GM1 Gangliosidosis Must Be Added to Statewide Newborn Screening in California

Urgent Request for Legislation Supporting CA Pilot Program of Newborn Screening for GM1 Gangliosidosis

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What is GM1 Gangliosidosis?

GM1 is rare, inherited lysosomal storage disease caused by a deficiency of beta-galactosidase. This leads to toxic accumulation of GM1 ganglioside in the brain and body.

- **Most cases affect babies:** Symptoms typically appear in the first 6 months of life.
- **Universally fatal:** GM1 is rapidly progressive with most children dying between ages 2 and 4.
- **No FDA-approved treatment** and **not included on the Recommended Uniform Screening Panel**.

A Diagnostic Crisis

Without newborn screening, families face a devastating diagnostic odyssey with months or years of specialist visits and misdiagnoses. **By the time a diagnosis is made, irreversible neurological damage has already occurred, and every intervention window has closed.** What's more, because GM1 is so rare, it will be nearly impossible to gather enough data to add it to the federal Recommended Uniform Screening Panel despite the fact that it meets the requirement of causing severe health issues or death if not treated early.

A Catch-22 for GM1 Families

- Without early identification, trials deeply struggle to enroll patients to illustrate a treatment works.
- Without trial data, treatments cannot receive approval.
- Without an approved treatment, NBS stalls.

We should not penalize babies because no treatment exists. Screening could help us find one.

The Treatment Pipeline *Is* Advancing

- **Enzyme Replacement Therapy (ERT):** Cure GM1 is funding the first-ever intracerebroventricular ERT for GM1 (modeled on FDA-approved Brineura for CLN2 Batten disease). ERT is the approach most likely to benefit infantile patients identified at birth.
- **Small molecule therapy:** Two drugs in trials; regulatory filings anticipated 2026.
- **Gene therapy:** Clinical trials ongoing; fetal gene therapy trials in planning.

NBS must exist before these treatments are approved to prevent needless pain and suffering.

We Must Screen Statewide for the Patient Pool to be Large Enough

GM1 is so rare that existing research pilots may not identify a single U.S. patient.

- New York's ScreenPlus pilot already includes GM1, but no U.S. patients have been identified because the pilot is not large enough.
- Florida, North Carolina, and other state pilots face the same limitation.
- California has ~420,000 births per year, which is the largest birth cohort of any state. Only statewide California screening can find GM1 babies at a rate that supports trial enrollment and treatment access.

A pilot is not enough for GM1. Only statewide screening generates the volume needed.

The Validated NBS Assay is Ready

Cure GM1 Foundation funded the first-ever NBS assay for GM1 (developed by Dr. Michael Gelb at the University of Washington).

- **Runs on existing mass spectrometry** equipment already used in California state labs (no new infrastructure required)
- **Two babies were identified in a pilot study** in Turkey, which proves the assay finds real patients when deployed at meaningful scale.
- **The assay was validated in multiple NBS studies** including New York's ScreenPlus program.
- **GM1 can be easily added to an existing metabolic panel** at a marginal cost

Why California, Why Now?

- Screening in California will generate data that can change the national landscape.
- Traditionally, California has been a leader in newborn screening; it's time to move faster and add non-RUSP diseases to the screening list.
- California is home to the Cure GM1 Foundation (Albany). **Multiple California families are living this crisis right now.**
- Every year that screening does not exist is another year families will suffer.

The assay is ready. The science is established. The precedent is set. We need the political will to act.

WE ASK CALIFORNIA TO ADD GM1 GANGLIOSIDOSIS TO STATEWIDE NEWBORN SCREENING.

LEARN MORE: Newborn screening: curegm1.org/nbs-gm1 · ERT program: curegm1.org/ert-project