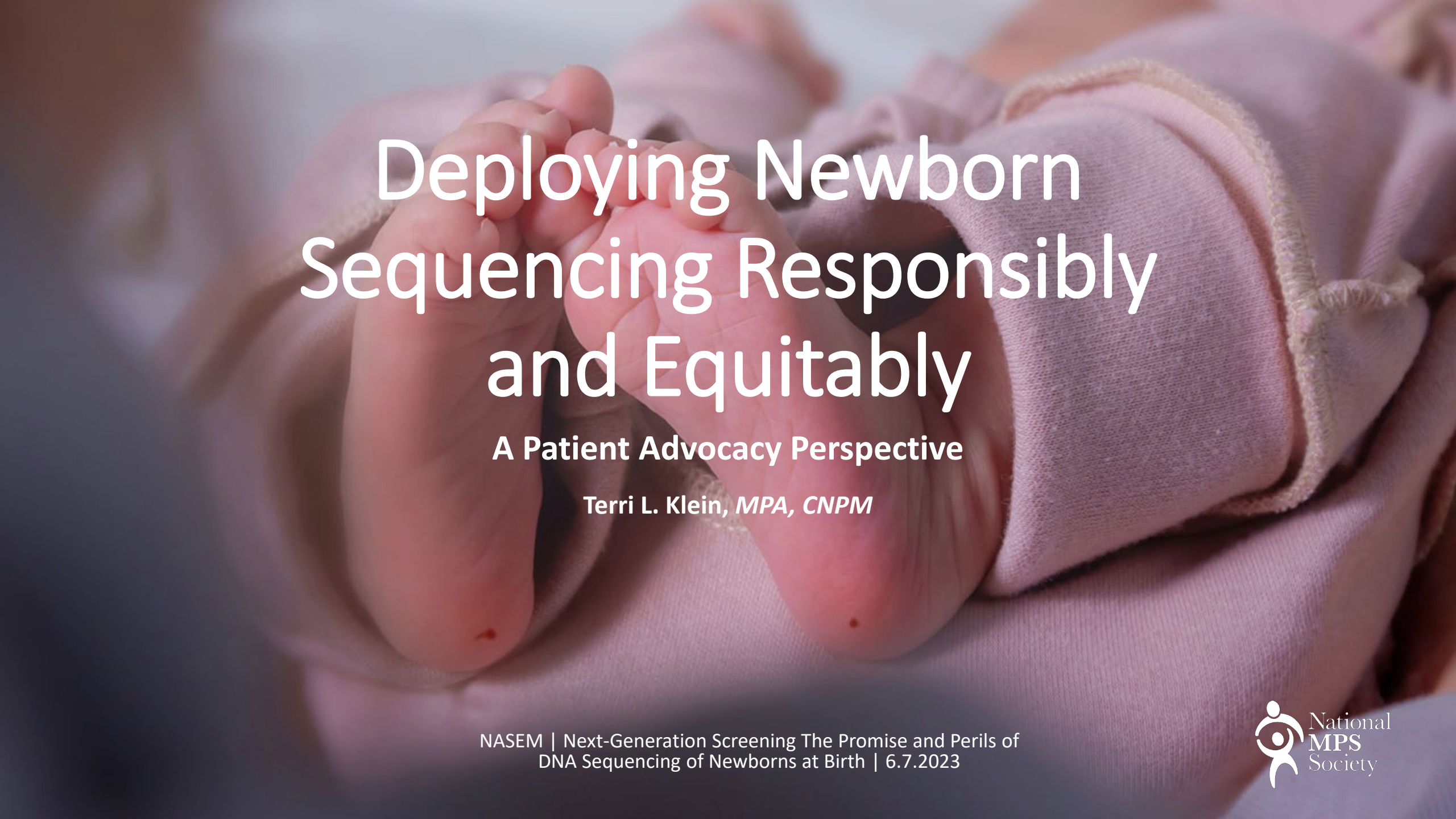


Deploying Newborn Sequencing Responsibly and Equitably

A Patient Advocacy Perspective

Terri L. Klein, *MPA, CNPM*

NASEM | Next-Generation Screening The Promise and Perils of DNA Sequencing of Newborns at Birth | 6.7.2023

