

## Supplementary Material

**Supplementary Table S1.** Search strategy to identify studies for the SLR.

### *Embase*

#	Searches	Strategy
1	exp newborn screening/ or ((new born or new-born or newborn) adj3 screening).mp.	NBS
2	(mucopolysaccharidosis or (hunter* adj (syndrome or disease)) or MPS).mp.	MPS
3	exp lysosome storage disease/ or lysosomal storage disease?.mp.	LSD
4	2 or 3	MPS or LSD
5	1 and 4	

### *MEDLINE*

#	Searches	Strategy
1	exp newborn screening/ or ((new born or new-born or newborn) adj3 screening).mp.	NBS
2	(mucopolysaccharidosis or (hunter* adj (syndrome or disease)) or MPS).mp.	MPS
3	exp lysosome storage disease/ or lysosomal storage disease?.mp.	LSD
4	2 or 3	MPS or LSD
5	1 and 4	

### *Cochrane libraries*

#	Searches	Strategy
1	exp newborn screening/ or ((new born or new-born or newborn) adj3 screening).mp.	NBS
2	(mucopolysaccharidosis or (hunter* adj (syndrome or disease)) or MPS).mp.	MPS
3	exp lysosome storage disease/ or lysosomal storage disease?.mp.	LSD
4	2 or 3	MPS or LSD
5	1 and 4	

LSD, lysosomal storage disease; MPS, mucopolysaccharidosis; NBS, newborn screening.

**Supplementary Table S2.** Eligibility criteria for studies identified by the SLR.

Category	Inclusion criteria
Population	Patients undergoing NBS for any LSD (initial search) or MPS II (updated search)
Interventions	Not restricted by intervention
Comparator	Not restricted by comparators
Outcomes	Status
	Countries with existing programs, pilot studies, or plans for NBS for any type of LSD (initial search) or MPS II (updated search)
	Guidelines and recommendations for existing NBS programs (updated search only)
	Technologies
	Methodology/technologies in use for NBS (brief overview only)
	Diagnostic testing validity and reliability assessments (initial search only)
	Management of false positives and pseudodeficiency; use of additional testing to minimize false positives (updated search only)
	Implementation
	Barriers to implementation of NBS in different countries/regions (e.g., infrastructure, perceived cost–benefit ratio, meeting criteria to be added to screening panel)
	Outcomes
Study designs	Outcomes of NBS pilot studies/existing programs on mortality and morbidity, including estimated incidence, prevalence, <sup>a</sup> and follow-up experience; impact on mortality and morbidity outcomes
	Patient follow-up and management (referral and support systems, uncertainty around prognosis/genotype–phenotype relationships, treatment decisions)
	Not restricted by study design
	Not restricted by date
	Not restricted by country
Language	English (initial search) or not limited by publication language (updated search)

<sup>a</sup> Prevalence estimates extracted if presented in included studies.

LSD, lysosomal storage disease; MPS, mucopolysaccharidosis; NBS, newborn screening; SLR, systematic literature review.

