

## Frequently Asked Questions

### A. General FAQs about the dGTEEx Project

#### *What is dGTEEx?*

dGTEEx is the Developmental Genotype-Tissue Expression Project. dGTEEx is an NIH-supported initiative that will establish a scientific research resource of data in pediatric tissues to better understand the impact of genetic variation on health and disease. The data from dGTEEx will help understand how tissues are different from one another, how those cells change during childhood development, and which cells might be the most important in the development and onset of childhood diseases. By studying how genes affect our health, scientists can better understand how to prevent and treat certain types of diseases and reduce health disparities among groups of people.

#### *What is the purpose of the project?*

Genes (*which are composed of DNA*) are found in our body's cells and hold all the information that we pass from parent to child, generation to generation. This information determines characteristics such as eye color, blood type, height, as well as susceptibility to certain diseases. Genes are the instructions our cells use for building and repairing our bodies and for controlling our day-to-day vital functions. Scientists know that certain changes to genes alter the normal instructions and can impact normal human development or increase a person's risk of getting diseases like cancer, diabetes, or cystic fibrosis. Exactly *how* the changes in genes do this is still poorly understood. Research in this area offers incredible potential for discovering how to treat and prevent many devastating diseases, including pediatric disease.

This project has three key goals:

1. This project will obtain samples from multiple tissues and organs from a donor's body. By collecting many different tissues, the project can examine how different body systems interact within an individual and across different individuals during critical developmental processes, like puberty, and in disease. For example, how does obesity impact brain development or heart function during childhood development. These tissues will be physically stored in a **Biobank** which will preserve the samples of donated tissue from many people for use in current and future research. There will be no identifying information about the donor accompanying these samples.
2. Scientists will analyze the donated tissues to determine the unique genetic makeup of the donor and that information will be stored in a genetic **Database** with the associated anonymous medical/clinical information about the donor.
3. The genetic **Database** and **Biobank** will be made available to approved researchers for use in multiple research studies. This will provide scientists with data that will help them investigate the links between genetic information and development within specific tissues.

dGTEEx will create a scientific resource of pediatric tissue and genetic data to examine how gene expression from different tissues changes across development to help advance the study of normal childhood development and pediatric diseases, such as cancer or autism spectrum disorder.

dGTEEx will profile gene expression from the many individual cells that make up each tissue to understand how tissues

are different from one another, how those cells change during development, and which cells might be the most important in the development and onset of childhood diseases. For example, the brain is made up of many different type of cells that are specialized to perform specific functions in the brain, so it is important to understand similarities and differences among those different cell types to understand their normal function in the brain, and to identify how those functions change during the development of disease.

### ***What impact will dGTEEx have on health and medicine?***

There is a potential to help other families in the future through the gift of donating tissues to research. Donating to dGTEEx will help researchers find out what changes in genes are occurring during development that may contribute to pediatric and adult diseases, including cancer, diabetes, and heart disease. There isn't much known about these changes in early development, so that's why donation of your child's normal tissue is so important for the research community. This project will create a much-needed resource of genetic data and tissues that will support a large number of investigations in the future.

Children are different from adults, however scientists don't yet understand why pediatric tissues are different from adult tissues. Understanding how children's tissues, and the cells in those tissues, change through childhood development will help in understanding how pediatric diseases develop and how changes early in development can also lead to adult diseases later on. The dGTEEx project will help in answering these questions by studying the molecular characteristics of a diverse array of pediatric tissues.

To date, no large-scale study has looked at the molecular differences across different tissues from children of different ages. The dGTEEx project will provide a valuable and unique resource of data for scientists to understand the make-up of different cells throughout the body and how cells and tissues change during development.

There are several diseases and disorders that researchers using dGTEEx tissues and data will study. Below are three examples:

- dGTEEx will allow scientists to explore how different cells within the brain may contribute to diseases such as epilepsy and autism spectrum disorder, potentially advancing our understanding of these neurological disorders commonly diagnosed in children.
- Pediatric cancers are distinct from adult cancers in how they develop and are treated. Normal pediatric tissue is needed to help improve diagnosis and treatments for pediatric cancer, like leukemia. By providing normal pediatric tissue to scientists, the dGTEEx project will be an invaluable resource to help medical advancements for families of children with pediatric cancer.
- Children's immune systems go through significant changes during the first 3 years of life. The dGTEEx project will allow scientists to evaluate immune cells to understand how they change throughout life and how allergies, and immune diseases, develop in different individuals.