National MPS Society

Disclosures

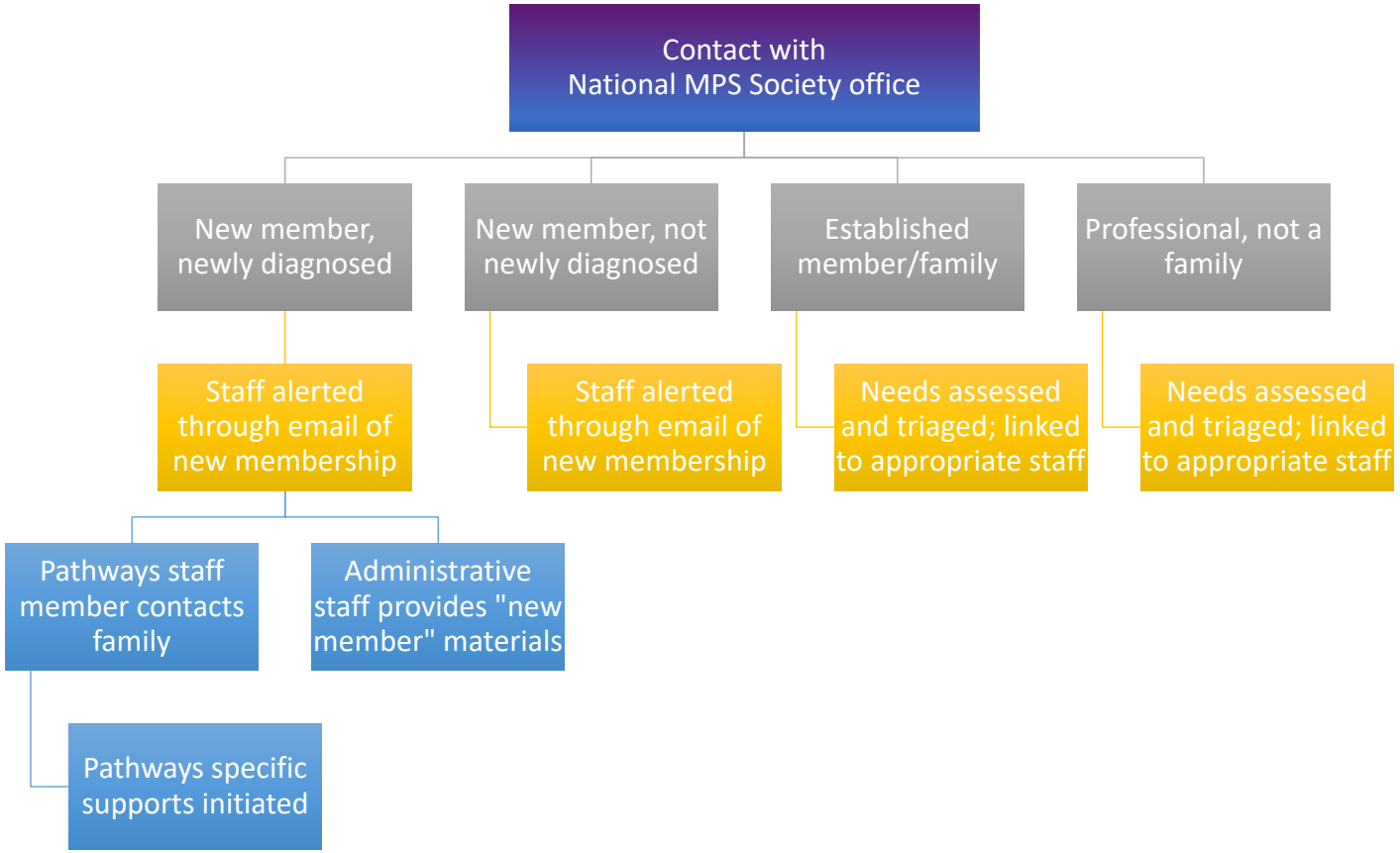
- Patient Advisory Board – Sanofi Genzyme
- Patient Advisor – BioMarin
- Patient Advisory – Takeda
- Patient Advisory – Ultragenyx
- Patient Advocate – Lobbyist

