GAUCHER DISEASE: A CASE STUDY OF GEOGRAPHIC DISPARITIES IN NEWBORN SCREENING

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NEWBORN SCREENING IN THE US

Early detection tool:

Newborn screening (NBS) identifies life-threatening but treatable disorders soon after birth, enabling timely intervention and better health outcomes.[1]

Historical growth:

The first NBS test, for phenylketonuria (PKU), began in 1961. By 2020, all 50 states had adopted screening panels, though the scope of testing varies widely.[1]

Ongoing disparities:

Some states now screen for nearly 70 conditions, while others test for far fewer, creating significant inequities in access to diagnosis and treatment.[2]

Uneven federal guidance:

Without consistent national standards, families' access to timely diagnosis and intervention often depends on where a child is born.[3]

Policy shifts:

In April 2025, the federal committee overseeing the Recommended Uniform Screening Panel (RUSP) was disbanded; even before then, only 13 states had adopted the full list of recommended conditions, and the approval process was criticized as slow and restrictive by the rare disease community.[3]

ABOUT GAUCHER DISEASE[4]

Disease overview:

- Gaucher disease is a lysosomal storage disorder, in the same family as Tay–Sachs disease.
- Caused by mutations in the GBA1 gene, leading to deficient enzyme activity and lipid accumulation in macrophages.
- Three subtypes:
  - Type 1 (85–90%): non-neuronopathic, variable onset.
  - Types 2 & 3: neuronopathic, with progressive neurological involvement.
- Clinical features: anemia, bone pain, hepatosplenomegaly, growth delay; severity ranges from mild to life-threatening.

Incidence and population differences:

- General population: ~1 in 50,000 births.
- Much higher among Ashkenazi Jewish populations: 1 in 350–1,200 births; carrier rate ~1 in 10.
- State pilot programs confirm variable incidence across the U.S.

Diagnosis challenges:

- 73% diagnosed within a year of first medical visit.
- 14% wait seven years or longer.
- Age at diagnosis: 37% in childhood (<10 years), 12% during adolescence.

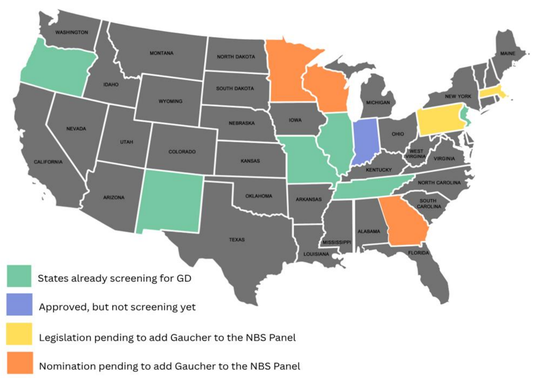
Treatment and barriers:

- FDA-approved therapies, including enzyme replacement (ERT) and substrate reduction (SRT), can significantly improve outcomes.
- Delayed diagnosis limits the effectiveness of treatment, as earlier intervention is critical to prevent irreversible disease complications.
- Barriers to care include misdiagnosis, low provider awareness, limited access to specialists, and insurance restrictions.

EQUITY THROUGH NEWBORN SCREENING AND EARLY INTERVENTION

Disparities by Population Prevalence:

Almost half of the U.S. Jewish population resides in just three states - New York, California, and Florida - yet none of these states currently include Gaucher disease in their newborn screening panels.[2,5] The table below highlights the top 10 states by Jewish population alongside their Gaucher screening status.



Top-10 States by Jewish Population (2020 Estimates)[2,5]

	State	Jewish Population (≈)	Pct. of Total US Jewish Pop	Screens for Gaucher
1	New York	1,598,000	21%	No
2	California	1,174,000	15%	No
3	Florida	778,000	10%	No
4	New Jersey	572,000	8%	Yes
5	Pennsylvania	348,000	5%	No
6	Illinois	335,000	4%	Yes
7	Massachusetts	335,000	4%	No
8	Texas	245,000	3%	No
9	Maryland	231,000	3%	No
10	Connecticut	155,000	2%	No

Gaucher NBS Results From a Sample of States[6-10]

State	Years Covered	No. Babies Screened	Gaucher Cases Confirmed	Rate of Confirmed Cases/Babies Screened Per 100K
Illinois	2015-2024	~1.3 million	21	1.62
Missouri	2013-2024	~850,000	15	1.76
New Jersey*	Jul 2019 – Dec 2023	~438,515	19	4.33
Tennessee	2018-2024	~612,500	4	0.65
New York Pilot Program**	2021-2025	~29,100	6	20.62

** 19 additional identified, lost to follow up before confirmation*
***select hospitals participated*

Early detection matters:

NBS enables timely diagnosis, allowing treatment to begin before irreversible damage occurs.

Improved outcomes:

Early and consistent treatment can significantly alter disease progression and quality of life.

Benefits of NBS include:

- Reduces diagnostic delays
- Enables treatment before symptoms worsen
- Supports proactive care planning
- Facilitates access to specialized care and support
- Expands eligibility for emerging therapies and clinical trials

Evidence:

States already testing for Gaucher, such as Missouri, Illinois, and New Jersey, demonstrate the feasibility of NBS and generate valuable data on incidence and treatment outcomes.

Accuracy of testing:

Second-tier testing (enzyme activity, lyso-Gb1 levels, *GBA* gene status) minimizes false positives.

Funding:

- Integration into existing NBS infrastructure is feasible without significant financial burden.
- Earlier diagnosis generates cost savings for both families and the healthcare system.

Support resources:

Early intervention allows families to receive guidance on care pathways, financial support, and provider access.

CONCLUSION

Geographic location should not determine health outcomes or access to preventive care.

Gaucher disease highlights how fragmented NBS standards create unequal access to early diagnosis and treatment.

Families affected by rare diseases face persistent barriers to timely care, compounded by inconsistent state and federal policies.

Expanded state programs and stronger, federally guided, patient-centered frameworks can ensure equity nationwide.

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REFERENCES

Please scan the QR code below to access the full list of literature cited.

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