Roberto Giugliani

List of Publications by Year in Descending Order

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	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
440	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
439	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
438	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
437	The Mucopolysaccharidoses **2022**, 1267-1286		0
436	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
435	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
434	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
433	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
432	Detection of Mosaic Variants in Mothers of MPS II Patients by Next Generation Sequencing. *Frontiers in Molecular Biosciences*, **2021**, 8, 789350	5.6	
431	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
430	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
429	Morquio-like dysostosis multiplex presenting with neuronopathic features is a distinct -related phenotype. *JIMD Reports*, **2021**, 60, 23-31	1.9	1
428	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
427	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
426	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
425	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

424	Mucopolysaccharidosis VII in Brazil: natural history and clinical findings. *Orphanet Journal of Rare Diseases*, **2021**, 16, 238	4.2	1
423	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	
422	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
421	Updated birth prevalence and relative frequency of mucopolysaccharidoses across Brazilian regions. *Genetics and Molecular Biology*, **2021**, 44, e20200138	2	5
420	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
419	Disruption of morphogenic and growth pathways in lysosomal storage diseases. *WIREs Mechanisms of Disease*, **2021**, 13, e1521	0.3	1
418	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
417	MPSBase: Comprehensive repository of differentially expressed genes for mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2021**, 133, 372-377	3.7	1
416	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
415	Schizophreniform presentation and abrupt neurologic decline in a patient with late-onset mucopolysaccharidosis type IIIB. *Psychiatric Genetics*, **2021**, 31, 199-204	2.9	1
414	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	
413	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
412	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
411	Mucopolysaccharidoses **2021**, 501-562		
410	Genome editing in lysosomal disorders. *Progress in Molecular Biology and Translational Science*, **2021**, 182, 289-325	4	
409	Mucopolysaccharidosis Type I. *Diagnostics*, **2020**, 10,	3.8	15
408	Nanoparticles containing Ecyclodextrin potentially useful for the treatment of Niemann-Pick C. *Journal of Inherited Metabolic Disease*, **2020**, 43, 586-601	5.4	5
407	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

406	Newborn screening for lysosomal disorders in Brazil: A pilot study using customized fluorimetric assays. *Genetics and Molecular Biology*, **2020**, 43, e20180334	2	5
405	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
404	Therapeutic Options for Mucopolysaccharidosis II (Hunter Disease). *Current Pharmaceutical Design*, **2020**, 26, 5100-5109	3.3	1
403	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
402	Estimated birth prevalence of mucopolysaccharidoses in Brazil. *American Journal of Medical Genetics, Part A*, **2020**, 182, 469-483	2.5	12
401	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
400	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
399	Therapy development for the mucopolysaccharidoses: Updated consensus recommendations for neuropsychological endpoints. *Molecular Genetics and Metabolism*, **2020**, 131, 181-196	3.7	8
398	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
397	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
396	Neonatal Screening for MPS Disorders in Latin America: A Survey of Pilot Initiatives. *International Journal of Neonatal Screening*, **2020**, 6,	2.6	2
395	Estimated prevalence of mucopolysaccharidoses from population-based exomes and genomes. *Orphanet Journal of Rare Diseases*, **2020**, 15, 324	4.2	3
394	Precision Medicine for Lysosomal Disorders. *Biomolecules*, **2020**, 10,	5.9	5
393	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
392	Oral, dental, and craniofacial features in chronic acid sphingomyelinase deficiency. *American Journal of Medical Genetics, Part A*, **2020**, 182, 2891-2901	2.5	
391	Assessing the impact of the five senses on quality of life in mucopolysaccharidoses. *Orphanet Journal of Rare Diseases*, **2020**, 15, 97	4.2	2
390	Diagnosis of Mucopolysaccharidoses. *Diagnostics*, **2020**, 10,	3.8	20
389	Clinical research challenges in rare genetic diseases in Brazil. *Genetics and Molecular Biology*, **2019**, 42, 305-311	2	5

