Guide for Sharing Your Health Information

Thank you for participating in the Rare Genomes Project!

We need copies of medical records so that we can learn more about your medical condition. This health information will be used along with the genomic data generated from your family's blood or tissue samples to help us study the rare genetic condition present in your family, and rare genetic conditions more generally.

Who in my family needs to provide medical information?

Only family members that have the medical condition and who 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.
  - These 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 would like to receive clinic notes from each of them.

- 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)
- Biochemical and/or metabolic testing (particularly if abnormal)

Other helpful records include:

- Additional clinical notes
- MRI imaging reports (particularly if abnormal)
- Pathology reports and information about stored tissue samples
- Photos
- Family history or family pedigree drawing
- Pediatric growth charts (if abnormal)

Please have records sent to us:

By Mail:

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

OR

By fax:

(617) 800-1909

Is there any additional information I can provide?

Yes. If you have had any of the following, please provide the following information to the best of your ability:

- Stored tissue samples
  - What is the tissue type?
  - Where was the biopsy performed?
  - What date was the biopsy performed?
  - What is the contact information for the facility with the sample?

- MRI brain images (on a CD).
  - Please provide the images if the results were abnormal or the undiagnosed condition is neurologic.

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

Yes. We can request copies of your medical records if you sign a medical record release form for each doctor or medical facility that provided care for you. 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 detailed description of the symptoms that are suspected to be due to the suspected genetic condition present in your family. This information, 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

Previous genetic testing helps provide us with both specific and general details of what has been previously considered as a cause for your condition. They provide us with a better idea of what known conditions have been considered yet have been ruled out.

MRI imaging reports and MRI brain images

MRI reports provide information about whether your condition has affected tissues and organs in your body. MRI images of the brain are useful for review by our team, especially if there are any abnormal findings.

Tissue samples

Unfixed, frozen tissue samples from medical procedures can be an additional way for us to understand how a genetic changes might be affecting the tissues of the body impacted by the disease.

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 interms of tissue amount, size, and structure. In the event that your stored tissue samples are not available, this report can help us understand how your tissues may be affected by your condition.

Photos

Some genetic conditions are known to cause characteristic or subtle facial features that we consider when determining if a participant's condition is related to a genetic variant. Photographs are considered part of the patient's medical record and will not be shared externally without separate permission.

rare genomes project

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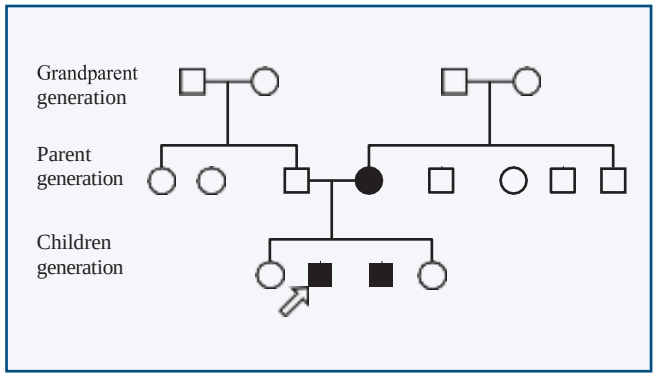
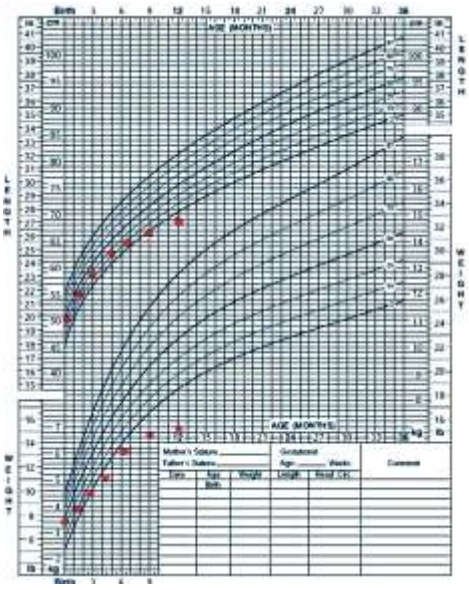
Family history or family pedigree drawing

A family history or family pedigree drawing (shown below on left) shows the structure of your biological family and indicates the medical conditions present in each person. Having this information is helpful since genetic conditions can pass through families in different ways, or in some cases, can arise anew in an individual. These are often drawn by a medical geneticist or genetic counselor and are helpfulto share if it already is part of your family's medical record. There is no need for you to create one of these if it is not part of your medical record.

Pediatric growth charts (if abnormal)

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





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.