
397	Genomic rearrangements in BRCA1 and BRCA2: A literature review. *Genetics and Molecular Biology*, **2009**, 32, 437-46	2	72
396	Therapies for the bone in mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2015**, 114, 94-109	3.7	70
395	Idursulfase treatment of Hunter syndrome in children younger than 6 years: results from the Hunter Outcome Survey. *Genetics in Medicine*, **2011**, 13, 102-9	8.1	69
394	Newborn screening for congenital infectious diseases. *Emerging Infectious Diseases*, **2004**, 10, 1068-73	10.2	69
393	Diagnosis and treatment trends in mucopolysaccharidosis I: findings from the MPS I Registry. *European Journal of Pediatrics*, **2012**, 171, 911-9	4.1	67
392	Identification and characterization of 13 new mutations in mucopolysaccharidosis type I patients. *Molecular Genetics and Metabolism*, **2003**, 78, 37-43	3.7	67
391	The role of enzyme replacement therapy in severe Hunter syndrome-an expert panel consensus. *European Journal of Pediatrics*, **2012**, 171, 181-8	4.1	66
390	Diagnosing mucopolysaccharidosis IVA. *Journal of Inherited Metabolic Disease*, **2013**, 36, 293-307	5.4	66
389	Enzyme replacement therapy for mucopolysaccharidosis VI: evaluation of long-term pulmonary function in patients treated with recombinant human N-acetylgalactosamine 4-sulfatase. *Journal of Inherited Metabolic Disease*, **2010**, 33, 51-60	5.4	66

388	Development and testing of new screening method for keratan sulfate in mucopolysaccharidosis IVA. *Pediatric Research*, **2004**, 55, 592-7	3.2	66
387	Keratan sulphate levels in mucopolysaccharidoses and mucolipidoses. *Journal of Inherited Metabolic Disease*, **2005**, 28, 187-202	5.4	65
386	Natural history and galsulfase treatment in mucopolysaccharidosis VI (MPS VI, Maroteaux-Lamy syndrome)--10-year follow-up of patients who previously participated in an MPS VI Survey Study. *American Journal of Medical Genetics, Part A*, **2014**, 164A, 1953-64	2.5	63
385	Neurocognitive and somatic stabilization in pediatric patients with severe Mucopolysaccharidosis Type I after 52 weeks of intravenous brain-penetrating insulin receptor antibody-iduronidase fusion protein (valanafusp alpha): an open label phase 1-2 trial. *Orphanet Journal of Rare Diseases*, **2018**, 13, 110	4.2	61
384	Selective screening of 10,000 high-risk Brazilian patients for the detection of inborn errors of metabolism. *European Journal of Pediatrics*, **1997**, 156, 650-4	4.1	61
383	A Phase 2 study of migalastat hydrochloride in females with Fabry disease: selection of population, safety and pharmacodynamic effects. *Molecular Genetics and Metabolism*, **2013**, 109, 86-92	3.7	60
382	The role of methylenetetrahydrofolate reductase in acute lymphoblastic leukemia in a Brazilian mixed population. *Leukemia Research*, **2006**, 30, 477-81	2.7	60
381	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, 229-40	5.4	59
380	Allelic heterogeneity in the COH1 gene explains clinical variability in Cohen syndrome. *American Journal of Human Genetics*, **2004**, 75, 138-45	11	59
379	Correlation of MR imaging and MR spectroscopy findings with cognitive impairment in mucopolysaccharidosis II. *American Journal of Neuroradiology*, **2007**, 28, 1029-33	4.4	58
378	Multi-domain impact of elosufase alfa in Morquio A syndrome in the pivotal phase III trial. *Molecular Genetics and Metabolism*, **2015**, 114, 178-85	3.7	57
377	A systematic review of the prevalence of Morquio A syndrome: challenges for study reporting in rare diseases. *Orphanet Journal of Rare Diseases*, **2014**, 9, 173	4.2	57
376	Heparan sulfate levels in mucopolysaccharidoses and mucolipidoses. *Journal of Inherited Metabolic Disease*, **2005**, 28, 743-57	5.4	56
375	Serum S100B protein is increased in fasting rats. *Archives of Medical Research*, **2006**, 37, 683-6	6.6	55
374	A dose-optimization trial of laronidase (Aldurazyme) in patients with mucopolysaccharidosis I. *Molecular Genetics and Metabolism*, **2009**, 96, 13-9	3.7	54
373	Hematopoietic Stem Cell Transplantation for Patients with Mucopolysaccharidosis II. *Biology of Blood and Marrow Transplantation*, **2017**, 23, 1795-1803	4.7	53
372	Investigation of oxidative stress parameters in treated phenylketonuric patients. *Metabolic Brain Disease*, **2006**, 21, 287-96	3.9	53
371	Enzyme replacement therapy started at birth improves outcome in difficult-to-treat organs in mucopolysaccharidosis I mice. *Molecular Genetics and Metabolism*, **2013**, 109, 33-40	3.7	52

370	Newborn screening for mucopolysaccharidoses: a pilot study of measurement of glycosaminoglycans by tandem mass spectrometry. *Journal of Inherited Metabolic Disease*, **2017**, 40, 151-158	5.4	52
369	Impact of enzyme replacement therapy and hematopoietic stem cell transplantation in patients with Morquio A syndrome. *Drug Design, Development and Therapy*, **2015**, 9, 1937-53	4.4	52
368	Importance of surgical history in diagnosing mucopolysaccharidosis type II (Hunter syndrome): data from the Hunter Outcome Survey. *Genetics in Medicine*, **2010**, 12, 816-22	8.1	52
367	Understanding the natural history of Gaucher disease. *American Journal of Hematology*, **2015**, 90 Suppl 1, S6-11	7.1	51
366	Enzyme replacement therapy for mucopolysaccharidosis VI: long-term cardiac effects of galsulfase (Naglazyme®) therapy. *Journal of Inherited Metabolic Disease*, **2013**, 36, 385-94	5.4	51
365	Diagnosis of neuronal ceroid lipofuscinosis type 2 (CLN2 disease): Expert recommendations for early detection and laboratory diagnosis. *Molecular Genetics and Metabolism*, **2016**, 119, 160-7	3.7	50
364	Expression of the disease on female carriers of X-linked lysosomal disorders: a brief review. *Orphanet Journal of Rare Diseases*, **2010**, 5, 14	4.2	50
363	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	1.4	50
362	Intrathecal administration of recombinant human N-acetylgalactosamine 4-sulfatase to a MPS VI patient with pachymeningitis cervicalis. *Molecular Genetics and Metabolism*, **2010**, 99, 346-50	3.7	49
361	Biochemical characterization of chitotriosidase enzyme: comparison between normal individuals and patients with Gaucher and with Niemann-Pick diseases. *Clinical Biochemistry*, **2004**, 37, 893-7	3.5	49
360	Development and validation of a simple questionnaire for the identification of hereditary breast cancer in primary care. *BMC Cancer*, **2009**, 9, 283	4.8	48
359	Glycosaminoglycans analysis in blood and urine of patients with mucopolysaccharidosis. *Molecular Genetics and Metabolism*, **2018**, 125, 44-52	3.7	47
358	Diagnosing Hunter syndrome in pediatric practice: practical considerations and common pitfalls. *European Journal of Pediatrics*, **2012**, 171, 631-9	4.1	46
357	Orthopedic manifestations in patients with mucopolysaccharidosis type II (Hunter syndrome) enrolled in the Hunter Outcome Survey. *Orthopedic Reviews*, **2010**, 2, e16	1.2	46
356	Pregnancy outcome after exposure to misoprostol in Brazil: a prospective, controlled study. *Reproductive Toxicology*, **1999**, 13, 147-51	3.4	46
355	Establishment of glycosaminoglycan assays for mucopolysaccharidoses. *Metabolites*, **2014**, 4, 655-79	5.6	45
354	Incidence of cystic fibrosis in five different states of Brazil as determined by screening of p.F508del, mutation at the CFTR gene in newborns and patients. *Journal of Cystic Fibrosis*, **2008**, 7, 15-22	4.1	45
353	Mucopolysaccharidoses in Brazil: what happens from birth to biochemical diagnosis?. *American Journal of Medical Genetics, Part A*, **2008**, 146A, 1741-7	2.5	45