388	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
387	Effects of gene therapy on cardiovascular symptoms of lysosomal storage diseases. *Genetics and Molecular Biology*, **2019**, 42, 261-285	2	4
386	Recommendations for the management of MPS VI: systematic evidence- and consensus-based guidance. *Orphanet Journal of Rare Diseases*, **2019**, 14, 118	4.2	18
385	Recommendations for the management of MPS IVA: systematic evidence- and consensus-based guidance. *Orphanet Journal of Rare Diseases*, **2019**, 14, 137	4.2	36
384	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
383	Genotype-phenotype relationships in mucopolysaccharidosis type I (MPS I): Insights from the International MPS I Registry. *Clinical Genetics*, **2019**, 96, 281-289	4	31
382	Lysosomal diseases: Overview on current diagnosis and treatment. *Genetics and Molecular Biology*, **2019**, 42, 165-177	2	29
381	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
380	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
379	Population medical genetics: translating science to the community. *Genetics and Molecular Biology*, **2019**, 42, 312-320	2	3
378	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
377	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
376	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
375	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
374	Clinical findings in Brazilian patients with adult GM1 gangliosidosis. *JIMD Reports*, **2019**, 49, 96-106	1.9	6
373	Phenotype-oriented NGS panels for mucopolysaccharidoses: Validation and potential use in the diagnostic flowchart. *Genetics and Molecular Biology*, **2019**, 42, 207-214	2	6
372	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
371	Characterization of glycan substrates accumulating in GM1 Gangliosidosis. *Molecular Genetics and Metabolism Reports*, **2019**, 21, 100524	1.8	10

370	Aortic root dilatation in patients with mucopolysaccharidoses and the impact of enzyme replacement therapy. *Heart and Vessels*, **2019**, 34, 290-295	2.1	13
369	Recommendations for clinical monitoring of patients with acid sphingomyelinase deficiency (ASMD). *Molecular Genetics and Metabolism*, **2019**, 126, 98-105	3.7	23
368	Long-term outcomes of systemic therapies for Hurler syndrome: an international multicenter comparison. *Genetics in Medicine*, **2018**, 20, 1423-1429	8.1	39
367	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
366	Cathepsin B inhibition attenuates cardiovascular pathology in mucopolysaccharidosis I mice. *Life Sciences*, **2018**, 196, 102-109	6.8	20
365	A simple protocol for transfecting human mesenchymal stem cells. *Biotechnology Letters*, **2018**, 40, 617-622		13
364	Neurological manifestations of lysosomal disorders and emerging therapies targeting the CNS. *The Lancet Child and Adolescent Health*, **2018**, 2, 56-68	14.5	27
363	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
362	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
361	Simple and efficient screening of patients with Fabry disease with high resolution melting. *Clinical Biochemistry*, **2018**, 53, 160-163	3.5	1
360	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
359	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
358	Spectrum of GALNS mutations and haplotype study in Brazilian patients with Mucopolysaccharidosis type IVA. *Meta Gene*, **2018**, 16, 77-84	0.7	5
357	CRISPR-Cas9-mediated gene editing in human MPS I fibroblasts. *Gene*, **2018**, 678, 33-37	3.8	7
356	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
355	Burden of Illness in Acid Sphingomyelinase Deficiency: A Retrospective Chart Review of 100 Patients. *JIMD Reports*, **2018**, 41, 119-129	1.9	13
354	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
353	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

352	Mucopolysaccharidosis VI and effects on growth of the apical bases: a case report. *Special Care in Dentistry*, **2018**, 38, 176-184	1.7	0
351	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
350	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
349	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
348	Intrathecal/Intracerebroventricular enzyme replacement therapy for the mucopolysaccharidoses: efficacy, safety, and prospects. *Expert Opinion on Orphan Drugs*, **2018**, 6, 403-411	1.1	10
347	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
346	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
345	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
344	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
343	Evaluating enzyme replacement therapies for Anderson-Fabry disease: commentary on a recent report. *Genetics and Molecular Biology*, **2018**, 41, 790-793	2	
342	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
341	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
340	In vivo genome editing of mucopolysaccharidosis I mice using the CRISPR/Cas9 system. *Journal of Controlled Release*, **2018**, 288, 23-33	11.7	36
339	Glycosaminoglycans analysis in blood and urine of patients with mucopolysaccharidosis. *Molecular Genetics and Metabolism*, **2018**, 125, 44-52	3.7	47
338	Spinal cord issues in adult patients with MPS: transition of care survey. *Childls Nervous System*, **2018** , 34, 1759-1765	1.7	2
337	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
336	Immune tolerance induction for laronidase treatment in mucopolysaccharidosis I. *Molecular Genetics and Metabolism Reports*, **2017**, 10, 61-66	1.8	10
335	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

