

Thank you for participating in the Rare Genomes Project!

We are asking you to obtain and send along your family's health information so that we can better evaluate your condition. Sharing your medical history will help us learn more about your family. Your family's health information will be used along with the genomic data generated from your family's DNA or tissue samples, so we can best understand the rare genetic condition present in your family, as well as rare genetic conditions more generally.

Who in my family needs to provide medical history?

Only family members that have signed a consent form to participate in the Rare Genomes Project should share their health information. If you have any questions about who in your family should share their health information, please ask us.

For family members who do not have signs of the suspected undiagnosed condition: no records are needed unless otherwise specified.

For family members who have signs of the suspected undiagnosed condition, we will ask for the following medical records:

- **Geneticist or other Specialist consultation notes**

- A copy of a clinic note from the first visit with the patient's geneticist or primary specialist, AND notes from the most recent clinic visit.
 - This should be from the clinician who is sending genetic testing or thinking about the underlying cause of the medical condition. If you have multiple physicians who fill this role, we are happy to receive clinic notes from multiple physicians.

- **Previous genetic testing results reports**

Please provide reports for any previously performed genetic testing. This may include:

- Karyotype/Chromosome analysis
- Microarray
- Gene Sequencing Tests (single gene and/or panels)
- Mitochondrial genome sequencing

- ♦ Whole exome sequencing (clinical or research)
- ♦ Whole genome sequencing (clinical or research)
- ♦ (Optional) Biochemical and/or metabolic testing (particularly if abnormal)

Other helpful records include:

- Additional clinical/summary notes.
- MRI imaging reports (particularly if abnormal).
- Pathology reports and information about stored tissue samples (particularly if abnormal).
- Photos (these should be provided only after signing our Photo Consent Form).
- Family history or family pedigree drawing.
- Most recent hospital admission note (if you had a recent hospitalization).
- Pediatric growth charts (if abnormal).
- Hospital discharge summary (if you had a recent hospitalization).

Please have records sent to us:**By Mail:**

Rare Genomes Project
Broad Institute of MIT and Harvard
415 Main Street
Cambridge, MA 02142

By fax:

(617) 800-1909

OR

Is there any additional information my family can provide?

Yes. If your family has any of the following, we will work with your hospital and pathology lab to obtain this information. Please provide the following information to the best of your abilities:

- **Stored tissue samples**
 - ♦ What is the tissue type?
 - ♦ At which facility was the biopsy completed?
 - ♦ What date was the biopsy completed? (Please be as specific as possible.)
 - ♦ Where is the biopsy stored?
 - ♦ How is the biopsy stored (frozen or fixed in chemicals)?
 - ♦ Please provide the contact information for the storage facility (Phone, Email, Contact Person, if available)
- **MRI brain images (on a CD).**
 - ♦ Please provide the images if the study was found to be abnormal or the condition is neurologic.

If my family sees multiple doctors or specialists, who should I contact for medical records?

Please request medical records from the specialist with whom you follow most closely through your health care facility's medical records office.

Can the Rare Genomes team reach out to my family's physicians on my behalf?

Yes. We would be able to reach out to your medical facilities for your medical history with a signed authorization from each hospital or medical center that provided care for your family. Please let us know if you would like us to do so and we would be happy to help.

How will my medical history help in this research?

Geneticist/Specialist consultation notes

Notes from your geneticist and/or specialist(s) will provide us a broad description of the symptoms and conditions you and/or your family member(s) have experienced due to the suspected genetic condition present in your family. This information, utilized

together with your family's genomic data, helps to build a more complete image of the condition present in your family and help us compare the similarities and differences across participants.

Previous genetic testing results or previous research genetic sequencing data

Previous genetic testing and research data could provide us with both specific and general details of what has been previously considered for your condition. Specific testing results could provide us with a better idea of what known conditions have been considered that are similar to your condition, yet have been ruled out. This could help us better characterize your condition – allowing us to compare and contrast between what is known and unknown.

MRI imaging reports and MRI brain images

MRI imaging can provide a visual of if/how your condition has affected the tissues and organs in your body. MRI imaging of the brain is particularly useful for providing more information if your condition involves any abnormal findings.

Tissue samples

Stored tissue samples are an invaluable resource for both the diagnosis and understanding the relationship between a genetic change in your DNA and the effects that change can have on structure and function of your tissues. In some cases, if a change in your DNA is rare or has never been seen before, we can obtain and study the effects on your tissue to help us understand if and how the change is causing your condition.

