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?
You will be asked to take part in a 50-minute information session and a 100-minute focus group session.

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

Where can I learn more?

Principal Investigator: Dr. Sylvia Stockler
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