334	Oxidative damage and redox in Lysosomal Storage Disorders: Biochemical markers. *Clinica Chimica Acta*, **2017**, 466, 46-53	6.2	14
333	Losartan improves aortic dilatation and cardiovascular disease in mucopolysaccharidosis I. *Journal of Inherited Metabolic Disease*, **2017**, 40, 311-312	5.4	4
332	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
331	Elevation of glycosaminoglycans in the amniotic fluid of a fetus with mucopolysaccharidosis VII. *Prenatal Diagnosis*, **2017**, 37, 435-439	3.2	13
330	Consensus recommendation for a diagnostic guideline for acid sphingomyelinase deficiency. *Genetics in Medicine*, **2017**, 19, 967-974	8.1	41
329	Globotriaosylsphingosine induces oxidative DNA damage in cultured kidney cells. *Nephrology*, **2017**, 22, 490-493	2.2	10
328	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
327	Cognitive endpoints for therapy development for neuronopathic mucopolysaccharidoses: Results of a consensus procedure. *Molecular Genetics and Metabolism*, **2017**, 121, 70-79	3.7	23
326	Hydrocephalus and mucopolysaccharidoses: what do we know and what do we not know?. *Childls Nervous System*, **2017**, 33, 1073-1080	1.7	12
325	Epidemiology of mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2017**, 121, 227-240	3.7	128
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	Phase I and II clinical trials for the mucopolysaccharidoses. *Expert Opinion on Investigational Drugs*, **2017**, 26, 1331-1340	5.9	13
322	Treatment of brain disease in the mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2017** , 122S, 25-34	3.7	41
321	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
320	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
319	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
318	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
317	Hematopoietic Stem Cell Transplantation for Patients with Mucopolysaccharidosis II. *Biology of Blood and Marrow Transplantation*, **2017**, 23, 1795-1803	4.7	53

316	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
315	. *FIRE Forum for International Research in Education*, **2017**, 5, 232640981769236	1.4	2
314	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
313	Newborn Screening for Pompe Disease. *Pediatrics*, **2017**, 140, S4-S13	7.4	43
312	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
311	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
310	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
309	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
308	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
307	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
306	Screening for Attenuated Forms of Mucopolysaccharidoses in Patients with Osteoarticular Problems of Unknown Etiology. *JIMD Reports*, **2016**, 26, 99-102	1.9	6
305	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
304	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
303	Development of a bile acid-based newborn screen for Niemann-Pick disease type C. *Science Translational Medicine*, **2016**, 8, 337ra63	17.5	75
302	A 15-Year Perspective of the Fabry Outcome Survey. *FIRE Forum for International Research in Education*, **2016**, 4, 232640981666629	1.4	5
301	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
300	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
299	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

298	Emerging drugs for the treatment of mucopolysaccharidoses. *Expert Opinion on Emerging Drugs*, **2016**, 21, 9-26	3.7	38
297	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
296	Current molecular genetics strategies for the diagnosis of lysosomal storage disorders. *Expert Review of Molecular Diagnostics*, **2016**, 16, 113-23	3.8	7
295	Correlation Between Flexible Fiberoptic Laryngoscopic and Polysomnographic Findings in Patients with Mucopolysaccharidosis Type VI. *JIMD Reports*, **2016**, 29, 53-58	1.9	3
294	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
293	Encapsulated Whole Bone Marrow Cells Improve Survival in Wistar Rats after 90% Partial Hepatectomy. *Stem Cells International*, **2016**, 2016, 4831524	5	3
292	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
291	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
290	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
289	Clinical course of sly syndrome (mucopolysaccharidosis type VII). *Journal of Medical Genetics*, **2016**, 53, 403-18	5.8	103
288	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
287	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
286	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
285	Treatment of Fabry Disease with the Pharmacologic Chaperone Migalastat. *New England Journal of Medicine*, **2016**, 375, 545-55	59.2	254
284	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
283	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
282	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
281	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

280	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
279	Look carefully to the heels! A potentially treatable cause of spastic paraplegia. *Journal of Inherited Metabolic Disease*, **2015**, 38, 363-4	5.4	10
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