352	Mucopolysacccharidoses: From understanding to treatment, a century of discoveries. *Genetics and Molecular Biology*, **2012**, 35, 924-31	2	44
351	The effect of idursulfase on growth in patients with Hunter syndrome: data from the Hunter Outcome Survey (HOS). *Molecular Genetics and Metabolism*, **2013**, 109, 41-8	3.7	44
350	Design, baseline characteristics, and early findings of the MPS VI (mucopolysaccharidosis VI) Clinical Surveillance Program (CSP). *Journal of Inherited Metabolic Disease*, **2013**, 36, 373-84	5.4	43
349	Newborn Screening for Pompe Disease. *Pediatrics*, **2017**, 140, S4-S13	7.4	43
348	Long-term endurance and safety of elosulfase alfa enzyme replacement therapy in patients with Morquio A syndrome. *Molecular Genetics and Metabolism*, **2016**, 119, 131-43	3.7	42
347	Consensus recommendation for a diagnostic guideline for acid sphingomyelinase deficiency. *Genetics in Medicine*, **2017**, 19, 967-974	8.1	41
346	Treatment of brain disease in the mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2017** , 122S, 25-34	3.7	41
345	LC-MS/MS based assay and reference intervals in children and adolescents for oxysterols elevated in Niemann-Pick diseases. *Clinical Biochemistry*, **2015**, 48, 596-602	3.5	41
344	Selective screening for organic acidemias by urine organic acid GC-MS analysis in Brazil: fifteen-year experience. *Clinica Chimica Acta*, **2009**, 400, 77-81	6.2	40
343	Long-term outcomes of systemic therapies for Hurler syndrome: an international multicenter comparison. *Genetics in Medicine*, **2018**, 20, 1423-1429	8.1	39
342	Response of women with Fabry disease to enzyme replacement therapy: comparison with men, using data from FOS--the Fabry Outcome Survey. *Molecular Genetics and Metabolism*, **2011**, 103, 207-14	3.7	39
341	Cause of death in patients with chronic visceral and chronic neurovisceral acid sphingomyelinase deficiency (Niemann-Pick disease type B and B variant): Literature review and report of new cases. *Molecular Genetics and Metabolism*, **2016**, 118, 206-213	3.7	38
340	Emerging drugs for the treatment of mucopolysaccharidoses. *Expert Opinion on Emerging Drugs*, **2016**, 21, 9-26	3.7	38
339	Practical and reliable enzyme test for the detection of mucopolysaccharidosis IVA (Morquio Syndrome type A) in dried blood samples. *Clinica Chimica Acta*, **2011**, 412, 1805-8	6.2	38
338	Identification of a common mutation in mucopolysaccharidosis IVA: correlation among genotype, phenotype, and keratan sulfate. *Journal of Human Genetics*, **2004**, 49, 490-494	4.3	38
337	Six novel beta-galactosidase gene mutations in Brazilian patients with GM1-gangliosidosis. *Human Mutation*, **1999**, 13, 401-9	4.7	38
336	Ten years of the Hunter Outcome Survey (HOS): insights, achievements, and lessons learned from a global patient registry. *Orphanet Journal of Rare Diseases*, **2017**, 12, 82	4.2	37
335	Recommendations for the management of MPS IVA: systematic evidence- and consensus-based guidance. *Orphanet Journal of Rare Diseases*, **2019**, 14, 137	4.2	36

334	Long-term experience with enzyme replacement therapy (ERT) in MPS II patients with a severe phenotype: an international case series. *Journal of Inherited Metabolic Disease*, **2014**, 37, 823-9	5.4	36
333	In vivo genome editing of mucopolysaccharidosis I mice using the CRISPR/Cas9 system. *Journal of Controlled Release*, **2018**, 288, 23-33	11.7	36
332	Efficacy of the pharmacologic chaperone migalastat in a subset of male patients with the classic phenotype of Fabry disease and migalastat-amenable variants: data from the phase 3 randomized, multicenter, double-blind clinical trial and extension study. *Genetics in Medicine*, **2019**, 21, 1987-1997	8.1	34
331	Oxidative stress and inflammation in mucopolysaccharidosis type IVA patients treated with enzyme replacement therapy. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2015**, 1852, 1012-9	6.9	34
330	Enzyme Replacement Therapy in Mucopolysaccharidosis II Patients Under 1 Year of Age. *JIMD Reports*, **2014**, 14, 99-113	1.9	33
329	A multinational, multidisciplinary consensus for the diagnosis and management of spinal cord compression among patients with mucopolysaccharidosis VI. *Molecular Genetics and Metabolism*, **2012**, 107, 15-24	3.7	33
328	Migalastat HCl reduces globotriaosylsphingosine (lyso-Gb3) in Fabry transgenic mice and in the plasma of Fabry patients. *PLoS ONE*, **2013**, 8, e57631	3.7	33
327	Genotype-phenotype relationships in mucopolysaccharidosis type I (MPS I): Insights from the International MPS I Registry. *Clinical Genetics*, **2019**, 96, 281-289	4	31
326	Genetic studies in a cluster of mucopolysaccharidosis type VI patients in Northeast Brazil. *Molecular Genetics and Metabolism*, **2011**, 104, 603-7	3.7	31
325	Metabolic effects and the methylenetetrahydrofolate reductase (MTHFR) polymorphism associated with neural tube defects in southern Brazil. *Birth Defects Research Part A: Clinical and Molecular Teratology*, **2004**, 70, 459-63		31
324	Glycosaminoglycan levels in dried blood spots of patients with mucopolysaccharidoses and mucolipidoses. *Molecular Genetics and Metabolism*, **2017**, 120, 247-254	3.7	30
323	Clinical outcomes in idursulfase-treated patients with mucopolysaccharidosis type II: 3-year data from the hunter outcome survey (HOS). *Orphanet Journal of Rare Diseases*, **2017**, 12, 161	4.2	30
322	Chondroitin 6-Sulfate as a Novel Biomarker for Mucopolysaccharidosis IVA and VII. *JIMD Reports*, **2014**, 16, 15-24	1.9	30
321	Non-immune hydrops fetalis: A prospective study of 53 cases. *American Journal of Medical Genetics, Part A*, **2013**, 161A, 3078-86	2.5	30
320	Oxidative stress in patients with mucopolysaccharidosis type II before and during enzyme replacement therapy. *Molecular Genetics and Metabolism*, **2011**, 103, 121-7	3.7	30
319	White matter lesions in Fabry disease before and after enzyme replacement therapy: a 2-year follow-up. *Arquivos De Neuro-Psiquiatria*, **2006**, 64, 711-7	1.6	30
318	Lysosomal diseases: Overview on current diagnosis and treatment. *Genetics and Molecular Biology*, **2019**, 42, 165-177	2	29
317	Longitudinal analysis of endurance and respiratory function from a natural history study of Morquio A syndrome. *Molecular Genetics and Metabolism*, **2015**, 114, 186-94	3.7	29

316	Guidelines for diagnosis and treatment of Hunter Syndrome for clinicians in Latin America. *Genetics and Molecular Biology*, **2014**, 37, 315-29	2	29
315	Di-sulfated Keratan Sulfate as a Novel Biomarker for Mucopolysaccharidosis II, IVA, and IVB. *JIMD Reports*, **2015**, 21, 1-13	1.9	29
314	Genetic causes of intellectual disability in a birth cohort: a population-based study. *American Journal of Medical Genetics, Part A*, **2015**, 167, 1204-14	2.5	28
313	Early hematopoietic stem cell transplantation in a patient with severe mucopolysaccharidosis II: A 7´years follow-up. *Molecular Genetics and Metabolism Reports*, **2017**, 12, 62-68	1.8	28
312	Prevalence of the BRCA1 founder mutation c.5266dupin Brazilian individuals at-risk for the hereditary breast and ovarian cancer syndrome. *Hereditary Cancer in Clinical Practice*, **2011**, 9, 12	2.3	28
311	Induction of lipid peroxidation and decrease of antioxidant defenses in symptomatic and asymptomatic patients with X-linked adrenoleukodystrophy. *International Journal of Developmental Neuroscience*, **2007**, 25, 441-4	2.7	28
310	Protracted course of Krabbe disease in an adult patient bearing a novel mutation. *Archives of Neurology*, **1999**, 56, 1014-7		28
309	Laronidase-functionalized multiple-wall lipid-core nanocapsules: promising formulation for a more effective treatment of mucopolysaccharidosis type I. *Pharmaceutical Research*, **2015**, 32, 941-54	4.5	27
308	Neurological manifestations of lysosomal disorders and emerging therapies targeting the CNS. *The Lancet Child and Adolescent Health*, **2018**, 2, 56-68	14.5	27
307	Novel heparan sulfate assay by using automated high-throughput mass spectrometry: Application to monitoring and screening for mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2014**, 113, 92-9	3.7	27
306	Sleep abnormalities in untreated patients with mucopolysaccharidosis type VI. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 1546-51	2.5	27
305	The natural history of growth in patients with Hunter syndrome: Data from the Hunter Outcome Survey (HOS). *Molecular Genetics and Metabolism*, **2016**, 117, 438-46	3.7	27
304	Relative frequency and estimated minimal frequency of Lysosomal Storage Diseases in Brazil: Report from a Reference Laboratory. *Genetics and Molecular Biology*, **2017**, 40, 31-39	2	26
303	A multicenter, open-label study evaluating safety and clinical outcomes in children (1.4-7.5 years) with Hunter syndrome receiving idursulfase enzyme replacement therapy. *Genetics in Medicine*, **2014**, 16, 435-41	8.1	26
302	Evidence of a progressive motor dysfunction in Mucopolysaccharidosis type I mice. *Behavioural Brain Research*, **2012**, 233, 169-75	3.4	26
301	Newborn screening for lysosomal diseases: current status and potential interface with population medical genetics in Latin America. *Journal of Inherited Metabolic Disease*, **2012**, 35, 871-7	5.4	25
300	Comparison between the biochemical properties of plasma chitotriosidase from normal individuals and from patients with Gaucher disease, GM1-gangliosidosis, Krabbe disease and heterozygotes for Gaucher disease. *Clinical Biochemistry*, **2007**, 40, 365-9	3.5	25
299	CFTR gene: molecular analysis in patients from South Brazil. *Molecular Genetics and Metabolism*, **2003**, 78, 259-64	3.7	25