Pathology reports

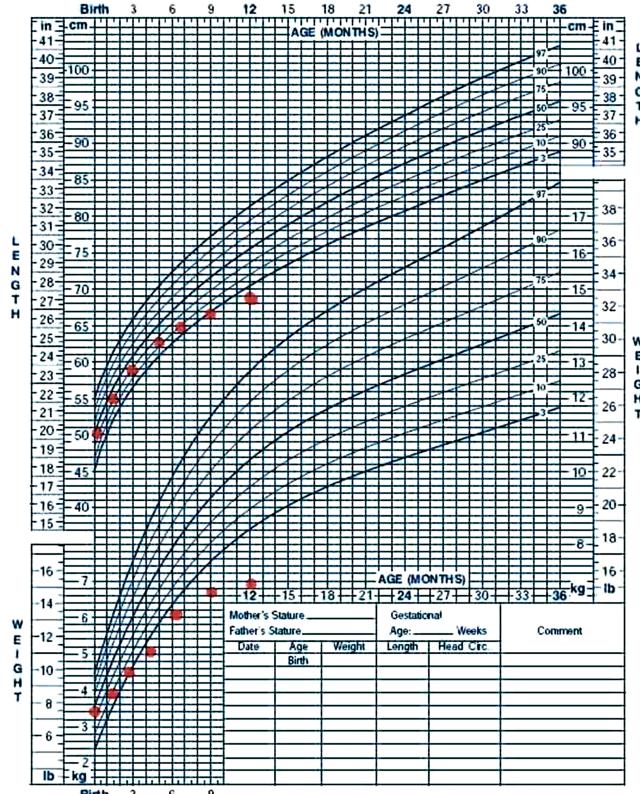
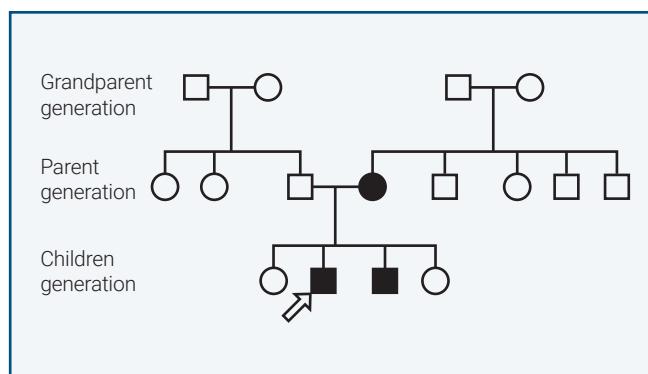
Pathology reports provide an evaluation of your tissues obtained through a biopsy. The report contains key descriptions characterizing how your tissues have been affected in terms of tissue amount, size, and structure. In the event that your stored tissue samples are not available, this report will be critical in helping us characterize and evaluate if/how your tissues have been affected by your condition.

Photos (only after signing our Photo Consent Form)

Photos could provide us with a visual image of how the condition has affected the individual(s) with rare disease. Additionally, some genetic conditions will lead to the development of specific physical characteristics, which can be very helpful in diagnosing certain rare conditions.

Family history or family pedigree drawing

Family history or family pedigree (shown below on left) could provide information to map out how the condition has affected different members of your family. Some genetic conditions are known to have certain inheritance patterns throughout a family tree. These are often drawn by a medical geneticist or genetic counselor and are helpful to share if it already is part of your family's medical record. We do not expect you to create one of these if it is not part of your family's medical record.



Pediatric growth charts (if abnormal)

Growth charts are used to document progression in height, weight, head circumference, and body mass over time.



Guide for Sharing Your Family's Health Information

Hospital admission notes and Hospital discharge summaries

Some symptoms may suddenly present themselves and lead to a required hospital stay. Thus, hospital admission notes and discharge summaries often provide a useful summary of information regarding the incidental situation that may be related to the condition. Hospital admission notes provide a baseline of your health condition at time of admittance, while the discharge notes will describe your health status upon leaving the hospital. Please note that we are requesting the discharge summary that is written by your care team and sent to your doctor, not the patient discharge instructions that are given to you before you leave the hospital.

Thank you! Together, we can learn more about the rare disease in your family.

Questions? Call us: 617-714-7395 or email raregenomes@broadinstitute.org.