Introducing Pathways

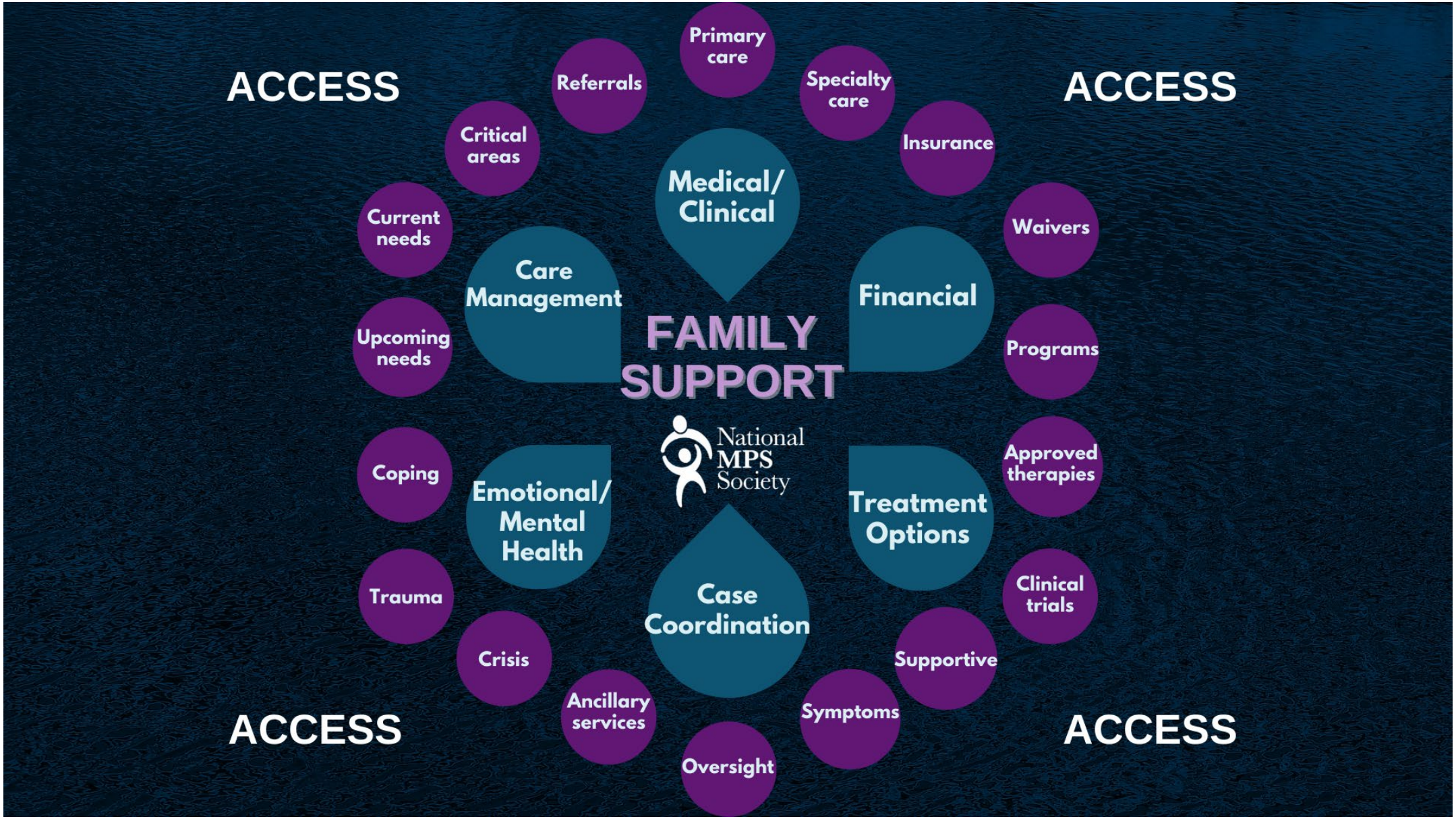
- Traveling the Journey Together
- Implemented in 2017
- Overseen by Licensed Social Workers
- Face-to-Face Care – traveling all 50 states
- 336 Families involved in the Pathways Program
- 81 Active Families
- Approximately 40% through Newborn Screening
- Referrals from HCPs, Online Social Media, Website, Google