298	Gene editing of MPS I human fibroblasts by co-delivery of a CRISPR/Cas9 plasmid and a donor oligonucleotide using nanoemulsions as nonviral carriers. *European Journal of Pharmaceutics and Biopharmaceutics*, **2018**, 122, 158-166	5.7	25
297	The Brazilian consensus on the management of Pompe disease. *Journal of Pediatrics*, **2009**, 155, S47-56	3.6	24
296	Cognitive endpoints for therapy development for neuronopathic mucopolysaccharidoses: Results of a consensus procedure. *Molecular Genetics and Metabolism*, **2017**, 121, 70-79	3.7	23
295	Investigation of newborns with abnormal results in a newborn screening program for four lysosomal storage diseases in Brazil. *Molecular Genetics and Metabolism Reports*, **2017**, 12, 92-97	1.8	23
294	Minimal clinically important difference for the 6-min walk test: literature review and application to Morquio A syndrome. *Orphanet Journal of Rare Diseases*, **2017**, 12, 78	4.2	23
293	A community-based study of mucopolysaccharidosis type VI in Brazil: the influence of founder effect, endogamy and consanguinity. *Human Heredity*, **2014**, 77, 189-96	1.1	23
292	Biomarkers for the mucopolysaccharidoses: discovery and clinical utility. *Molecular Genetics and Metabolism*, **2012**, 106, 395-402	3.7	23
291	Fabry disease in hemodialysis patients in southern Brazil: prevalence study and clinical report. *Renal Failure*, **2008**, 30, 825-30	2.9	23
290	Molecular characterization of phenylketonuria in South Brazil. *Molecular Genetics and Metabolism*, **2003**, 79, 17-24	3.7	23
289	Recommendations for clinical monitoring of patients with acid sphingomyelinase deficiency (ASMD). *Molecular Genetics and Metabolism*, **2019**, 126, 98-105	3.7	23
288	Pathogenesis of lumbar spine disease in mucopolysaccharidosis VII. *Molecular Genetics and Metabolism*, **2012**, 107, 153-60	3.7	22
287	Recombinant encapsulated cells overexpressing alpha-L-iduronidase correct enzyme deficiency in human mucopolysaccharidosis type I cells. *Cells Tissues Organs*, **2012**, 195, 323-9	2.1	22
286	Genotypic and phenotypic characterization of Brazilian patients with GM1 gangliosidosis. *Gene*, **2013**, 512, 113-6	3.8	21
285	Amino acids levels and lipid peroxidation in maple syrup urine disease patients. *Clinical Biochemistry* , **2009**, 42, 462-6	3.5	21
284	The effect of Lorenzo's oil on oxidative stress in X-linked adrenoleukodystrophy. *Journal of the Neurological Sciences*, **2006**, 247, 157-64	3.2	21
283	Nerve conduction studies, electromyography and sympathetic skin response in Fabry's disease. *Journal of the Neurological Sciences*, **2003**, 214, 21-5	3.2	21
282	Triagem neonatal para hemoglobinopatias: um estudo piloto em Porto Alegre, Rio Grande do Sul, Brasil. *Cadernos De Saude Publica*, **2002**, 18, 833-841	3.2	21
281	Services for the prevention and management of genetic disorders and birth defects in developing countries. *Public Health Genomics*, **1999**, 2, 196-201	1.9	21

280	Biomolecules damage and redox status abnormalities in Fabry patients before and during enzyme replacement therapy. *Clinica Chimica Acta*, **2016**, 461, 41-6	6.2	21
279	DNA damage in Fabry patients: An investigation of oxidative damage and repair. *Mutation Research - Genetic Toxicology and Environmental Mutagenesis*, **2015**, 784-785, 31-6	3	20
278	Plasma Pharmacokinetics of Valanafusp Alpha, a Human Insulin Receptor Antibody-Iduronidase Fusion Protein, in Patients with Mucopolysaccharidosis Type I. *BioDrugs*, **2018**, 32, 169-176	7.9	20
277	Cathepsin B inhibition attenuates cardiovascular pathology in mucopolysaccharidosis I mice. *Life Sciences*, **2018**, 196, 102-109	6.8	20
276	Intellectual Disability in a Birth Cohort: Prevalence, Etiology, and Determinants at the Age of 4 Years. *Public Health Genomics*, **2016**, 19, 290-7	1.9	20
275	Functional capacity evaluation of patients with mucopolysaccharidosis. *Journal of Pediatric Rehabilitation Medicine*, **2012**, 5, 37-46	1.4	20
274	Hexacosanoic and docosanoic acids plasma levels in patients with cerebral childhood and asymptomatic X-linked adrenoleukodystrophy: Lorenzo's oil effect. *Metabolic Brain Disease*, **2008**, 23, 43-9	3.9	20
273	Maple syrup urine disease in treated patients: biochemical and oxidative stress profiles. *Clinical Biochemistry*, **2008**, 41, 317-24	3.5	20
272	Adult derived mononuclear bone marrow cells improve survival in a model of acetaminophen-induced acute liver failure in rats. *Toxicology*, **2008**, 247, 1-5	4.4	20
271	Diagnosis of Mucopolysaccharidoses. *Diagnostics*, **2020**, 10,	3.8	20
270	Interleukin-15 favors the expansion of central memory CD8+ T cells in ex vivo generated, antileukemia human cytotoxic T lymphocyte lines. *Journal of Immunotherapy*, **2008**, 31, 385-93	5	19
269	Fibroblasts of skin fragments as a tool for the investigation of genetic diseases: technical recommendations. *Genetics and Molecular Biology*, **2000**, 23, 269-271	2	19
268	Recommendations for the management of MPS VI: systematic evidence- and consensus-based guidance. *Orphanet Journal of Rare Diseases*, **2019**, 14, 118	4.2	18
267	Genome Editing: Potential Treatment for Lysosomal Storage Diseases. *Current Stem Cell Reports*, **2015**, 1, 9-15	1.8	18
266	Impact of long-term elosulfase alfa on activities of daily living in patients with Morquio A syndrome in an open-label, multi-center, phase 3 extension study. *Molecular Genetics and Metabolism*, **2018**, 123, 127-134	3.7	18
265	Characterization of individuals at high risk of developing melanoma in Latin America: bases for genetic counseling in melanoma. *Genetics in Medicine*, **2016**, 18, 727-36	8.1	18
264	Retroviral-vector-mediated gene therapy to mucopolysaccharidosis I mice improves sensorimotor impairments and other behavioral deficits. *Journal of Inherited Metabolic Disease*, **2013**, 36, 499-512	5.4	18
263	Diagnostic and treatment strategies in mucopolysaccharidosis VI. *The Application of Clinical Genetics*, **2015**, 8, 245-55	3.1	18

262	Brain magnetic resonance imaging findings in patients with mucopolysaccharidosis VI. *Journal of Inherited Metabolic Disease*, **2013**, 36, 357-62	5.4	18
261	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, 206-10	3	18
260	Oxidative stress is induced in female carriers of X-linked adrenoleukodystrophy. *Journal of the Neurological Sciences*, **2008**, 266, 79-83	3.2	18
259	Clinical characterization and risk profile of individuals seeking genetic counseling for hereditary breast cancer in Brazil. *Journal of Genetic Counseling*, **2007**, 16, 363-71	2.5	18
258	In vitro correction of ARSA deficiency in human skin fibroblasts from metachromatic leukodystrophy patients after treatment with microencapsulated recombinant cells. *Metabolic Brain Disease*, **2008**, 23, 469-84	3.9	18
257	Some observations on genetic factors in urolithiasis. *Urology*, **1981**, 17, 33-8	1.6	18
256	Impact of long-term elosulfase alfa treatment on respiratory function in patients with Morquio A syndrome. *Journal of Inherited Metabolic Disease*, **2016**, 39, 839-847	5.4	18
255	The laboratory diagnosis of mucopolysaccharidosis III (Sanfilippo syndrome): A changing landscape. *Molecular Genetics and Metabolism*, **2014**, 113, 34-41	3.7	17
254	Fabry disease: a new approach for the screening of females in high-risk groups. *Clinical Biochemistry* , **2014**, 47, 657-62	3.5	17
253	Enzyme replacement therapy for Mucopolysaccharidosis Type I among patients followed within the MPS Brazil Network. *Genetics and Molecular Biology*, **2014**, 37, 23-9	2	17
252	GSTM1, GSTT1, and GSTP1 polymorphisms, breast cancer risk factors and mammographic density in women submitted to breast cancer screening. *Revista Brasileira De Epidemiologia*, **2012**, 15, 246-55	1.3	17
251	Age-dependent availability and functionality of bone marrow stem cells in an experimental model of acute and chronic myocardial infarction. *Cell Transplantation*, **2011**, 20, 407-19	4	17
250	Recent Advances in Treatment Approaches of Mucopolysaccharidosis VI. *Current Pharmaceutical Biotechnology*, **2011**, 12, 956-62	2.6	17
249	Consistency of self-reported first-degree family history of cancer in a population-based study. *Familial Cancer*, **2009**, 8, 195-202	3	17
248	S100B content and SOD activity in amniotic fluid of pregnancies with Down syndrome. *Clinical Biochemistry*, **2004**, 37, 134-7	3.5	17
247	Identification of a novel mutation in the ARSB gene that is frequent among Brazilian MPSVI patients. *Genetic Testing and Molecular Biomarkers*, **2003**, 7, 347-9		17
246	Evaluation of glucose-6-phosphate dehydrogenase stability in blood samples under different collection and storage conditions. *Clinical Chemistry*, **2005**, 51, 1080-1	5.5	17
245	Clinical and laboratorial study of 19 cases of mucopolysaccharidoses. *Revista Do Hospital Das Clinicas*, **2000**, 55, 213-8		17

