

The Genotype-Tissue Expression Project (GTEx)

Organ and Tissue Donors

What will GTEx collect and study from organ and tissue donors?

GTEx will collect and study numerous donated tissue samples, such as small amounts of skin, organs or blood.

Health information will also be collected and saved in a secure database at the NIH. This will, for example, include a donor's history of diseases, medications, and the cause of death, and will exclude personal information that might allow others to identify the names of the donors and their family members.

Scientists will use the donated organ and tissue samples and health information to study how changes in genes affect human health.

Why does GTEx need tissue and organ donations?

Common diseases affect many types of organs and tissues. GTEx researchers need to examine samples from many types of tissues and organs donated by as many people as possible in order to facilitate the understanding of these diseases.

Donors or donor families must give permission for organs or tissues to be donated to GTEx. Trained personnel from the hospital or a tissue and organ procurement organization will explain the benefits and risks of participating in GTEx. If they decide to donate, donors or families will be asked to sign a consent form which describes the study. All donors or families receive a copy of the consent form. The form will have the name and contact information of someone who can answer questions.

No organ or tissue donations are ever accepted without the donor's or the family's consent.

What types of tissues and organs may be donated?

Donors who have passed away may donate organs and tissues, such as:

- Fat
- Internal organs (e.g. lung or liver)
- Skin
- Brain
- Blood

What types of studies will the tissues and organs be used for?

First, scientists will be mapping all genes for a donor, also called "**genotyping**." A person's **genotype** is unique, like a fingerprint. Genotyping is usually performed using a blood sample.

Then scientists will study the activity of a donor's genes in different organs and tissues. We are all made up of thousands of genes. Our collection of genes and their specific order is called our genome. But, not every gene in an individual's genome is "expressed" or "turned on." This is referred to as "**gene expression**." These activated genes give us green eyes and/or red hair. They also play important roles in the diseases or conditions we may have, although their influence on health is not completely understood.

Comparing a person's genotype with their "**gene expression**" patterns will help researchers learn how the mapping of each gene affects the gene's activity in different tissues and in different people.

These and other future studies will allow a wide range of scientific questions to be addressed. To also ensure that tissues from GTEx donors are studied among many scientists in the future, some of the skin and blood cells are treated so that they remain alive for a very long time.

Where will donated tissues be stored? For how long?

Tissues and organs donated to GTEx are carefully stored in a "**biobank**" that preserves them for studies now and in the future. The biobank used for GTEx is called the **cancer Human Biobank (caHUB)**. **caHUB** stores tissues on behalf of the GTEx project and is sponsored by the National Cancer Institute, part of the NIH. Brains donated from those who have passed away will be kept in an NIH-supported biobank that specializes in storage of brains and associated nervous system tissues.

The tissue samples donated to GTEx will be stored by NIH indefinitely. Some tissue samples, such as skin and blood cells, will be treated so that they remain alive for long periods of time, allowing them to be studied for many years.

Where can I sign up to be a GTEx donor?

Currently, there is no way to directly sign up to be a GTEx donor. GTEx has agreements with organizations that are involved in organ or tissue transplantations, donations for medical research and training and surgery departments in various parts of the United States, i.e.:

- a. Disease Research Interchange, Philadelphia, Pa.
- b. Gift of Life Donor Program, Philadelphia, Pa.
- c. LifeNet Health, Virginia Beach, Va.
- d. Roswell Park Cancer Institute, Buffalo, N.Y.
- e. Upstate New York Transplant Service, Buffalo, N.Y.

If you register as an organ or tissue donor and pass away near one of these organizations, you may qualify for this study.

How are the donor's and his/her family's identities protected?

During the GTEx study enrollment, every donor is assigned a code number that replaces the donor's name and other personal information. Links from the code number to the donor's name will not be shared outside of the institution where the tissues were collected. No one from the NIH, its partners or researchers using the data or samples will have access to the donor's personal information.

All genetic information will be kept in a secure NIH database. No personal information, such as name or address, will be included in this database. A scientist needs to apply to be able to use most of the data or samples. These applications are reviewed by the NIH to ensure that the scientist's proposal is consistent with the consent provided by donors and families. Only NIH-approved researchers will have access to an individual's genetic information and associated health information. The agreement signed by researchers states that they will not try to find out who the donors are.

Are there risks associated with being a GTEx tissue or organ donor?

GTEx researchers are careful to ensure that a donor's medical and genetic information remains confidential. The NIH strives to protect donors and their families' identities in several ways. All genetic information will be kept in a secure NIH database. However, some genetic data that is summarized from all GTEx donors may be available on public resources on the Internet, such as "**genome browsers**." Genome browsers provide scientific data to the research community in useful and convenient formats in a similar way as the Census data are available to the public. These browsers have been used to display gene sequences and genetic variation data on humans and many other animal and plant species.

Despite taking these precautions, there is a small possibility that someone might connect the name of a donor, or a blood relative, to this information. This may represent the most significant risk of participating. Several state laws and a federal law, the **Genetic Information Non-discrimination Act (GINA)**, have been passed to help protect people from unfair treatment because of their genetic information. (See: **Genetic Information Nondiscrimination Act (GINA) of 2008**)

Will donors and their families receive any benefits from this study?

For some people, knowing that they are contributing to medical research is the primary benefit. Interested individuals may follow general news about the kinds of studies being done at the GTEx Common Fund site and caHUB, which coordinates the recruitment of donors and collecting organs and tissues on behalf of GTEx.

Donors and their families will not receive any direct benefits from participating, such as payment for donating tissues or organs, medical treatments, or test results. If the tissue sample is used in research that develops a new medical product or treatment, donors and their families will not receive any compensation. Donors will not receive individual results from the research done in GTEx.

Can I withdraw my organ and tissue donations and medical information from GTEx in the future?

Participation in GTEx is voluntary. At any time, you may withdraw your tissues and related medical information or those of your family member who died. Donors or families considering withdrawal of their or their loved one's tissues should contact the representative indicated in their GTEx project research consent form, which was completed prior to any donation.

If you withdraw from the study, any samples remaining in the biobank will be destroyed. Any donor-associated information will be permanently removed from the database. However, information and tissue samples already provided to researchers cannot be destroyed or withdrawn.

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