# An Open-label, Phase 1/2 Study to Evaluate the Safety and Efficacy of Single-dose PR001A in Infants with Type 2 Gaucher Disease

**Status:** Recruiting

## Eligibility Criteria

**Sex:** All
**Age:** up to 24 Months old
This study is NOT accepting healthy volunteers

**Inclusion Criteria:**

• Bi-allelic GBA1 mutations consistent with a diagnosis of GD2 confirmed by the central laboratory.

• Clinical diagnosis of GD2

• Parent/legal guardian has the ability to understand the purpose and risks of the study and provide written informed consent and authorization to use protected health information in accordance with national and local privacy regulations.

• Patient has a reliable informant (i.e., parent/legal guardian) willing and able to participate in the study as a source of information on the patient's health status and cognitive and functional abilities (including providing input into the rating scales).

**Exclusion Criteria:**

• Diagnosis of a significant CNS disease other than GD2 that may be a cause for the patient's GD symptoms or may confound study objectives.

• Achieved independent gait.

• Severe peripheral symptoms of GD which, in the opinion of the Investigator, would pose an unacceptable risk to the patient or interfere with the patient's ability to comply with study procedures or interfere with the conduct of the study.

• Concomitant disease, condition, or treatment which, in the opinion of the Investigator, would pose an unacceptable risk to the patient or interfere with the patient's ability to comply with study procedures or interfere with the conduct of the study.

• Use of any GD treatment-related substrate reduction therapy.

• Use of strong inhibitors or inducers of cytochrome P450 3A4 (CYP3A4) or P-glycoprotein (P-gp) medications, herbals, or over-the-counter agents.

• Any type of prior gene or cell therapy.

• Live vaccine Immunizations within 4 weeks, or non-live vaccines within 2 weeks prior to the start of required immunosuppressive regimen.

• Use of blood thinners. Antiplatelet therapies are acceptable if the patient is medically able to temporarily stop them from 7 days prior to dosing and through at least 48 hours after the intracisternal injection and lumbar puncture.

• Use of systemic immunosuppressant or corticosteroid therapy other than protocol-specified (topical or inhaled preparations for dermatological conditions or asthma are allowed).

• Participation in another investigational drug or device study within the past 3 months.

• Brain MRI (magnetic resonance imaging) and MRA (magnetic resonance angiography) showing clinically significant abnormality deemed a contraindication to intracisternal injection.

• Clinically significant laboratory test result abnormalities assessed at screening.

• Contraindications or intolerance to radiographic visualization methods (e.g. MRI, MRA, CT), and intolerance to contrast agents used for MRI or CT scans.

• Contraindications to general anesthesia or sedation. Other protocol-defined inclusion/exclusion criteria may apply.

## Conditions & Interventions

**Interventions:**
Biological: LY3884961, Drug: Methylprednisolone, Drug: Sirolimus, Drug: Prednisone
**Conditions:**
Gaucher Disease, Type 2
**Keywords:**
Gaucher Disease, GD, Gaucher, Type 2 Gaucher, Neuronopathic Gaucher, nGD, AAV9, GBA, Gene Therapy, Glucocerebrosidase, GBA1 mutation, Infants

## More Information

**Description:** This is a study to assess the safety and efficacy of PR001A, an Aden-associated (AAV9) viral vector to treat neuronopathic Gaucher disease type 2 (GD2) in infants. PRA001A will be administered via suboccipital injection to the cisterna magna during a single neurosurgical session. GD2 is a fatal disease of early infancy that does not have any therapeutic options beyond palliative care. This study will enroll infants 0-24 months of age.
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**Phase:** Phase 1/Phase 2
**IRB Number:** STUDY00008823
**System ID:** NCT04411654