244	Safety of intrathecal delivery of recombinant human arylsulfatase A in children with metachromatic leukodystrophy: Results from a phase 1/2 clinical trial. *Molecular Genetics and Metabolism*, **2020**, 131, 235-244	3.7	17
243	Molecular and biochemical biomarkers for diagnosis and therapy monitorization of Niemann-Pick type C patients. *International Journal of Developmental Neuroscience*, **2018**, 66, 18-23	2.7	17
242	Oxidative and nitrative stress and pro-inflammatory cytokines in Mucopolysaccharidosis type II patients: effect of long-term enzyme replacement therapy and relation with glycosaminoglycan accumulation. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2016**, 1862, 1608-16	6.9	16
241	Migalastat improves diarrhea in patients with Fabry disease: clinical-biomarker correlations from the phase 3 FACETS trial. *Orphanet Journal of Rare Diseases*, **2018**, 13, 68	4.2	16
240	Characterization of joint disease in mucopolysaccharidosis type I mice. *International Journal of Experimental Pathology*, **2013**, 94, 305-11	2.8	16
239	The CDKN2A p.A148T variant is associated with cutaneous melanoma in Southern Brazil. *Experimental Dermatology*, **2011**, 20, 890-3	4	16
238	UGT1A1, SLCO1B1, and SLCO1B3 polymorphisms vs. neonatal hyperbilirubinemia: is there an association?. *Pediatric Research*, **2012**, 72, 169-73	3.2	16
237	New GLB1 mutation in siblings with Morquio type B disease presenting with mental regression. *Molecular Genetics and Metabolism*, **2009**, 96, 148	3.7	16
236	Population prevalence of hereditary breast cancer phenotypes and implementation of a genetic cancer risk assessment program in southern Brazil. *Genetics and Molecular Biology*, **2009**, 32, 447-55	2	16
235	Ontogenetic changes in serum S100B in Down syndrome patients. *Clinical Biochemistry*, **2005**, 38, 433-5	3.5	16
234	GM1 gangliosidosis: clinical and laboratory findings in eight families. *Human Genetics*, **1985**, 70, 347-54	6.3	16
233	Mucopolysaccharidosis Type I. *Diagnostics*, **2020**, 10,	3.8	15
232	Shotgun proteomics reveals possible mechanisms for cognitive impairment in Mucopolysaccharidosis I mice. *Molecular Genetics and Metabolism*, **2015**, 114, 138-45	3.7	15
231	Intraperitoneal implant of recombinant encapsulated cells overexpressing alpha-L-iduronidase partially corrects visceral pathology in mucopolysaccharidosis type I mice. *Cytotherapy*, **2012**, 14, 860-7	4.8	15
230	Severe phenotype in MPS II patients associated with a large deletion including contiguous genes. *American Journal of Medical Genetics, Part A*, **2012**, 158A, 1055-9	2.5	15
229	Placenta analysis of prenatally diagnosed patients reveals early GAG storage in mucopolysaccharidoses II and VI. *Molecular Genetics and Metabolism*, **2011**, 103, 197-8	3.7	15
228	Population analysis of the GLB1 gene in South Brazil. *Genetics and Molecular Biology*, **2011**, 34, 45-8	2	15
227	Cell microencapsulation: a potential tool for the treatment of neuronopathic lysosomal storage diseases. *Journal of Inherited Metabolic Disease*, **2011**, 34, 983-90	5.4	15

226	X-linked adrenoleukodystrophy: clinical course and minimal incidence in South Brazil. *Brain and Development*, **2010**, 32, 180-90	2.2	15
225	Molecular analysis of the Pi*Z allele in patients with liver disease. *American Journal of Medical Genetics Part A*, **2001**, 104, 287-90		15
224	Screening for germline BRCA1, BRCA2, TP53 and CHEK2 mutations in families at-risk for hereditary breast cancer identified in a population-based study from Southern Brazil. *Genetics and Molecular Biology*, **2016**, 39, 210-22	2	15
223	Oxidative damage and redox in Lysosomal Storage Disorders: Biochemical markers. *Clinica Chimica Acta*, **2017**, 466, 46-53	6.2	14
222	Effects of enzyme replacement therapy started late in a murine model of mucopolysaccharidosis type I. *PLoS ONE*, **2015**, 10, e0117271	3.7	14
221	Microarray-based comparative genomic hybridization analysis in neonates with congenital anomalies: detection of chromosomal imbalances. *Jornal De Pediatria*, **2015**, 91, 59-67	2.6	14
220	A useful multi-analyte blood test for cerebrotendinous xanthomatosis. *Clinical Biochemistry*, **2014**, 47, 860-3	3.5	14
219	Clinical and biochemical study of 29 Brazilian patients with metachromatic leukodystrophy. *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S257-62	5.4	14
218	Novel mutations in the glucocerebrosidase gene of brazilian patients with Gaucher disease. *JIMD Reports*, **2013**, 9, 7-16	1.9	14
217	Effects of cryopreservation and hypothermic storage on cell viability and enzyme activity in recombinant encapsulated cells overexpressing alpha-L-iduronidase. *Artificial Organs*, **2010**, 34, 434-9	2.6	14
216	Biochemical study on beta-glucosidase in individuals with Gaucher disease and normal subjects. *Clinica Chimica Acta*, **2004**, 343, 145-53	6.2	14
215	Application of N-palmitoyl-O-phosphocholineserine for diagnosis and assessment of response to treatment in Niemann-Pick type C disease. *Molecular Genetics and Metabolism*, **2020**, 129, 292-302	3.7	14
214	Impairment of Body Growth in Mucopolysaccharidoses **2012**, 2091-2117		14
213	Elevation of glycosaminoglycans in the amniotic fluid of a fetus with mucopolysaccharidosis VII. *Prenatal Diagnosis*, **2017**, 37, 435-439	3.2	13
212	Phase I and II clinical trials for the mucopolysaccharidoses. *Expert Opinion on Investigational Drugs*, **2017**, 26, 1331-1340	5.9	13
211	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	2	13
210	A simple protocol for transfecting human mesenchymal stem cells. *Biotechnology Letters*, **2018**, 40, 617-622		13
209	Burden of Illness in Acid Sphingomyelinase Deficiency: A Retrospective Chart Review of 100 Patients. *JIMD Reports*, **2018**, 41, 119-129	1.9	13