National MPS Society
Support for Families. Research for a Cure.

Initial Communication



National MPS Society
Support for Families. Research for a Cure.



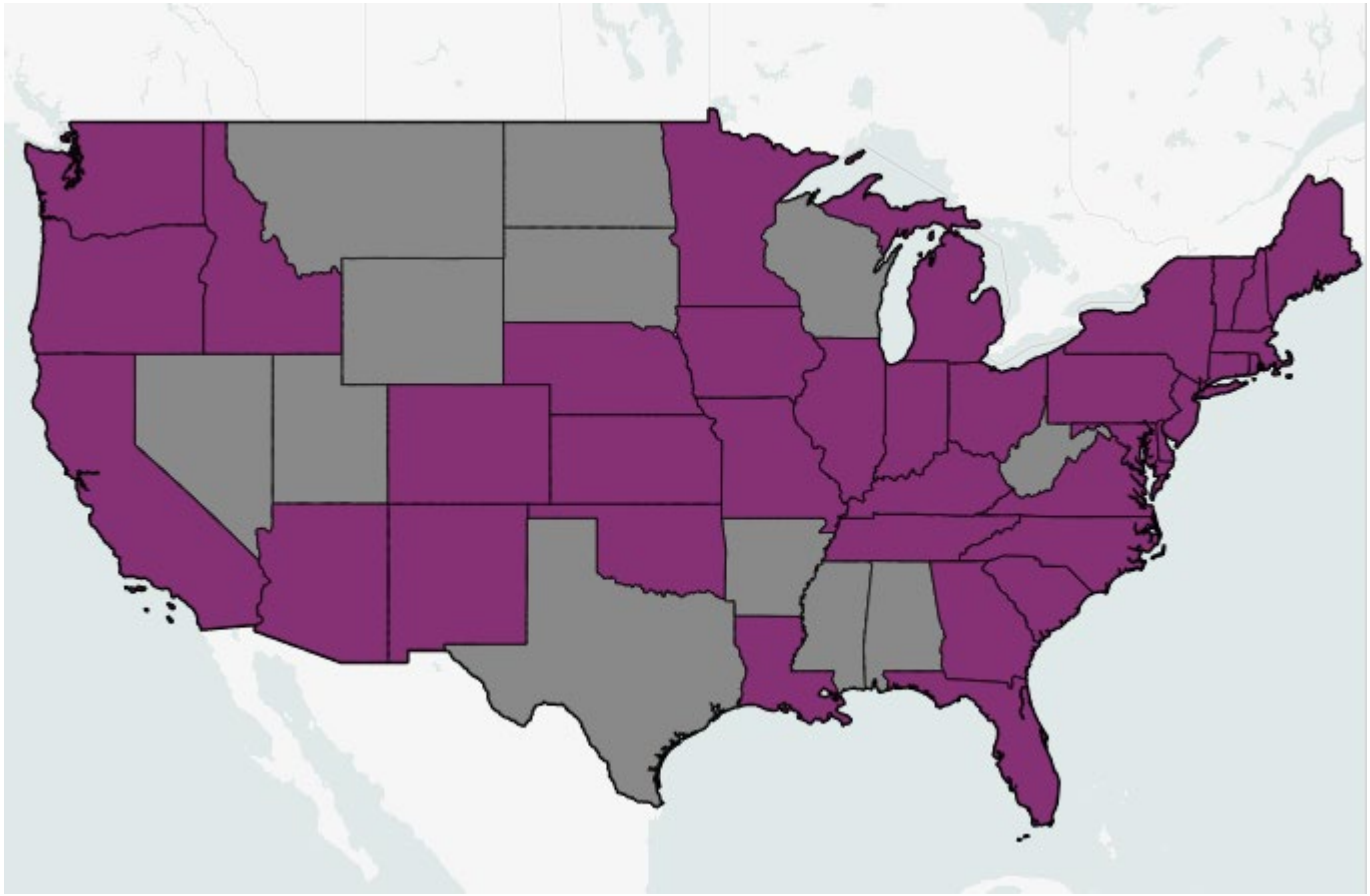
National MPS Society
Support for Families. Research for a Cure.