**Supplementary Table S3.** Summary of included articles on MPS II.

First author, year (publication type)	Study design	Country	Number of patients/samples	LSDs covered	Status	Technology	Estimated birth prevalence	Follow- up and outcomes
<b>Total number of articles on topic</b>					21	44	17	6
<b>Burton, 2023 [38] (full publication)</b>	Laboratory study	USA	586,323 infants screened	MPS II	✓	✓	✓	
<b>Ream, 2023 [29] (full publication)</b>	Narrative review	USA	NR	MPS II	✓			✓
<b>Brower, 2022 [35] (full publication)</b>	Narrative review and survey	USA	36 experts completed the survey	Various LSDs, including MPS II	✓			
<b>Chuang, 2022 [41] (full publication)</b>	Laboratory study	Taiwan	546,040 infants screened for MPS II	MPS I, II, IVA, and VI	✓	✓	✓	
<b>Jones, 2022 [48] (full publication)</b>	Evaluative analysis	Europe	NR	Various IMDs, including MPS II				✓
<b>Lin, 2022 [53] (full publication)</b>	Prospective observational study	Taiwan	548,624 newborns screened (study is focused on the 202 infants referred for confirmatory diagnosis)	MPS II	✓	✓	✓	✓
<b>Millington, 2022 [30] (full publication)</b>	Editorial	USA	NR	MPS II	✓	✓	✓	
<b>Okuyama, 2022 [79] (abstract)</b>	Laboratory study	Japan	Over 30,000 newborns	MPS I, II, and VI, and Pompe disease	✓	✓	✓	
<b>Pique, 2022 [78] (abstract)</b>	Cross- sectional study	USA	NR	Various LSDs, including MPS II	✓			
<b>Chuang, 2021 [27] (full publication)</b>	Laboratory study	Taiwan	More than 600,000 infants total, including 307,731 screened for MPS II	MPS I, II, VI, and IVA	✓	✓	✓	
<b>Quadri, 2021 [73] (abstract)</b>	Laboratory study	USA	24 newborns	MPS II	✓	✓		
<b>Bilyeu, 2020 [34] (full publication)</b>	Laboratory study	USA	NR	MPS II	✓	✓	✓	
<b>Burton, 2020 [37] (full publication)</b>	Laboratory study	USA	339,269 infants screened	MPS II	✓	✓	✓	
<b>Chien, 2020 [66] (abstract)</b>	Screening study	Taiwan	73,743 newborns screened	Fabry disease, Gaucher disease, MPS I, II, IIIB, IVA, and VI, and Pompe disease	✓	✓	✓	

First author, year (publication type)	Study design	Country	Number of patients/samples	LSDs covered	Status	Technology	Estimated birth prevalence	Follow- up and outcomes
Chien, 2020 [23] (full publication)	Laboratory study	Taiwan	73,743 newborns screened	Fabry disease, Gaucher disease, MPS I, II, IIIB, IVA, and VI, and Pompe disease	✓	✓	✓	
Fdil, 2020 [45] (full publication)	Laboratory study <sup>a</sup>	Morocco	88 patients with suspected MPS	MPS I and II		✓		
Gelb, 2020 [69] (abstract)	Laboratory study	USA	NR	Fabry disease, Gaucher disease, Krabbe disease, MPS I, II, IIIA, IIIB, IIIC, IIID, IVA, VI, and VII, Niemann–Pick disease type A/B/C, Pompe disease, and others		✓		
Gelb, 2020 [70] (abstract)	Laboratory study	USA	NR	MPS I, II, IIIA, IIIB, IVA, IV, and VII		✓		
Khaledi, 2020 [49] (full publication)	Laboratory study	USA	NR	All MPS types except MPS IX		✓		
Oguni, 2020 [60] (full publication)	Laboratory study	Japan	672 DBS samples from healthy newborns and 23 patients with MPS	MPS I, II, IIIB, IVA, and VI		✓		
Pollard, 2020 [72] (abstract)	Laboratory study	USA	446 infants	MPS I and II	✓	✓		
Sabir, 2020 [62] (full publication)	Laboratory study	Morocco	47 patients with MPS	MPS I and II		✓		
Scott, 2020 [22] (full publication)	Laboratory study	USA	~106,000 newborns	MPS II, IIIB, IVA, VI, and VII	✓	✓	✓	
Stapleton, 2020 [25] (full publication)	Laboratory study	Japan	18,222 DBS samples	MPS II		✓	✓	
Wasserstein, 2020 [77] (abstract)	Laboratory study	USA	150,000–175,000 infants	CLN2, CTX, Fabry disease, Gaucher disease, lysosomal acid lipase deficiency, MLD, MPS II, IIIB, IVA, VI, and VII, and Niemann–Pick	✓	✓		

