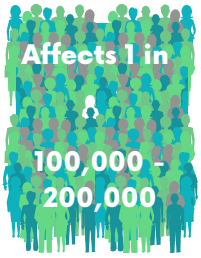
GM1 GANGLIOSIDOSIS

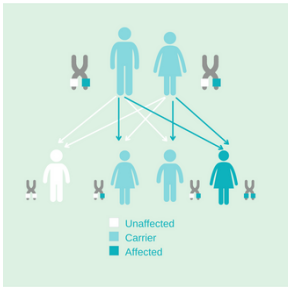
A FATAL GENETIC DISEASE, MISTAKEN FOR DEVELOPMENTAL DELAY, CAUSING A DIAGNOSTIC ODYSSEY



Help accelerate diagnosis, increase awareness, and improve quality of life

GM1 GANGLIOSIDOSIS OVERVIEW

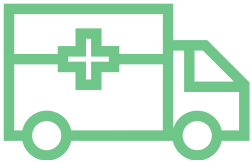
GM1 gangliosidosis (GM1) is a rare, autosomally inherited genetic disorder that primarily affects children. Over 50% of those impacted die before their fifth birthday. It has many debilitating ramifications, some of which are developmental regression, mobility deterioration, seizures, visual impairment, and neurodegeneration.



autosomal recessive inheritance

THE IMPACT

- Death in most forms of the disease
- Progressive worsening of symptoms
- Ineligibility for clinical trials due to delayed diagnosis



TREATMENTS & RESEARCH

As of yet, there is no approved treatment for GM1 gangliosidosis, but there are ways to manage symptoms via medications, physical and occupational therapy, and palliative care. Research into enzyme replacement therapy, gene therapy, and small molecules is ongoing. Early diagnosis is still helpful. We recommend genetic testing, screening of the GLB1 gene, and a lysosomal storage disease enzyme screening panel.

SIGNS & SYMPTOMS

Symptom Severity ⟶ Enzyme activity Age of onset

TYPE 1 INFANTILE

most common

- severe hypotonia by six months of age
- enlarged spleen, liver
- enlarged head
- inability to sit, roll
- loss of head control
- macular cherry-red spots
- nystagmus
- heart failure, cardiomyopathy
- dystonia
- skeletal abnormalities
- coarse facial features
- hearing impairment
- breathing difficulties
- in utero development non immune hydrops fetalis

TYPES 2A & 2B LATE INFANTILE & JUVENILE

note loss of skills

- hypotonia, muscle weakness
- loss of speech, slurred speech
- impaired eyesight, strabismus, nystagmus
- clumsiness, falling, lack of coordination
- loss of fine and gross motor skills
- cognitive impairment, intellectual disability
- cerebral atrophy
- gait disturbance, ataxia
- seizures
- lethargy
- scoliosis, flattened vertebrae
- dystonia

TYPE 3 ADULT ONSET

least common

- dystonia
- speech disturbance
- imbalance; gait disorders
- ataxia
- uncoordinated movements
- speech problems
- visual problems
- vertigo
- slowly progressive dementia
- intellectual degradation
- skeletal dysplasias that can include spine deformities
- skeletal muscle atrophy
- most reported cases are in patients of Japanese origin

ABOUT CURE GM1 FOUNDATION

Cure GM1 is dedicated to GM1 gangliosidosis research, drug development, awareness, and advocacy. If you believe your patient or loved one may have GM1, please direct them to our website, curegm1.org. We have information about clinical trials and preclinical research as well as provide access to an engaged community.

CUREGM1.ORG