Newborn Screening for MPS I and MPS II in the U.S.

NASEM | Next-Generation Screening The Promise and Perils of DNA Sequencing of Newborns at Birth | 6.7.2023

National MPS Society
Support for Families. Research for a Cure.

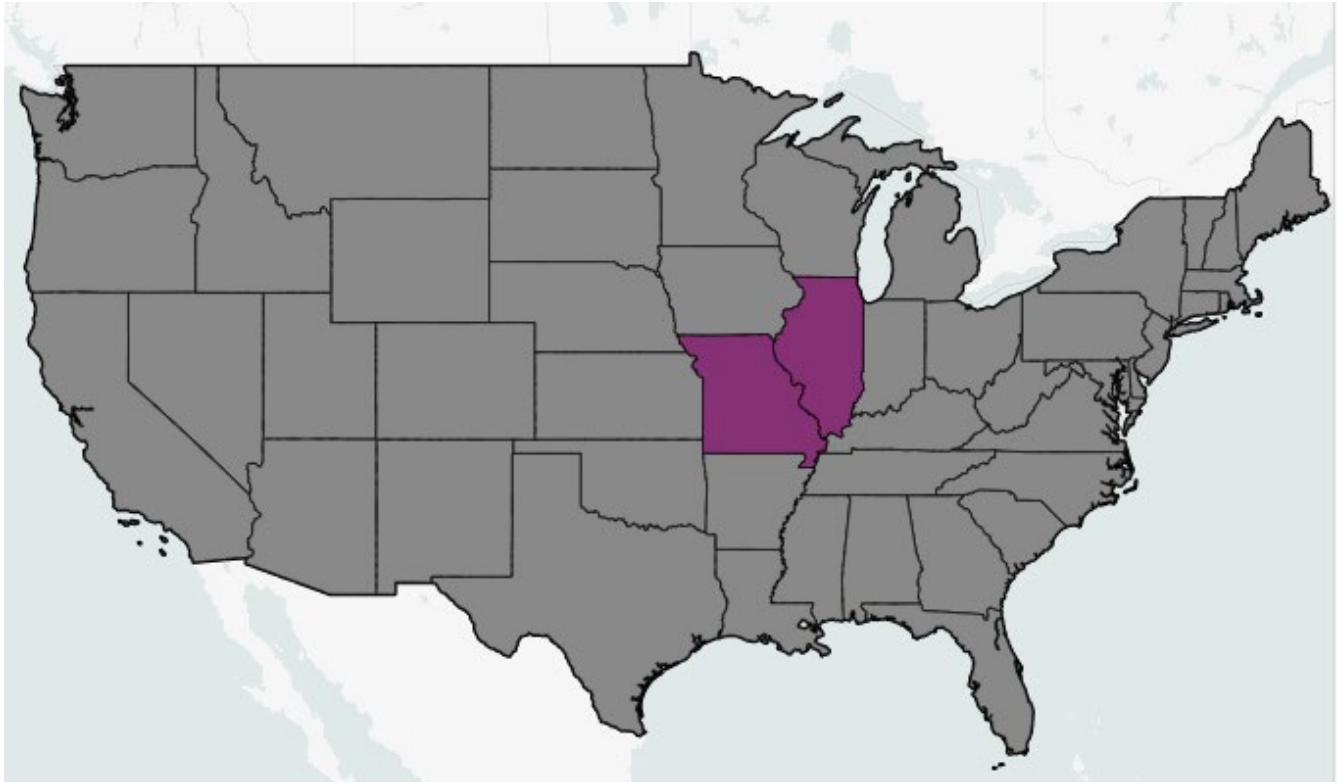
Mucopolysaccharidosis Type I



82% Currently Screened
With the addition of Texas – we will be screening 90%
WV is screening now

National MPS Society
Support for Families. Research for a Cure.

