rare genomes project

For your physician

The Rare Genomes Project is a research study housed 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 investigators Daniel MacArthur and Heidi Rehm, has a specific focus on understanding the genetic basis of very rare and undiagnosed conditions. Our mission in the Rare Genomes Project is to partner directly with patients, families, advocates, and clinicians like you, to speed the rate of rare disease diagnosis and 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 "Count Me In," visitors to the site will be directed to complete a survey so that our team can begin to understand the condition affecting the family. Our clinical team will review each submission and re-contact eligible families for participation. A videoconference will be scheduled to discuss the project in more detail; families interested in participating will electronically sign the informed consent. As part of the consent process, families will sign a medical records authorization so that our team can access a copy of the participant's medical records.

We will send a collection kit to enrolled families who can choose to have their blood drawn at a Quest Diagnostics Patient Service Center (www.questdiagnostics.com/appointment) or by a phlebotomist of their choice (including at their local physician's or hospital's lab). Saliva kits are also available for participants who are unable to have a blood draw. The sample collection kits will be mailed back to the Broad Institute Genomics Platform, where DNA extraction and DNA sequencing will be performed.

rare genomes project

For your physician

Our team will analyze genomic data alongside each family's clinical and personal story 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 request a second sample for clinical validation, which will be performed at the Partners Laboratory for Molecular Medicine. A clinical report describing our findings will be provided directly to the family and the family's doctor.

This project is dedicated to rapid and open data sharing. 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.

Eligibility criteria for families

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

- Understand English
- 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 known not to involve a genetic link
- Have a genetic diagnosis that fully explains the patient's condition.

How you can help

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.

Your role in the project

We are interested in partnering with enrolled families' clinicians to facilitate the return of genomic clinically-confirmed research results discovered in this project.

Other optional 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.

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

Contact us

raregenomes@broadinstitute.org
617-714-7395