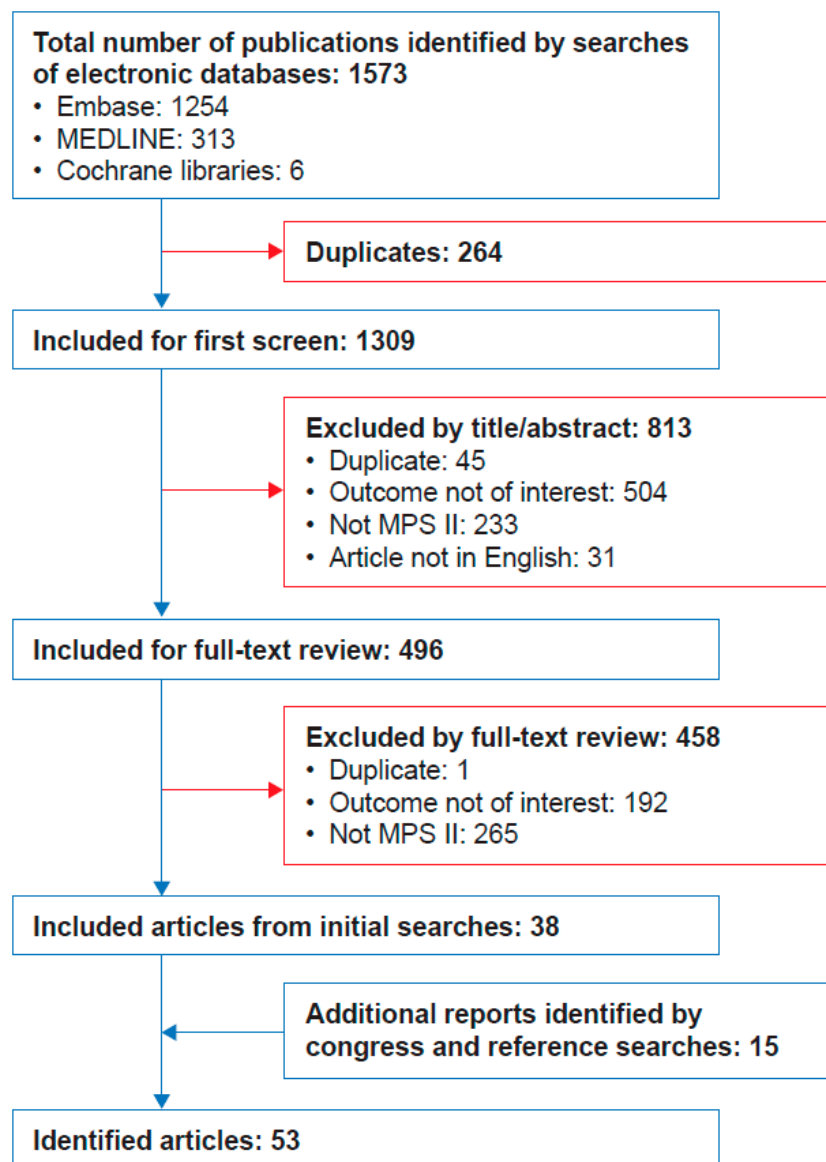
First author, year (publication type)	Study design	Country	Number of patients/samples	LSDs covered	Status	Technology	Estimated birth prevalence	Follow- up and outcomes
				disease type A/B/C				
Burton, 2019 [36] (full publication)	Laboratory study	USA	162,000 infant samples screened	MPS II	✓	✓	✓	
Chan, 2019 [39] (full publication)	Laboratory study	Taiwan	130,237 newborns screened	MPS I, II, and VI	✓	✓	✓	
Chuang, 2019 [67] (abstract)	Laboratory study	Taiwan	280,984 infants screened for MPS	MPS I, II, and VI	✓	✓	✓	
Fabie, 2019 [44] (full publication)	Narrative review	USA	NR	Fabry disease, Gaucher disease, Krabbe disease, MPS I and II, Niemann–Pick disease type A/B, and Pompe disease	✓			
Gelb, 2019 [46] (full publication)	SLR	Italy, Taiwan, USA	NR	Fabry disease, Gaucher disease, Krabbe disease, MLD, MPS I, II, IIIA, IIIB, IVA, VI, and VII, Niemann–Pick disease type A/B/C, Pompe disease, and others		✓		
Kumar, 2019 [51] (full publication)	Laboratory study	USA	NR	Fabry disease, Gaucher disease, Krabbe disease, MPS I, II, IIIB, IVA, VI, and VII, Niemann–Pick disease type A/B, and Pompe disease		✓		
Menkovic, 2019 [59] (full publication)	Laboratory study	Canada	500 urine samples	MPS I, II, III, VI, and VII		✓		
Tomatsu, 2019 [74] (abstract)	Laboratory study	NR	17,467 DBS samples from newborns and 14 DBS samples from newborns with MPS (two with MPS II)	MPS I, II, and III		✓		