208	Anthropometry, nutritional status, and dietary intake in pediatric patients with osteogenesis imperfecta. *Journal of the American College of Nutrition*, **2014**, 33, 18-25	3.5	13
207	Pathogenesis of mitral valve disease in mucopolysaccharidosis VII dogs. *Molecular Genetics and Metabolism*, **2013**, 110, 319-28	3.7	13
206	Selective screening for lysosomal storage diseases with dried blood spots collected on filter paper in 4,700 high-risk colombian subjects. *JIMD Reports*, **2013**, 11, 107-16	1.9	13
205	Kidney function and 24-hour proteinuria in patients with Fabry disease during 36 months of agalsidase alfa enzyme replacement therapy: a Brazilian experience. *Renal Failure*, **2009**, 31, 773-8	2.9	13
204	Prevalence of G6PD deficiency in newborns in the south of Brazil. *Journal of Medical Screening*, **2006**, 13, 85-6	1.4	13
203	Unique frequency of known mutations in Brazilian MPS I patients. *American Journal of Medical Genetics Part A*, **2000**, 90, 108-9		13
202	Heterozygous cystinuria and urinary lithiasis. *American Journal of Medical Genetics Part A*, **1985**, 22, 703-15		13
201	Chloramphenicol enhances IDUA activity on fibroblasts from mucopolysaccharidosis I patients. *Current Pharmaceutical Biotechnology*, **2013**, 14, 194-8	2.6	13
200	Aortic root dilatation in patients with mucopolysaccharidoses and the impact of enzyme replacement therapy. *Heart and Vessels*, **2019**, 34, 290-295	2.1	13
199	Hydrocephalus and mucopolysaccharidoses: what do we know and what do we not know?. *Childls Nervous System*, **2017**, 33, 1073-1080	1.7	12
198	Gene delivery strategies for the treatment of mucopolysaccharidoses. *Expert Opinion on Drug Delivery*, **2014**, 11, 449-59	8	12
197	Clinical and molecular characterization of patients at risk for hereditary melanoma in southern Brazil. *Journal of Investigative Dermatology*, **2008**, 128, 421-5	4.3	12
196	An evaluation of four methods for the detection of heterozygous cystinuria. *Clinica Chimica Acta*, **1987**, 164, 227-33	6.2	12
195	Mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome): assessment of joint mobility and grip and pinch strength. *Jornal De Pediatria*, **2008**, 84, 130-5	2.6	12
194	Estimated birth prevalence of mucopolysaccharidoses in Brazil. *American Journal of Medical Genetics, Part A*, **2020**, 182, 469-483	2.5	12
193	Long-Term Galsulfase Treatment Associated With Improved Survival of Patients With Mucopolysaccharidosis VI (Maroteaux-Lamy Syndrome): 15-Year Follow-Up From the Survey Study. *FIRE Forum for International Research in Education*, **2018**, 6, 232640981875580	1.4	12
192	Progressive heart disease in mucopolysaccharidosis type I mice may be mediated by increased cathepsin B activity. *Cardiovascular Pathology*, **2017**, 27, 45-50	3.8	11
191	Intrafamilial variability in the clinical manifestations of mucopolysaccharidosis type II: Data from the Hunter Outcome Survey (HOS). *American Journal of Medical Genetics, Part A*, **2018**, 176, 301-310	2.5	11

190	Neonatal screening for four lysosomal storage diseases with a digital microfluidics platform: Initial results in Brazil. *Genetics and Molecular Biology*, **2018**, 41, 414-416	2	11
189	Detection of organic acidemias in Brazil. *Archives of Medical Research*, **2002**, 33, 581-5	6.6	11
188	Characterization of beta-galactosidase in leukocytes and fibroblasts of GM1 gangliosidosis heterozygotes compared to normal subjects. *Clinical Biochemistry*, **2000**, 33, 125-9	3.5	11
187	Neonatal nonviral gene editing with the CRISPR/Cas9 system improves some cardiovascular, respiratory, and bone disease features of the mucopolysaccharidosis I phenotype in mice. *Gene Therapy*, **2020**, 27, 74-84	4	11
186	Rare disease landscape in Brazil: report of a successful experience in inborn errors of metabolism. *Orphanet Journal of Rare Diseases*, **2016**, 11, 76	4.2	11
185	Immune tolerance induction for laronidase treatment in mucopolysaccharidosis I. *Molecular Genetics and Metabolism Reports*, **2017**, 10, 61-66	1.8	10
184	Globotriaosylsphingosine induces oxidative DNA damage in cultured kidney cells. *Nephrology*, **2017**, 22, 490-493	2.2	10
183	Look carefully to the heels! A potentially treatable cause of spastic paraplegia. *Journal of Inherited Metabolic Disease*, **2015**, 38, 363-4	5.4	10
182	Intrathecal/Intracerebroventricular enzyme replacement therapy for the mucopolysaccharidoses: efficacy, safety, and prospects. *Expert Opinion on Orphan Drugs*, **2018**, 6, 403-411	1.1	10
181	Lysosomal enzymes may cross the blood-brain-barrier by pinocytosis: implications for enzyme replacement therapy. *Medical Hypotheses*, **2014**, 82, 478-80	3.8	10
180	Glucose-6-phosphate-dehydrogenase deficiency and its correlation with other risk factors in jaundiced newborns in Southern Brazil. *Asian Pacific Journal of Tropical Biomedicine*, **2011**, 1, 110-3	1.4	10
179	Bone marrow mononuclear cell transplantation improves survival and induces hepatocyte proliferation in rats after CCl(4) acute liver damage. *Digestive Diseases and Sciences*, **2010**, 55, 3384-92	4	10
178	Characterization of glycan substrates accumulating in GM1 Gangliosidosis. *Molecular Genetics and Metabolism Reports*, **2019**, 21, 100524	1.8	10
177	Subcutaneous implantation of microencapsulated cells overexpressing L-iduronidase for mucopolysaccharidosis type I treatment. *Journal of Materials Science: Materials in Medicine*, **2017**, 28, 43	4.5	9
176	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, e141-8	2	9
175	Oxidative profile exhibited by Mucopolysaccharidosis type IVA patients at diagnosis: Increased keratan urinary levels. *Molecular Genetics and Metabolism Reports*, **2017**, 11, 46-53	1.8	9
174	Treatment of MPS I mice with microencapsulated cells overexpressing IDUA: effect of the prednisolone administration. *Journal of Microencapsulation*, **2013**, 30, 383-9	3.4	9
173	G(M1)-ganglioside degradation and biosynthesis in human and murine G(M1)-gangliosidosis. *Clinica Chimica Acta*, **2005**, 354, 131-9	6.2	9

172	Biochemical properties of beta-glucosidase in leukocytes from patients and obligated heterozygotes for Gaucher disease carriers. *Clinica Chimica Acta*, **2005**, 362, 101-9	6.2	9
171	Serum S100B levels in patients with neural tube defects. *Clinica Chimica Acta*, **2006**, 364, 275-8	6.2	9
170	Triagem neonatal de distúrbios metabólicos. *Ciencia E Saude Coletiva*, **2002**, 7, 129-137	2.2	9
169	Mutation analysis of phenylketonuria in south Brazil. *Human Mutation*, **1996**, 8, 262-4	4.7	9
168	One-year results of a clinical trial of olipudase alfa enzyme replacement therapy in pediatric patients with acid sphingomyelinase deficiency. *Genetics in Medicine*, **2021**, 23, 1543-1550	8.1	9
167	Long-term outcomes with agalsidase alfa enzyme replacement therapy: Analysis using deconstructed composite events. *Molecular Genetics and Metabolism Reports*, **2018**, 14, 31-35	1.8	9
166	Niemann-Pick Disease Type C: Mutation Spectrum and Novel Sequence Variations in the Human NPC1 Gene. *Molecular Neurobiology*, **2019**, 56, 6426-6435	6.2	8
165	Brain imaging and genetic risk in the pediatric population, part 1: inherited metabolic diseases. *Neuroimaging Clinics of North America*, **2015**, 25, 31-51	3	8
164	A novel LC-MS/MS assay to quantify dermatan sulfate in cerebrospinal fluid as a biomarker for mucopolysaccharidosis II. *Bioanalysis*, **2018**, 10, 825-838	2.1	8
163	Polymorphic variants of UGT1A1 in neonatal jaundice in southern Brazil. *Journal of Tropical Pediatrics*, **2010**, 56, 366-7	1.2	8
162	Urinary oligosaccharides: a peripheral marker for Sida carpinifolia exposure or poisoning. *Toxicon*, **2009**, 53, 591-4	2.8	8
161	Inborn errors of metabolism in Latin America: challenges and opportunities. *Journal of Inherited Metabolic Disease*, **2010**, 33, S315-20	5.4	8
160	Cancer genetic counseling in public health care hospitals: the experience of three Brazilian services. *Public Health Genomics*, **2007**, 10, 110-9	1.9	8
159	Application of a comprehensive protocol for the identification of Gaucher disease in Brazil. *American Journal of Medical Genetics, Part A*, **2005**, 136, 58-62	2.5	8
158	Effects of imilglucerase withdrawal on an adult with Gaucher disease. *British Journal of Haematology*, **2001**, 113, 1089	4.5	8
157	Metabolic factors in urolithiasis: a study in Brazil. *Journal of Urology*, **1980**, 124, 503-7	2.5	8
156	Therapy development for the mucopolysaccharidoses: Updated consensus recommendations for neuropsychological endpoints. *Molecular Genetics and Metabolism*, **2020**, 131, 181-196	3.7	8
155	Recent advances in molecular testing to improve early diagnosis in children with mucopolysaccharidoses. *Expert Review of Molecular Diagnostics*, **2018**, 18, 855-866	3.8	8

