rare genomes project

For your physician

The Rare Genomes Project is a research study at The Broad Institute of MIT and Harvard, a non-profit academic research institution focused on using genomic data to better understand human health and disease. Our team, led by investigator Heidi Rehm, has a specific focus on understanding the genetic basis of very rare and undiagnosed conditions. Our mission is to partner directly with patients, families, advocates, and clinicians and to increase patients' access to genomic research.

How it works

The Rare Genomes Project provides undiagnosed families with the opportunity to participate in a genomic research study, regardless of where in the United States they live. To ensure that participation is made broadly available to any eligible family, all study procedures can be completed from, or nearby, the patient's home.

Interested participants and families can visit www.raregenomes.org to learn more about the project. By clicking "Start an Application," they can apply to participate. Our clinical team will review each application and eligible families will be invited to enroll.

We will send a collection kit to enrolled families who can choose to have blood drawn at a Quest Diagnostics Patient Service Center (www.questdiagnostics.com/appointment) or at another phlebotomy location of their choice. In some cases, we may be able to request DNA from prior genetic testing. The sample collection kits will be mailed back to our lab, where DNA extraction and genomic sequencing will be performed.

Our team will analyze genomic data alongside each family's clinical information with the goal of identifying the genetic basis of the condition present in the family. If we are able to identify the cause of disease, we will re-contact the family and pursue clinical confirmation in partnership with a local provider of their choosing. The clinical confirmation report will be issued to the local ordering provider.

This project is dedicated to rapid and open data sharing. De-identified genetic and clinical data generated in this project will be shared broadly with the wider rare and undiagnosed community, as well as other clinicians and researchers, through controlled-access genomic databases, as well as the Matchmaker Exchange and Clinvar.

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Your role in the project

We are interested in partnering with clinical providers of participants to facilitate the return of medically-relevant, clinically-confirmed results discovered in this project.

Other ways that you could be involved:

- Aggregating and summarizing the family's medical records and clinical information
- Addressing follow-up questions that arise during the family's involvement in the project

Eligibility Criteria for Families

Patients and families with a rare and genetically undiagnosed condition who:

- Live in the United States
- Are currently under the care of a doctor that is helping to understand the cause of the patient's condition

At this time, we are unable to include patients and families that:

- Have a suspected or known environmentally caused condition
- Have any condition not likely to be monogenic
- Have a genetic diagnosis that fully explains the patient's condition.

How to refer a family

If you currently provide care for families that you think would be a good fit for participation in this project, please refer them to our website www.raregenomes.org. We also partner directly with clinicians, hospitals, and institutions to increase awareness about the project. If you would like us to provide brochures and informational materials for patients that visit your clinic, please contact us.

Thank you for your support. We look forward to learning with you!

Contact Us

www.raregenomes.org
617-714-7395
Toll-free: 855-534-4300