First author, year (publication type)	Study design	Country	Number of patients/samples	LSDs covered	Status	Technology	Estimated birth prevalence	Follow- up and outcomes
Joseph, 2018 [47] (full publication)	SLR	NR	NR	MPS II			✓	✓
Maccari, 2018 [56] (abstract)	Laboratory study	Italy	600 healthy newborns and six newborns with MPS (MPS II, n = 2)	MPS unspecified		✓		
Gelb, 2017 [68] (abstract)	Laboratory study	USA	~40,000 newborns screened	CLN1, CLN2, CTX, Krabbe disease, MLD, MPS I, II, IIIB, IVA, VI, and VII, Niemann–Pick disease type C, and Pompe disease		✓		
Liu, 2017 [55] (full publication)	Laboratory study	USA	DBS samples from 62 healthy newborns	CLN2 and MPS I, II, IIIB, IVA, VI, and VII		✓		
Ullal, 2017 [75] (abstract)	Laboratory study	USA	200 presumed normal and 26 MPS II DBS samples	MPS I and II, and Pompe disease		✓		
Lisi, 2016 [54] (full publication)	Survey	USA, Canada, India	38 HCPs	Fabry disease, Gaucher disease, Krabbe disease, MPS I and II, and Pompe disease				✓
Kumar, 2015 [50] (full publication)	Laboratory study	USA	NR	MPS II, IVA, and VI		✓		
Lee, 2015 [52] (article)	Laboratory study	South Korea	DBS from 110 healthy newborns and from three patients with MPS II	MPS II		✓		
Chennamaneni, 2014 [40] (full publication)	Laboratory study	USA	NR	MPS I, II, and VI		✓		
Matern, 2014 [71] (abstract)	Laboratory study	USA	100,000 newborns	Various LSDs, including MPS II			✓	
Ruijter, 2014 [61] (full publication)	Laboratory study	Netherlands	1426 newborns	MPS II		✓		
Shimada, 2014 [63] (full publication)	Laboratory study	NR	Blood (plasma or serum) samples from 55 patients	MPS II, III, and IV		✓		

First author, year (publication type)	Study design	Country	Number of patients/samples	LSDs covered	Status	Technology	Estimated birth prevalence	Follow- up and outcomes
			with MPS II; DBS sample from one patient with MPS II					
van Vlies, 2013 [76] (abstract)	Laboratory study	NR	One patient	MPS I, II, and III		✓		
de Ruijter, 2012 [43] (full publication)	Laboratory study	Netherlands	18 newborn DBS samples screened	MPS I, II, and III		✓		
Alonso- Fernández, 2010 [33] (full publication)	Laboratory study	Argentina/Spain	903 newborn samples screened	MPS I, II, and VI		✓		
Yamaguchi, 2008 [65] (full publication)	Narrative review	Japan	NR	Various LSDs, including MPS II				✓
Wang, 2007 [64] (full publication)	Laboratory study	USA	13 patients with MPS II and 57 randomly chosen newborns	MPS II		✓		
Civallero, 2006 [42] (full publication)	Laboratory study	Brazil	NR	Fabry disease, Gaucher disease, MPS I, II, IIIB, VI, and VII, and others		✓		
Meikle, 2006 [58] (full publication)	Laboratory study	Australia, Denmark	Samples from 92 individuals after diagnosis of an LSD	MLD, MPS I, II, IIIA, and VI, and Niemann–Pick disease type A/B		✓		
Meikle, 2004 [57] (full publication)	Laboratory study	Denmark, Australia	Guthrie cards retrieved from 47 patients with LSDs (n = 47)	Fabry disease, Gaucher disease, I-cell disease, Krabbe disease, MPS II, IVA, and IIIA, Pompe disease, and Tay–Sachs disease		✓		

<sup>a</sup> Targeted screening program of patients with suspected MPS aged 9 months–15.6 years. CLN, neuronal ceroid lipofuscinosis; CTX, cerebrotendinous xanthomatosis; DBS, dried blood spot(s); IMD, inherited metabolic disease; HCP, healthcare professional; LSD, lysosomal storage disease; MLD, metachromatic leukodystrophy; MPS, mucopolysaccharidosis; NBS, newborn screening; NR, not reported; SLR, systematic literature review.

**Supplementary Figure 1.** PRISMA diagram of studies excluded from and included in the SLR. MPS, mucopolysaccharidosis; PRISMA, Preferred Reporting Items for Systematic reviews and Meta-Analyses; SLR, systematic literature review.





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