154	Iduronate-2-sulfatase fused with anti-hTfR antibody, pabinafusp alfa, for MPS-II: A phase 2 trial in Brazil. *Molecular Therapy*, **2021**, 29, 2378-2386	11.7	8
153	Improvement in time to treatment, but not time to diagnosis, in patients with mucopolysaccharidosis type I. *Archives of Disease in Childhood*, **2021**, 106, 674-679	2.2	7
152	Current molecular genetics strategies for the diagnosis of lysosomal storage disorders. *Expert Review of Molecular Diagnostics*, **2016**, 16, 113-23	3.8	7
151	CRISPR-Cas9-mediated gene editing in human MPS I fibroblasts. *Gene*, **2018**, 678, 33-37	3.8	7
150	Are MPS II heterozygotes actually asymptomatic? A study based on clinical and biochemical data, X-inactivation analysis and imaging evaluations. *American Journal of Medical Genetics, Part A*, **2011**, 155A, 50-7	2.5	7
149	Mucopolissacaridose tipo VI (sÍndrome de Maroteaux-Lamy): avaliaÇÃo da mobilidade articular e das forÇas de garra e de pinÇa. *Jornal De Pediatria*, **2008**, 84,	2.6	7
148	Detection of mucopolysaccharidosis type I heterozygotes based on the biochemical characteristics of leukocyte alpha-L-iduronidase. *Archives of Medical Research*, **2002**, 33, 20-4	6.6	7
147	Neurological outcomes after hematopoietic stem cell transplantation for cerebral X-linked adrenoleukodystrophy, late onset metachromatic leukodystrophy and Hurler syndrome. *Arquivos De Neuro-Psiquiatria*, **2016**, 74, 953-966	1.6	7
146	Cardio- Renal Outcomes With Long- Term Agalsidase Alfa Enzyme Replacement Therapy: A 10- Year Fabry Outcome Survey (FOS) Analysis. *Drug Design, Development and Therapy*, **2019**, 13, 3705-3715	4.4	7
145	Information and Diagnosis Networks - tools to improve diagnosis and treatment for patients with rare genetic diseases. *Genetics and Molecular Biology*, **2019**, 42, 155-164	2	6
144	A consensus statement on birth defects surveillance, prevention, and care in Latin America and the Caribbean. *Revista Panamericana De Salud Publica/Pan American Journal of Public Health*, **2019**, 43, e2	4.1	6
143	Safety of switching to Migalastat from enzyme replacement therapy in Fabry disease: Experience from the Phase 3 ATTRACT study. *American Journal of Medical Genetics, Part A*, **2019**, 179, 1069-1073	2.5	6
142	Screening for Attenuated Forms of Mucopolysaccharidoses in Patients with Osteoarticular Problems of Unknown Etiology. *JIMD Reports*, **2016**, 26, 99-102	1.9	6
141	Investigation of correlation of urinary globotriaosylceramide (Gb3) levels with markers of renal function in patients with Fabry disease. *Clinica Chimica Acta*, **2018**, 478, 62-67	6.2	6
140	Alternative laronidase dose regimen for patients with mucopolysaccharidosis I: a multinational, retrospective, chart review case series. *Orphanet Journal of Rare Diseases*, **2016**, 11, 51	4.2	6
139	Clinical findings in Brazilian patients with adult GM1 gangliosidosis. *JIMD Reports*, **2019**, 49, 96-106	1.9	6
138	Phenotype-oriented NGS panels for mucopolysaccharidoses: Validation and potential use in the diagnostic flowchart. *Genetics and Molecular Biology*, **2019**, 42, 207-214	2	6
137	TGFA/Taq I polymorphism and environmental factors in non-syndromic oral clefts in Southern Brazil. *Brazilian Oral Research*, **2012**, 26, 431-5	2.6	6

136	Impact of measures to enhance the value of observational surveys in rare diseases: the Fabry Outcome Survey (FOS). *Value in Health*, **2011**, 14, 862-6	3.3	6
135	Consumption of folic acid-fortified flour and folate-rich foods among women at reproductive age in South Brazil. *Public Health Genomics*, **2008**, 11, 179-84	1.9	6
134	Molecular characterization of glucose-6-phosphate dehydrogenase deficiency in patients from the southern Brazilian city of Porto Alegre, RS. *Genetics and Molecular Biology*, **2007**, 30, 10-13	2	6
133	An improved specific laboratory test for homocystinuria. *Clinica Chimica Acta*, **1982**, 125, 367-9	6.2	6
132	Prospective study of 11 Brazilian patients with mucopolysaccharidosis II. *Jornal De Pediatria*, **2006**, 82, 273-8	2.6	6
131	Treatment of inborn errors of metabolism. *Jornal De Pediatria*, **2008**, 84, S8-19	2.6	6
130	Clinical research challenges in rare genetic diseases in Brazil. *Genetics and Molecular Biology*, **2019**, 42, 305-311	2	5
129	Diagnosis and therapy options in mucopolysaccharidosis II (Hunter syndrome). *Expert Opinion on Orphan Drugs*, **2015**, 3, 141-150	1.1	5
128	Identification of mutations in Colombian patients affected with Fabry disease. *Gene*, **2015**, 574, 325-9	3.8	5
127	Nanoparticles containing Ecyclodextrin potentially useful for the treatment of Niemann-Pick C. *Journal of Inherited Metabolic Disease*, **2020**, 43, 586-601	5.4	5
126	Spectrum of GALNS mutations and haplotype study in Brazilian patients with Mucopolysaccharidosis type IVA. *Meta Gene*, **2018**, 16, 77-84	0.7	5
125	A 15-Year Perspective of the Fabry Outcome Survey. *FIRE Forum for International Research in Education*, **2016**, 4, 232640981666629	1.4	5
124	Pitfalls in the prenatal diagnosis of mucolipidosis II alpha/beta: A case report. *Meta Gene*, **2014**, 2, 403-6	0.7	5
123	Identification of a novel missense mutation in Brazilian patient with a severe form of mucopolysaccharidosis type IVA. *Gene*, **2013**, 517, 112-5	3.8	5
122	Mucopolysaccharidosis: Caregiver Quality of Life. *FIRE Forum for International Research in Education* , **2015**, 3, 232640981561380	1.4	5
121	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, 150-4	3.8	5
120	Optimized loading test to evaluate responsiveness to tetrahydrobiopterin (BH4) in Brazilian patients with phenylalanine hydroxylase deficiency. *Molecular Genetics and Metabolism*, **2011**, 104 Suppl, S80-5	3.7	5
119	Mucopolysaccharidoses in northern Brazil: Targeted mutation screening and urinary glycosaminoglycan excretion in patients undergoing enzyme replacement therapy. *Genetics and Molecular Biology*, **2011**, 34, 410-5	2	5

118	Alpha-mannosidase activity in goats fed with Sida carpinifolia. *Experimental and Toxicologic Pathology*, **2010**, 62, 191-5		5
117	Prenatal diagnosis of fetal chromosomal abnormalities: report of an 18-year experience in a Brazilian public hospital. *Genetics and Molecular Biology*, **2008**, 31, 829-833	2	5
116	Further cases of "neighbor" mutations in mucopolysaccharidosis type II. *American Journal of Medical Genetics, Part A*, **2006**, 140, 1684-6	2.5	5
115	Newborn screening for lysosomal disorders in Brazil: A pilot study using customized fluorimetric assays. *Genetics and Molecular Biology*, **2020**, 43, e20180334	2	5
114	Precision Medicine for Lysosomal Disorders. *Biomolecules*, **2020**, 10,	5.9	5
113	Updated birth prevalence and relative frequency of mucopolysaccharidoses across Brazilian regions. *Genetics and Molecular Biology*, **2021**, 44, e20200138	2	5
112	Understanding the Early Presentation of Mucopolysaccharidoses Disorders: Results of a Systematic Literature Review and Physician Survey. *FIRE Forum for International Research in Education*, **2018**, 6, 232640981880034	1.4	5
111	The Mucopolysaccharidoses94-100		5
110	Losartan improves aortic dilatation and cardiovascular disease in mucopolysaccharidosis I. *Journal of Inherited Metabolic Disease*, **2017**, 40, 311-312	5.4	4
109	Birth weight in patients with mucopolysaccharidosis type II: Data from the Hunter Outcome Survey (HOS). *Molecular Genetics and Metabolism Reports*, **2017**, 11, 62-64	1.8	4
108	Molecular characterization of a large group of Mucopolysaccharidosis type IIIC patients reveals the evolutionary history of the disease. *Human Mutation*, **2019**, 40, 1084-1100	4.7	4
107	Effects of gene therapy on cardiovascular symptoms of lysosomal storage diseases. *Genetics and Molecular Biology*, **2019**, 42, 261-285	2	4
106	Risks of long-term port use in enzyme replacement therapy for lysosomal storage disorders. *Molecular Genetics and Metabolism Reports*, **2018**, 15, 71-73	1.8	4
105	The migalastat GLP-HEK assay is the gold standard for determining amenability in patients with Fabry disease. *Molecular Genetics and Metabolism Reports*, **2019**, 20, 100494	1.8	4
104	Evaluation of galsulfase for the treatment of mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). *Expert Opinion on Orphan Drugs*, **2014**, 2, 407-417	1.1	4
103	Inclusion of medical genetics in primary health care: report of a pilot project in Brazil. *Journal of Community Genetics*, **2013**, 4, 137-45	2.5	4
102	Abnormal polyamine metabolism is unique to the neuropathic forms of MPS: potential for biomarker development and insight into pathogenesis. *Human Molecular Genetics*, **2017**, 26, 3837-3849	5.6	4
101	Assessment of a pioneer metabolic information service in Brazil. *Public Health Genomics*, **2006**, 9, 127-32	1.9	4

