About Us

The Manton Center for Orphan Disease Research is a philanthropically funded center at Boston Children's Hospital that is dedicated to rare disease research.

The Gene Discovery Core (GDC) is a genetic research study within the Manton Center that focuses on learning more about rare and undiagnosed genetic conditions.

We work with patients and families at Boston Children's Hospital and across the world who are searching for a diagnosis or a greater understanding of their rare disease.

Our Participants and Collaborators Around the World!

Contact Us

For families interested in enrolling and for providers who wish to refer patients to our research study, please reach us at:

617-919-3378

GDC@childrens.harvard.edu

For more information, visit **www.childrenshospital.org/mantoncenter**

Interested in donating to our research? **Visit www.childrenshospital.org/SupportTheGDC**

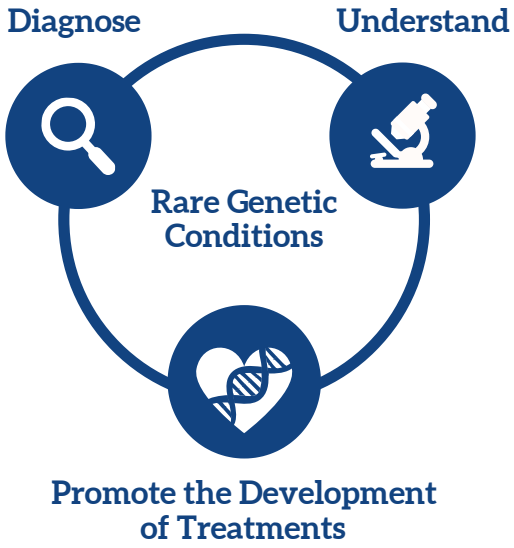
The Manton Center for Orphan Disease Research

Gene Discovery Core

Boston Children's Hospital
Until every child is well™

Our Mission

At the GDC, our goals are to:



The knowledge gained from this research will help families find diagnoses and will contribute to scientific advancements that improve human health. The information discovered will lead to a better understanding of rare diseases and assist in the development of treatments.

By collecting DNA and combining genetic information with clinical information, the GDC will serve as a valuable genetic library that scientists at Boston Children's Hospital and their collaborators around the world can use to study rare diseases.

About Our Research Study

Who We Enroll

We enroll participants of any age with a rare or unknown diagnosis and their family members. Enrolling parents and siblings can be helpful in finding a diagnosis but is not required for participation.

Research Opportunities

For those seeking a diagnosis, our research program may be able to complete genetic studies on a research basis. If relevant results are found, we can work with your healthcare providers to confirm the results with a clinical test.

Genetic studies may include:

Whole Exome/Genome Sequencing (WES & WGS) - Genetic tests that sequence DNA with the hopes of identifying causative gene(s).

Genetic Reanalysis - Involves reviewing existing genetic data for those with non-diagnostic clinical testing.

Follow-up Studies - Involves working with your physician and other scientists to better understand an unclear genetic result.

How to Enroll

Enrollment includes:

- Written and verbal consent (phone call or in-person meeting) for each family member participating
- Questions about medical and family history/sharing relevant medical records
- DNA samples (blood or saliva)
- If applicable, access to tissue sample(s)
- Collaboration with your/your child's clinicians to assist in the enrollment and research process

Enrollment can be done by mail, email, and phone, so no trips to BCH are required for enrollment or study participation. There is no cost to participate.