rare genomes project

Making genomic research accessible to the rare disease community

Who We Are

The Rare Genomes Project is a patient-driven research study led by genomics experts and clinicians who believe that the latest advances in genomic sequencing are changing medicine, and that they should be accessible to families with rare and undiagnosed conditions.

What We Do

Sequence families

We will use genomic sequencing to look for the cause of the disease in your family. If we find a genetic change that may be the cause of the rare disease, we will confirm it in a clinical lab and work with your doctor to return the result to you.

Generate new discoveries

The genetic information generated in this project will be shared with other scientists to ensure it has the widest possible impact.

Empower patients as partners

We believe that families should be at the center of research involving their diseases. We encourage family engagement throughout the study.

Is the Rare Genomes Project right for me?

Our project is looking to enroll patients who:

- Have a condition that is suspected to be caused by a change in a single gene
- Do not yet have a genetic diagnosis
- Are currently under the care of a clinician
- Are English- or Spanish-speaking
- Live in the United States

What does participation involve?

Participation is remote and free of cost. Eligible families will be asked to donate a blood sample using a collection kit mailed to their home and provide relevant medical records.

How can I get involved?

Visit www.raregenomes.org to learn more about this study and tell us your story by filling out an online application.

Questions? Contact Us
raregenomes@broadinstitute.org
617-714-7395

Thank you to awareofangels.org and participating families for sharing their photos.