100	The Latin-American network of human genetics. *Public Health Genomics*, **2004**, 7, 74-5	1.9	4
99	Specificity and sensitivity of S100B levels in amniotic fluid for Down syndrome diagnosis. *Life Sciences*, **2004**, 76, 379-84	6.8	4
98	Detection of mucopolysaccharidosis type I heterozygotes on the basis of the biochemical properties of plasma alpha-L-iduronidase. *Clinica Chimica Acta*, **2001**, 312, 81-6	6.2	4
97	Melanoma hereditário: prevalência de fatores de risco em um grupo de pacientes no Sul do Brasil. *Anais Brasileiros De Dermatologia*, **2004**, 79, 53-60	1.6	4
96	A useful routine for biochemical detection and diagnosis of mucopolysaccharidoses. *Genetics and Molecular Biology*, **1998**, 21, 163-167	2	4
95	Application of a glycinated bile acid biomarker for diagnosis and assessment of response to treatment in Niemann-pick disease type C1. *Molecular Genetics and Metabolism*, **2020**, 131, 405-417	3.7	4
94	Multiplex testing for the screening of lysosomal storage disease in urine: Sulfatides and glycosaminoglycan profiles in 40 cases of sulfatiduria. *Molecular Genetics and Metabolism*, **2020**, 129, 106-110	3.7	4
93	Genotype-phenotype studies in a large cohort of Brazilian patients with Hunter syndrome. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2021**, 187, 349-356	3.1	4
92	Deleterious effects of interruption followed by reintroduction of enzyme replacement therapy on a lysosomal storage disorder. *Translational Research*, **2016**, 176, 29-37.e1	11	4
91	Hunter syndrome: Long-term idursulfase treatment does not protect patients against DNA oxidation and cytogenetic damage. *Mutation Research - Genetic Toxicology and Environmental Mutagenesis*, **2018**, 835, 21-24	3	4
90	Population medical genetics: translating science to the community. *Genetics and Molecular Biology*, **2019**, 42, 312-320	2	3
89	Progressive eye pathology in mucopolysaccharidosis type I mice and effects of enzyme replacement therapy. *Clinical and Experimental Ophthalmology*, **2020**, 48, 334-342	2.4	3
88	Correlation of CSF flow using phase-contrast MRI with ventriculomegaly and CSF opening pressure in mucopolysaccharidoses. *Fluids and Barriers of the CNS*, **2017**, 14, 23	7	3
87	Lessons from molecular modeling human α-L-iduronidase. *Journal of Molecular Graphics and Modelling*, **2014**, 54, 107-13	2.8	3
86	Fabry disease: Evidence for a regional founder effect of the gene mutation 30delG in Brazilian patients. *Molecular Genetics and Metabolism Reports*, **2014**, 1, 414-421	1.8	3
85	Correlation Between Flexible Fiberoptic Laryngoscopic and Polysomnographic Findings in Patients with Mucopolysaccharidosis Type VI. *JIMD Reports*, **2016**, 29, 53-58	1.9	3
84	RELAGH - The challenge of having a scientific network in Latin America: An account from the presidents. *Genetics and Molecular Biology*, **2014**, 37, 305-9	2	3
83	Emerging treatment options for the mucopolysaccharidoses. *Research and Reports in Endocrine Disorders*, **2012**, 53		3

82	Non Viral Gene Transfer Approaches for Lysosomal Storage Disorders **2011**,		3
81	Enzyme replacement therapy for mucopolysaccharidosis I: laronidase. *Pediatric Health*, **2010**, 4, 133-145		3
80	Analysis of cDNA molecules is not suitable for the molecular diagnosis of Mucopolysaccharidosis type I. *Diagnostic Molecular Pathology*, **2012**, 21, 53-5		3
79	Comparison of the measurement of lysosomal hydrolase activity in mycoplasma-contaminated and non-contaminated human fibroblast cultures treated with mycoplasma removal agent. *Clinical Biochemistry*, **2007**, 40, 521-5	3.5	3
78	Conventional MRI and MR spectroscopy in nonclassical mitochondrial disease: report of three patients with mitochondrial DNA deletion. *Childls Nervous System*, **2006**, 22, 1355-9	1.7	3
77	Application of a protocol for the detection of disorders of sialic acid metabolism to 124 high-risk Brazilian patients. *American Journal of Medical Genetics Part A*, **2003**, 119A, 348-51		3
76	Aspartame loading test in PKU heterozygous individuals bearing severe and moderate mutations. *Clinical Genetics*, **2000**, 58, 86-8	4	3
75	Congenital malformations detected in 731 autopsies of children aged 0 to 14 years. *Teratology*, **1987**, 35, 305-7		3
74	Tetrahydrobiopterin responsiveness of patients with phenylalanine hydroxylase deficiency. *Jornal De Pediatria*, **2011**, 87, 245-51	2.6	3
73	Prevalence and Phenotypic Expression of Mutations in the MYH7, MYBPC3 and TNNT2 Genes in Families with Hypertrophic Cardiomyopathy in the South of Brazil: A Cross-Sectional Study. *Arquivos Brasileiros De Cardiologia*, **2016**, 107, 257-265	1.2	3
72	Enzyme Replacement Therapy with Pabinafusp Alfa for Neuronopathic Mucopolysaccharidosis II: An Integrated Analysis of Preclinical and Clinical Data. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	3
71	Estimated prevalence of mucopolysaccharidoses from population-based exomes and genomes. *Orphanet Journal of Rare Diseases*, **2020**, 15, 324	4.2	3
70	Encapsulated Whole Bone Marrow Cells Improve Survival in Wistar Rats after 90% Partial Hepatectomy. *Stem Cells International*, **2016**, 2016, 4831524	5	3
69	A charitable access program for patients with lysosomal storage disorders in underserved communities worldwide. *Orphanet Journal of Rare Diseases*, **2021**, 16, 8	4.2	3
68	Long-term follow-up of renal function in patients treated with migalastat for Fabry disease. *Molecular Genetics and Metabolism Reports*, **2021**, 28, 100786	1.8	3
67	Analysis of C6S/C4S ratio in Morquio syndrome type A patients. *Molecular Genetics and Metabolism*, **2015**, 114, S66	3.7	2
66	The Latin American School of Human and Medical Genetics: promoting education and collaboration in genetics and ethics applied to health sciences across the continent. *Journal of Community Genetics*, **2015**, 6, 189-91	2.5	2
65	Diagnosing Mucopolysaccharidosis type IV a by the fluorometric assay of N-Acetylgalactosamine-6-sulfate sulfatase activity. *Journal of Diabetes and Metabolic Disorders*, **2017**, 16, 37	2.5	2

64	Outcomes of a Physician Survey on the Type, Progression, Assessment, and Treatment of Neurological Disease in Mucopolysaccharidoses. *FIRE Forum for International Research in Education*, **2018**, 6, 232640981875937	1.4	2
63	High-Risk Screening and Diagnosis of Inborn Errors of Metabolism: A Practical Guide for Laboratories. *FIRE Forum for International Research in Education*, **2018**, 6, 232640981879206	1.4	2
62	. *FIRE Forum for International Research in Education*, **2017**, 5, 232640981769236	1.4	2
61	Niemann-Pick disease type C: a case series of Brazilian patients. *Arquivos De Neuro-Psiquiatria*, **2014**, 72, 214-8	1.6	2
60	Polymorphic variation of mononucleotide microsatellites in healthy humans and its implication for microsatellite instability screening. *Arquivos De Gastroenterologia*, **2007**, 44, 64-7	1.3	2
59	Effect of CuCl2, NaCl and EDTA on the enzyme alpha-L-iduronidase in the plasma of normal individuals and heterozygotes for MPS I. *Clinica Chimica Acta*, **2002**, 318, 83-9	6.2	2
58	Clinical and biochemical findings in 7 patients with X-linked adrenoleukodystrophy treated with Lorenzo's Oil. *Genetics and Molecular Biology*, **2000**, 23, 697-701	2	2
57	X-linked adrenoleukodystrophy: clinical and laboratory findings in 15 Brazilian patients. *Genetics and Molecular Biology*, **2000**, 23, 261-264	2	2
56	Inborn errors of metabolism. Sensitivity of screening tests in high risk patients. *Clinical Pediatrics*, **1989**, 28, 494-7	1.2	2
55	Frequency of cystinuria among stone-forming patients in region of Brazil. *Urology*, **1986**, 27, 38-40	1.6	2
54	Cancer-related worry and risk perception in Brazilian individuals seeking genetic counseling for hereditary breast cancer. *Genetics and Molecular Biology*, **2020**, 43, e20190097	2	2
53	Development of a Clinical Algorithm for the Early Diagnosis of Mucopolysaccharidosis III. *Journal of Inborn Errors of Metabolism and Screening*,8,	0.8	2
52	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*,8,	0.8	2
51	Evaluation of the long-term treatment effects of intravenous idursulfase in patients with mucopolysaccharidosis II (MPS II) using statistical modeling: data from the Hunter Outcome Survey (HOS). *Orphanet Journal of Rare Diseases*, **2021**, 16, 456	4.2	2
50	Clinical relevance of endpoints in clinical trials for acid sphingomyelinase deficiency enzyme replacement therapy. *Molecular Genetics and Metabolism*, **2020**, 131, 116-123	3.7	2
49	Neonatal Screening for MPS Disorders in Latin America: A Survey of Pilot Initiatives. *International Journal of Neonatal Screening*, **2020**, 6,	2.6	2
48	Assessing the impact of the five senses on quality of life in mucopolysaccharidoses. *Orphanet Journal of Rare Diseases*, **2020**, 15, 97	4.2	2
47	Spinal cord issues in adult patients with MPS: transition of care survey. *Childls Nervous System*, **2018**, 34, 1759-1765	1.7	2

