

# Newborn Screening for Lysosomal Storage Disorders in Experience

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Citation Report

#	ARTICLE	IF	CITATIONS
1	Searching for the Best Oral Treatment for Hypoglycemic Newborns. <i>Journal of Pediatrics</i> , 2017, 190, 10-12.	0.9	2
2	Newborn Screening for Spinal Muscular Atrophy and Lysosomal Storage Disorders Takes Advantage of Novel Therapies. <i>Journal of Pediatrics</i> , 2017, 190, 9-10.	0.9	2
3	Recent advances in the diagnosis and management of Gaucher disease. <i>Expert Review of Endocrinology and Metabolism</i> , 2018, 13, 107-118.	1.2	78
4	Newborn screening for Pompe disease: impact on families. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 1189-1203.	1.7	27
5	Newborn screening for lysosomal storage disorders by tandem mass spectrometry in North East Italy. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 209-219.	1.7	114
6	Precision newborn screening for lysosomal disorders. <i>Genetics in Medicine</i> , 2018, 20, 847-854.	1.1	99
7	Performance of the Four-Plex Tandem Mass Spectrometry Lysosomal Storage Disease Newborn Screening Test: The Necessity of Adding a 2nd Tier Test for Pompe Disease. <i>International Journal of Neonatal Screening</i> , 2018, 4, 41.	1.2	17
8	Newborn screening in mucopolysaccharidoses. <i>Italian Journal of Pediatrics</i> , 2018, 44, 126.	1.0	50
9	Mutations in the GLA Gene and LysoGb3: Is It Really Anderson-Fabry Disease?. <i>International Journal of Molecular Sciences</i> , 2018, 19, 3726.	1.8	63
10	The new frame for Mucopolysaccharidoses. <i>Italian Journal of Pediatrics</i> , 2018, 44, 117.	1.0	6
11	Pharmacological treatment of pediatric Gaucher disease. <i>Expert Review of Clinical Pharmacology</i> , 2018, 11, 1183-1194.	1.3	20
12	International working group identifies need for newborn screening for mucopolysaccharidosis type I but states that existing hurdles must be overcome. <i>Acta Paediatrica, International Journal of Paediatrics</i> , 2018, 107, 2059-2065.	0.7	10
13	GNPTAB c.2404C>T nonsense mutation in a patient with mucopolipidosis III alpha/beta: a case report. <i>BMC Medical Genetics</i> , 2018, 19, 162.	2.1	2
14	Current State of the Art of Newborn Screening for Lysosomal Storage Disorders. <i>International Journal of Neonatal Screening</i> , 2018, 4, 24.	1.2	16
15	Incidence of 4 Lysosomal Storage Disorders From 4 Years of Newborn Screening. <i>JAMA Pediatrics</i> , 2018, 172, 696.	3.3	43
16	Status of newborn screening and follow up investigations for Mucopolysaccharidoses I and II in Taiwan. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 84.	1.2	50
17	Failure to shorten the diagnostic delay in two ultra-orphan diseases (mucopolysaccharidosis types I) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 5	1.2	47
18	Updates in Newborn Screening. <i>Pediatric Annals</i> , 2018, 47, e187-e190.	0.3	21

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19	Galactosidase A-deficient rats accumulate glycosphingolipids and develop cardiorenal phenotypes of Fabry disease. <i>FASEB Journal</i> , 2019, 33, 418-429.	0.2	24
20	The New York pilot newborn screening program for lysosomal storage diseases: Report of the First 65,000 Infants. <i>Genetics in Medicine</i> , 2019, 21, 631-640.	1.1	113
21	Symptoms and Quality of Life in Patients with Fabry Disease: Results from an International Patient Survey. <i>Advances in Therapy</i> , 2019, 36, 2866-2880.	1.3	17
22	Implementation of Second-Tier Tests in Newborn Screening for Lysosomal Disorders in North Eastern Italy. <i>International Journal of Neonatal Screening</i> , 2019, 5, 24.	1.2	45
23	Rats deficient in galactosidase A develop ocular manifestations of Fabry disease. <i>Scientific Reports</i> , 2019, 9, 9392.	1.6	9
24	Population-Based Newborn Screening for Mucopolysaccharidosis Type II in Illinois: The First Year Experience. <i>Journal of Pediatrics</i> , 2019, 214, 165-167.e1.	0.9	14
25	Niemann-Pick disease A or B in four pediatric patients and SMPD1 mutation carrier frequency in the Mexican population. <i>Annals of Hepatology</i> , 2019, 18, 613-619.	0.6	6
26	The North Carolina Experience with Mucopolysaccharidosis Type I Newborn Screening. <i>Journal of Pediatrics</i> , 2019, 211, 193-200.e2.	0.9	22
27	Genotype-phenotype relationships in mucopolysaccharidosis type I (MPS I): Insights from the International MPS I Registry. <i>Clinical Genetics</i> , 2019, 96, 281-289.	1.0	54
28	Newborn Screening for Lysosomal Storage Disorders: Methodologies for Measurement of Enzymatic Activities in Dried Blood Spots. <i>International Journal of Neonatal Screening</i> , 2019, 5, 1.	1.2	38
29	Consensus recommendations for diagnosis, management and treatment of Fabry disease in paediatric patients. <i>Clinical Genetics</i> , 2019, 96, 107-117.	1.0	87
30	The Role of Technology in Newborn Screening. <i>North Carolina Medical Journal</i> , 2019, 80, 49-53.	0.1	8
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32	Recent trends in mucopolysaccharidosis research. <i>Journal of Human Genetics</i> , 2019, 64, 127-137.	1.1	31
33	Evaluation of the detection of GBA missense mutations and other variants using the Oxford Nanopore MinION. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2019, 7, e564.	0.6	65
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35	Expanding the clinical utility of glucosylsphingosine for Gaucher disease. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 558-563.	1.7	26
36	High-risk screening for Fabry disease in a Canadian cohort of chronic kidney disease patients. <i>Clinica Chimica Acta</i> , 2020, 501, 234-240.	0.5	11

