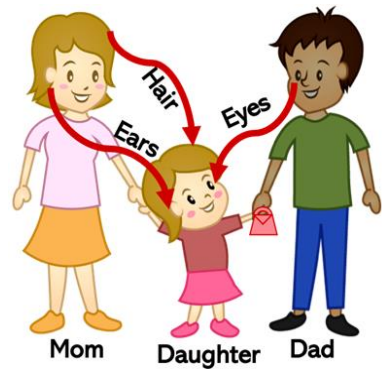


## What is DNA?

Our genetic information, or DNA, is a molecule that contains code for our body's functions. Specific parts of our DNA are called 'genes'. Genes provide instructions for carrying out specific functions of the body. For example, some genes are important in informing the proper function of our kidneys, while others are important for normal function of our lungs.



Our DNA is inherited, or passed down from parents to their children. We inherit half of our DNA from each parent. This means we have two copies of each gene, one copy passed down from our mother and the second copy from our father.

## What is genetics research?

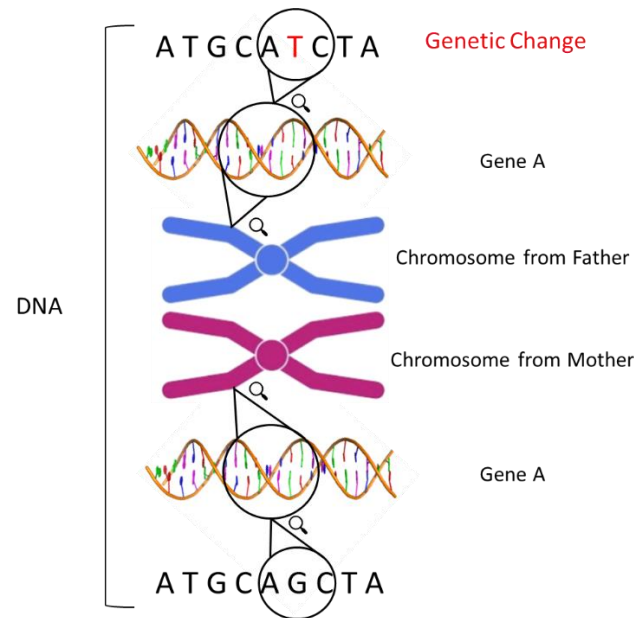
The purpose of genetic research, like this study, is to gain **NEW** knowledge about genetic factors that can help doctors treat patients better in the future.

By participating in a research study, you are contributing to the knowledge about your illness. This helps to improve health care for future generations but in most cases, you will not benefit directly from participating.

Sometimes genetic research may uncover information that may be relevant to your health, but such information would have to be verified by **clinical genetic testing**.

## What is clinical genetic testing?

Clinical genetic testing is a medical test that looks for known genetic diseases. When clinical testing is done, it is associated with your personal information and its results will be added to your medical record. You can always pursue clinical testing separate from this research study.



## Who can participate in this research study?

People or family members of people with medical a problem that may be due to genetic factors can participate (such as heart, liver, endocrine, lung, or kidney conditions). Children and adults are eligible for this study.

## What does participating in genetics research involve?

- Sharing your personal and family health history
- Allowing us to review your medical records
- Giving a DNA sample (usually blood)
- We may ask for other samples or imaging to help understand your condition.
- Answering a few surveys if we contact you to inform you that we found a genetic change (only if you choose to learn such results).

## Will I learn the results of this study?

You will have the option to be contacted if we find a genetic change that might be important for your health. This may be a genetic change that:

- Causes a medical condition that you already have OR
- Increases the risk of having a medical condition you may not already have

You will indicate your choice when you join the study. The timeline for receiving genetic results is unpredictable and may take a long time. If we identify a genetic change and you chose to be contacted, we will offer to confirm the research finding using a clinical genetic test.

If we do not identify a genetic change, we do not inform participants, as we continue searching for genetic causes.

The Genetic Studies of Constitutional Disorders of the Center for Precision Medicine and Genomics aims to improve our understanding of the genetic factors that put people at risk for common diseases such as heart, liver, endocrine, lung, or kidney disorders. We hope that the knowledge we gain from this study will help us improve medical care by contributing to better prevention, diagnosis, and treatment of illnesses.

### What is precision medicine?

Precision medicine is an extension of traditional approaches to understanding and treating diseases. It incorporates each person's unique genetic, environmental, and demographic profiles in order to offer tailored recommendations for their healthcare.

### What if I want to participate or I have more questions?

We will be very happy to meet with you and answer all of your questions, provide you with more details, and invite you to participate in the study. If you are interested, contact us by email at [cpmg\\_info@cumc.columbia.edu](mailto:cpmg_info@cumc.columbia.edu) or by phone at 212-851-4927.

[www.columbiamedicine.org/cpmg](http://www.columbiamedicine.org/cpmg)



## Genetic Studies of Constitutional Disorders

### INFORMATION FOR PATIENTS



The goal of this study is to learn the relationship between genetic factors and the development of medical conditions.