Mucopolysaccharidosis Type II



- Added to RUSP in August 2022
- Anticipated State Labs adding MPS II faster than MPS I

National MPS Society
Support for Families. Research for a Cure.

Projected 3-year Window on MPS II Adoption

Operational Alignment Legislation	37.25%
NY & TX	15.76%
MO (RUSP enabling Pilot)	1.88%
IL (RUSP enabling Pilot)	3.85%
Total Projected Screened	58.74%

Building a Program, New challenge of Diversity and Equity

NASEM | Next-Generation Screening The Promise and Perils of DNA Sequencing of Newborns at Birth | 6.7.2023

National MPS Society
Support for Families. Research for a Cure.

Is the Pathways Program Identifying Diverse Families and Families with Disparities?

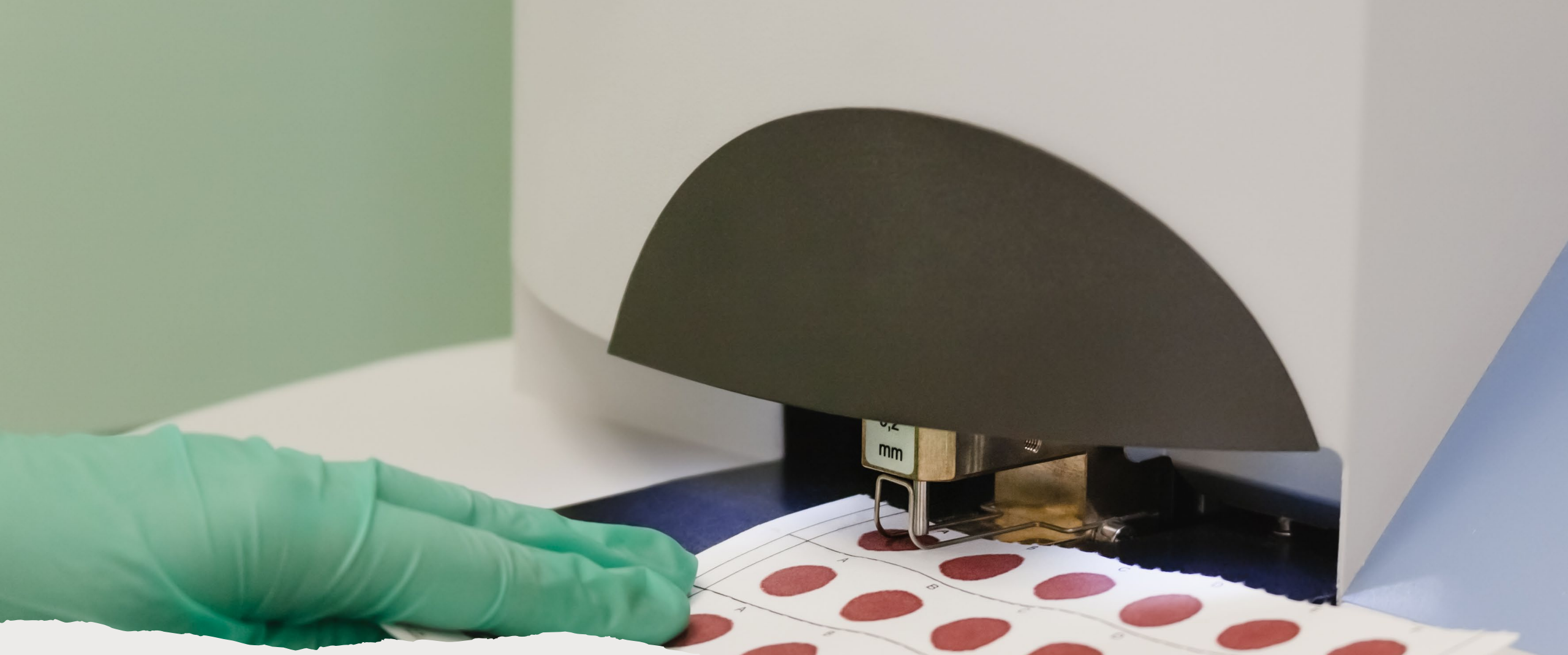
Acknowledging we need to do more

Exposing Payor Data to understand Membership – 40-45%

Ran data sets for MPS I and MPS II populations in the U.S.

Learning that most of our Members are White Caucasian, some Hispanic, and fewer in our Black Community

National MPS Society
Support for Families. Research for a Cure.



Pushing the Envelope in Evidence-base Criteria to Demonstrate Benefits of Newborn Screening Programs

NASEM | Next-Generation Screening The Promise and Perils of DNA Sequencing of Newborns at Birth | 6.7.2023

National MPS Society
Support for Families. Research for a Cure.

Acknowledging Difficult Learnings – Dec. 2022

- MPS are genetic diseases
- Child 1 diagnosed with outside of NBS; 6 years without therapies
- Child 3 – first child caught in district with NBS
- Child 3 – moved immediately towards optional therapies/HSCT
- Child 1 – now on therapy
- Different trajectories for the children

Gaps in Outreach We Must Bridge in 2023

Pilot Outreach Atlanta, GA 2022

Revamp Family Regional Gatherings in 2023

Moving into five inner-city areas in 2023

- Bergen, NJ; Cleveland, OH, Los Angeles, CA, San Antonio, TX, Denver, CO

4. Outreach program with Hospitals, care centers, and local families