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37	The role of Immunity in Fabry Disease and Hypertension: A Review of a Novel Common Pathway. High Blood Pressure and Cardiovascular Prevention, 2020, 27, 539-546.	1.0	9
38	Second-Generation Pharmacological Chaperones: Beyond Inhibitors. Molecules, 2020, 25, 3145.	1.7	31
39	Newborn Screening for Mucopolysaccharidosis I: Moving Forward Learning from Experience. International Journal of Neonatal Screening, 2020, 6, 91.	1.2	8
40	Fabry Disease practice resource: Focused revision. Journal of Genetic Counseling, 2020, 29, 715-717.	0.9	1
41	Fabry disease screening in high-risk populations in Japan: a nationwide study. Orphanet Journal of Rare Diseases, 2020, 15, 220.	1.2	9
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45	A highly multiplexed biochemical assay for analytes in dried blood spots: application to newborn screening and diagnosis of lysosomal storage disorders and other inborn errors of metabolism. Genetics in Medicine, 2020, 22, 1262-1268.	1.1	18
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47	Estimated birth prevalence of Menkes disease and ATP7A-related disorders based on the Genome Aggregation Database (gnomAD). Molecular Genetics and Metabolism Reports, 2020, 24, 100602.	0.4	20
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50	Newborn Screening for Pompe Disease in Illinois: Experience with 684,290 Infants. International Journal of Neonatal Screening, 2020, 6, 4.	1.2	19
51	A Comparative Effectiveness Study of Newborn Screening Methods for Four Lysosomal Storage Disorders. International Journal of Neonatal Screening, 2020, 6, 44.	1.2	23
52	Screening for Fabry Disease in Patients With Juvenile Systemic Lupus Erythematosus. Archives of Rheumatology, 2020, 35, 7-12.	0.3	4
53	Lessons Learned from Pompe Disease Newborn Screening and Follow-up. International Journal of Neonatal Screening, 2020, 6, 11.	1.2	20
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61	Newborn Screening for Pompe Disease. <i>International Journal of Neonatal Screening</i> , 2020, 6, 31.	1.2	28
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63	Toward newborn screening of metachromatic leukodystrophy: results from analysis of over 27,000 newborn dried blood spots. <i>Genetics in Medicine</i> , 2021, 23, 555-561.	1.1	31
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67	Epidemiology of Mucopolysaccharidoses Update. <i>Diagnostics</i> , 2021, 11, 273.	1.3	50
68	Current and emerging pharmacotherapy for Gaucher disease in pediatric populations. <i>Expert Opinion on Pharmacotherapy</i> , 2021, 22, 1489-1503.	0.9	10
70	Newborn Screening for Krabbe Disease – Illinois Experience: Role of Psychosine in Diagnosis of the Disease. <i>International Journal of Neonatal Screening</i> , 2021, 7, 24.	1.2	11
71	Opinions of adults affected with later-onset lysosomal storage diseases regarding newborn screening: A qualitative study. <i>Journal of Genetic Counseling</i> , 2021, 30, 1544-1558.	0.9	5
72	Epidemiology of mucopolysaccharidoses (MPS) in United States: challenges and opportunities. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 241.	1.2	33
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75	Diagnosis of Mucopolysaccharidoses and Mucolipidosis by Assaying Multiplex Enzymes and Glycosaminoglycans. <i>Diagnostics</i> , 2021, 11, 1347.	1.3	5
76	The future of newborn screening for lysosomal disorders. <i>Neuroscience Letters</i> , 2021, 760, 136080.	1.0	17
77	Detecting lysosomal storage disorders by glycomic profiling using liquid chromatography mass spectrometry. <i>Molecular Genetics and Metabolism</i> , 2021, 134, 43-52.	0.5	11
78	High-Risk Screening for Fabry Disease: A Nationwide Study in Japan and Literature Review. <i>Diagnostics</i> , 2021, 11, 1779.	1.3	5
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81	Plasma lyso-sphingomyelin levels are positively associated with clinical severity in acid sphingomyelinase deficiency. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 28, 100780.	0.4	13
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86	Mutational spectrum of SMPD1 gene in Pakistani Niemann-Pick disease patients. <i>Pakistan Journal of Medical Sciences</i> , 2020, 36, 479-484.	0.3	9
87	Cornea verticillata and acroparesthesia efficiently discriminate clusters of severity in Fabry disease. <i>PLoS ONE</i> , 2020, 15, e0233460.	1.1	9
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111	Prevalence of Fabry disease-causing variants in the UK Biobank. Journal of Medical Genetics, 2023, 60, 391-396.	1.5	10
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123	Importance to include differential diagnostics for acid sphingomyelinase deficiency (ASMD) in patients suspected to have to Gaucher disease. Molecular Genetics and Metabolism, 2023, 139, 107563.	0.5	1
124	The <sc>D409H</sc> variant in <sc><i>GBA1</i></sc> : Challenges in predicting the Gaucher phenotype in the newborn screening era. American Journal of Medical Genetics, Part A, 0, , .	0.7	0