

# GLB1-related Morquio B Disease and GM1 gangliosidosis community: We want to hear from you!

Researchers across the globe are studying treatments to improve care for individuals with GLB1-related Morquio B Disease and GM1 gangliosidosis. These clinical trials and research projects need to measure appropriate outcomes for these disorders.

## What is GLB1-related Morquio B Disease (GLB1-related MBD)?

- GLB1-related MBD is a "storage disease" where individuals with MPS lack the ability to produce sufficient amounts of the enzyme beta-galactosidase. This enzyme is crucial for breaking down undesirable substances in their bodies. Due to the deficiency of this enzyme, these substances accumulate over time, leading to a worsening of the condition.
- There are numerous symptoms that people with GLB1-related MBD can have, especially problems with bone and muscle development that can cause severe pain and mobility issues.

## What is GM1 gangliosidosis (MBD-GM1)?

- People with MBD-GM1 also lack the ability to produce sufficient amounts of the beta-galactosidase enzyme, which causes an accumulation of undesirable substances in their bodies.
- There are three types of MBD-GM1 identified by the medical community; infantile (type I), juvenile (type II), and adult (type III).
- Symptoms in individuals with MBD-GM1 can present differently than those in GLB1-related MBD, and usually includes a range of neurodevelopmental issues such as speech, cognitive, and/or motor functioning, in addition to skeletal and mobility issues.

## What are outcomes?

- Outcomes refer to various aspects that can be observed or measured to assess the effectiveness of a treatment.
- These aspects include how a person feels, what they are able to do, any observable symptoms they experience, or the results obtained from laboratory tests.

## What is a Core Outcome Set (COS) and why is a COS used?

- A 'Core Outcome Set' is a small group of outcomes that are considered important and should be consistently measured in every research study focused on a particular disease.
- The purpose of a COS is to ensure that studies do not overlook key outcomes or measure irrelevant ones related to the specific disorder.

## Why is a COS important?

When researchers measure different outcomes in studies, it is difficult to compare and evaluate the effectiveness of different interventions or treatments. A COS helps standardize the outcomes measured across studies, enabling meaningful comparisons and identification of studies and determining the best treatment options.

Clinical trials are exploring therapies for GLB1-related MBD and MBD-GM1; however, their potential impact remains uncertain as these trials are primarily conducted in animal or cell models but not on humans models.

### **How can I contribute?**

Researchers are developing a core outcome set for GLB1-related MBD and MBD-GM1 and we need your input—we want to know which outcomes are important to you!

In a focus group session, you will be invited to share the outcomes related to GLB1-related MBD and MBD-GM1 most important to you. As a rare disease community, we need as many patients and parents/caregivers as possible to participate!

### **What is a focus group?**

A focus group is a small group of 6-10 people who participate in an open discussion led by a facilitator. The goal of the focus group is to gather the various opinions and ideas from as many different people in the time allotted.

Focus groups are structured around a set of carefully predetermined questions; however, the format allows for the freedom to discuss ideas related to these questions. For our study, we will be covering six questions about GLB1-related MBD and MBD-GM1 disease and outcomes that you think should be measured in clinical trials and research studies.

For more information, please visit our supplemental focus group guideline PDF.

### **Who is eligible?**

Any individuals diagnosed with GLB1-related MBD and MBD-GM1 (Type II or III).

A parent or adult caregiver of a person diagnosed with GLB1-related MBD and MBD-GM1 (Type II or III).

### **What is the time commitment? Will I be compensated?**

You will be asked to take part in a 50-minute information session and a 100-minute focus group session. Yes, you will be compensated for your participation with a \$100 (USD) Amazon online gift card.

### **How can I participate?**

Recruitment will take place from March to May, 2024. Fill out our short form to see if you are eligible to participate.

### **Questions?**

Email us at [gm1cos@bcchr.ca](mailto:gm1cos@bcchr.ca)

### **Where can I learn more?**



**Principal Investigator:** Dr. Sylvia Stockler

Professor Pediatrics, Department of Pediatrics, UBC

Clinical Biochemical Geneticist, Division of Biochemical Genetics, BCCH

**Research Assistants:** Zahra Nasser Moghaddam, Maria Bleier, Jasmine Li