5. Provide our Social Workers, Adults with MPS, Diverse Board/Staff, and

Translators for our Hispanic population

National MPS Society
Support for Families. Research for a Cure.

How will we measure effectiveness? Outcomes?

- What will mean a good outcome?
- How can we ensure that we do not know everyone in a clinical trial?
- How do we change our forward-facing education materials?
- Embracing hard is crucial – we are not fulfilling our mission completely if we do not address the disparities within follow-up care and education in our diverse populations.
- Newborn Screening is health equity for babies; follow-up care and education is critical.

National MPS Society
Support for Families. Research for a Cure.

Thank you!

National MPS Society
Support for Families. Research for a Cure.

References

Goldenberg, Aaron. *Including ELSI research questions in newborn screening pilot studies.* Genetics in Medicine, American College of Medical Genetics and Genomics. (2019), 21.

Heeju Sohn and Timmermans, Stefan. *Inequities in newborn screening: Rrace and the role of Medicaid.* SSM-Population Health. 2019, 9.

Herbst, Z.M.; Urdaneta, L.; Klein, T.; Fuller, M.; Gelb, M.H. *Evaluation of Multiple Methods for Quantification of Glycosaminoaminoglycan Biomarkers in Newborn Dried Blood Spots from Patients with Severe and Attenuated Mucopolysaccharidosis-I*. *Int. J. Neonatal Screen.* **2020**, *6*, 69.

Newborn Screening as a Public Health Program. Available from: https://www.ncbi.nlm.nih.gov/books/NBK52731/

NC Pilot screening project - http://ncnewbornscreeningpilots.org/MPS-I/index.php

NewSTEPS: The Establishment of a National Newborn Screening Technical Assistance Resource Center, J Ojuda; Dec 2019

RUSP application - www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html

Taylor, J.L. & Lee, S. *Lessons Learned from Newborn Screening in Pilot Studies.* NC Medical Journal, Vol. 80, No. 1, 55.

National MPS Society
Support for Families. Research for a Cure.