46	Dermal melanocytosis associated with GM1-gangliosidosis type 1. *Acta Dermato-Venereologica*, **2006**, 86, 156-8	2.2	2
45	Simple and efficient screening of patients with Fabry disease with high resolution melting. *Clinical Biochemistry*, **2018**, 53, 160-163	3.5	1
44	Important aspects in the molecular diagnosis of mucopolysaccharidoses. *Journal of Inherited Metabolic Disease*, **2013**, 36, 905-6	5.4	1
43	Chloramphenicol Enhances IDUA Activity on Fibroblasts from Mucopolysaccharidosis I Patients. *Current Pharmaceutical Biotechnology*, **2013**, 14, 194-198	2.6	1
42	Detection of metabolic disorders in high-risk patients. A pilot study in Salvador, Bahia. *Arquivos De Neuro-Psiquiatria*, **1997**, 55, 209-12	1.6	1
41	Common N-acetylgalactosamine-6-sulfate sulfatase (GALNS) exon mutations in Brazilian patients with mucopolysaccharidosis IVA (MPS IVA). *Genetics and Molecular Biology*, **2007**, 30, 524-528	2	1
40	Avaliaã prospectiva de 11 pacientes brasileiros com mucopolissacaridose II. *Jornal De Pediatria*, **2006**, 82, 273-278	2.6	1
39	S100B protein and amniotic fluid. *Clinica Chimica Acta*, **2003**, 335, 165-6; author reply 167	6.2	1
38	Application of a flowchart for the detection of lysosomal storage diseases in 105 high-risk Brazilian patients. *American Journal of Medical Genetics Part A*, **1990**, 37, 534-8		1
37	Brain and visceral gene editing of mucopolysaccharidosis I mice by nasal delivery of the CRISPR/Cas9 system.. *Journal of Gene Medicine*, **2022**, e3410	3.5	1
36	Sanfilippo Syndrome: The Tale of a Challenging Diagnosis. *Journal of Inborn Errors of Metabolism and Screening*,8,	0.8	1
35	Therapeutic Options for Mucopolysaccharidosis II (Hunter Disease). *Current Pharmaceutical Design*, **2020**, 26, 5100-5109	3.3	1
34	Morquio-like dysostosis multiplex presenting with neuronopathic features is a distinct -related phenotype. *JIMD Reports*, **2021**, 60, 23-31	1.9	1
33	Prospective study of the natural history of chronic acid sphingomyelinase deficiency in children and adults: eleven years of observation. *Orphanet Journal of Rare Diseases*, **2021**, 16, 212	4.2	1
32	Mucopolysaccharidosis VII in Brazil: natural history and clinical findings. *Orphanet Journal of Rare Diseases*, **2021**, 16, 238	4.2	1
31	Cardiac pathology in mucopolysaccharidosis I mice: Losartan modifies ERK1/2 activation during cardiac remodeling. *Journal of Inherited Metabolic Disease*, **2021**, 44, 740-750	5.4	1
30	Disruption of morphogenic and growth pathways in lysosomal storage diseases. *WIREs Mechanisms of Disease*, **2021**, 13, e1521	0.3	1
29	MPSBase: Comprehensive repository of differentially expressed genes for mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2021**, 133, 372-377	3.7	1

28	Molecular basis of mucopolysaccharidosis IVA (Morquio A syndrome): A review and classification of GALNS gene variants and reporting of 68 novel variants. *Human Mutation*, **2021**, 42, 1384-1398	4.7	1
27	Schizophreniform presentation and abrupt neurologic decline in a patient with late-onset mucopolysaccharidosis type IIIB. *Psychiatric Genetics*, **2021**, 31, 199-204	2.9	1
26	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	1.8	1
25	Epidemiology of rare diseases in Brazil: protocol of the Brazilian Rare Diseases Network (RARAS-BRDN).. *Orphanet Journal of Rare Diseases*, **2022**, 17, 84	4.2	1
24	Mucopolysaccharidosis VI and effects on growth of the apical bases: a case report. *Special Care in Dentistry*, **2018**, 38, 176-184	1.7	0
23	COVID-19 impact on the diagnosis of Inborn Errors of Metabolism: Data from a reference center in Brazil.. *Genetics and Molecular Biology*, **2021**, 45, e20210253	2	0
22	Progression of Cardiovascular Manifestations in Adults and Children With Mucopolysaccharidoses With and Without Enzyme Replacement Therapy.. *Frontiers in Cardiovascular Medicine*, **2021**, 8, 801147	5.4	0
21	Fifteen years of enzyme replacement therapy for mucopolysaccharidosis type VI (Maroteaux-Lamy syndrome): a case report.. *Journal of Medical Case Reports*, **2022**, 16, 46	1.2	0
20	Measurement of sulfatides in the amniotic fluid supernatant: A useful tool in the prenatal diagnosis of metachromatic leukodystrophy.. *JIMD Reports*, **2022**, 63, 162-167	1.9	0
19	Plasma neurofilament light, glial fibrillary acidic protein and lysosphingolipid biomarkers for pharmacodynamics and disease monitoring of GM2 and GM1 gangliosidoses patients.. *Molecular Genetics and Metabolism Reports*, **2022**, 30, 100843	1.8	0
18	Magnetic resonance imaging findings of the posterior fossa in 47 patients with mucopolysaccharidoses: A cross-sectional analysis. *JIMD Reports*, **2021**, 60, 32-41	1.9	0
17	The Mucopolysaccharidoses **2022**, 1267-1286		0
16	A Case of Early Infantile Pompe Disease with Atypical Manifestation. *Journal of Neuromuscular Diseases*, **2015**, 2, S15-S15	5	
15	Punctate calcifications in lysosomal storage disorders. *Clinical Dysmorphology*, **2009**, 18, 172-177	0.9	
14	Microsatellite instability testing in genetically heterogeneous populations. *Journal of Clinical Oncology*, **2007**, 25, 913-4; author reply 914-5	2.2	
13	Chromosome analysis in 31 cases of benign and malignant breast tumors: a study in Brazil. *Hereditas*, **2002**, 137, 57-64	2.4	
12	Re: Metabolic factors in urolithiasis: a study in Brazil. *Journal of Urology*, **1987**, 137, 320	2.5	
11	Detection of Mosaic Variants in Mothers of MPS II Patients by Next Generation Sequencing. *Frontiers in Molecular Biosciences*, **2021**, 8, 789350	5.6	

10	Cockayne syndrome: report of a Brazilian family with confirmation of impaired RNA synthesis after UV-irradiation. *Genetics and Molecular Biology*, **2000**, 23, 273-275	2
9	Prevalence of the serpin peptidase inhibitor (alpha-1-antitrypsin) PI*S and PI*Z alleles in Brazilian children with liver disease. *Genetics and Molecular Biology*, **2008**, 31, 423-426	2
8	Early infantile form of galactosialidosis in a female baby with a prenatal diagnosis of fetal ascites: First case in Brazil. *Genetics and Molecular Biology*, **1998**, 21, 443-446	2
7	Oral, dental, and craniofacial features in chronic acid sphingomyelinase deficiency. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2891-2901	2.5
6	Demographic, clinical, and ancestry characterization of a large cluster of mucopolysaccharidosis IV A in the Brazilian Northeast region. *American Journal of Medical Genetics, Part A*, **2021**, 185, 2929-2940	2.5
5	Evaluating enzyme replacement therapies for Anderson-Fabry disease: commentary on a recent report. *Genetics and Molecular Biology*, **2018**, 41, 790-793	2
4	Clinical trials for genetic diseases in Latin America. *American Journal of Medical Genetics, Part C: Seminars in Medical Genetics*, **2021**, 187, 381-387	3.1
3	Mucopolysaccharidoses **2021**, 501-562	
2	Genome editing in lysosomal disorders. *Progress in Molecular Biology and Translational Science*, **2021**, 182, 289-325	4
1	A Case of Early Infantile Pompe Disease with Atypical Manifestation. *Journal of Neuromuscular Diseases*, **2015**, 2, S15	5