### ***What does Biobank and genetic Database mean?***

A Biobank is a facility that stores donated tissues from many people so that it can be used in future medical research studies. The genetic Database is where the genetic data and associated anonymous medical/clinical information will be stored.

### ***Where is the Biobank located?***

Brain tissue will be stored at the University of Maryland Brain and Tissue Bank, a not-for-profit organization contracted by the National Institutes of Health to store the tissues. Lung tissue will be stored at the University of Rochester. All other tissue will be stored at the Broad Institute in Boston. The Biobanks are secured facilities and only approved personnel are allowed to access the biobank.

## **B. FAQs about dGTEEx tissue donation**

### ***Who will have access to the tissue samples and the medical/genetic information about the donor?***

Researchers and scientists from academic, government and commercial labs, worldwide will be able to request access. To access the tissues and the full genetic database, researchers will undergo a formal review process before research projects are approved. Researchers will sign an agreement to protect privacy and confidentiality of the donor.

### ***How does the project protect the confidentiality/privacy of the donor and blood relatives?***

The project makes every effort to ensure the confidentiality of the donor and his/her blood relatives. Safeguards include the following:

- Identifying information such as donor's name, address, date of birth, and medical record number are excluded from the Database and the way tissues are labeled.
- No researchers using the genetic Database or tissues in the Biobank will know the identity of the donor, as the information and tissues will be labeled only with a unique code number.
- Researchers using the genetic Database and/or tissues in the Biobank must agree to never try to discover the identity of the donor.

Despite this, there is a small, though highly unlikely, chance that one day this coded data could be combined with other information to reveal the donor's or their biological relative's identity or personal information (re-identification). This information could then somehow be used against the donor's blood relatives, such as impacting the ability for a relative to obtain health insurance. In the unlikely event of a breach of confidentiality where a donor's genetic data become identifiable, individuals and families are protected against discrimination by employers or health insurance companies through the Genetic Information Nondiscrimination Act (GINA). Again, the chances of this occurring are very low and the protections described above are meant to avoid this scenario. Information donor families choose to share information publicly could increase the risk of re-identification.

### ***Can someone make a profit off the tissue and genetic information from this donation?***

For-profit companies will be allowed to apply to use the tissue and genetic database. For example, a pharmaceutical company could use the tissue or data to help discover a new drug to treat heart disease. Families who donate will not receive any money from cures or treatments developed using dGTEEx donations. For-profit companies will undergo the same approval process as academic or government labs and will sign an agreement to protect privacy and confidentiality.

### ***Why are tissues and cells stored indefinitely?***

A unique part of the dGTEEx project is that the future use of information in the genetic Database and tissues in the Biobank is still unknown and will be informed by science and findings that have not yet been discovered. Therefore, it is important to store tissues in the Biobank long-term to address these unknown research questions in the future. In

In addition to tissues, cell lines created from tissues can be stored long-term. Cell lines are useful in research because they allow rapid and wide-ranging research to be performed on live cells. For example, does a potential new cancer drug kill cancer cells and not healthy cells. To access the tissues in the Biobank and create cell lines, researchers will undergo a formal review process before research projects are approved.

***What if I change my mind about the donation?***

You can change your mind and withdraw the tissues and medical information at any time by contacting National Disease Research Interchange (NDRI). Any tissue in the biobank will be destroyed; health information and genetic information will be removed from the Database. However, samples/information that is already part of a study cannot be recalled.

***If the researchers identify a potential disease or disorder using the donated tissues, will the family be notified?***

No. Donor families will not receive any information regarding what the Biobank learns about the donor's health. The Biobank is not organized in a way to provide testing results about diseases. First, the disease testing is not the same as a diagnostic test your doctor would perform. Second, the researchers conducting the testing won't know the identity of the family who donated, so there would be no way for the family to be contacted.

## **C. FAQs about the U-SPEAK study**

***What is the dGTEEx sub-study being led by Johns Hopkins All Children's Hospital?***

This sub-study is called U-SPEAK (UnderStanding Parental Expectations Attitudes and Knowledge Regarding Pediatric Tissue Donation for Research). The study is trying to learn what parents think about donating children's tissue for genetic research after their child's passing.

***What will be involved in the U-SPEAK study if I participate?***

Parents who choose to be in this study may be asked to participate in a one-time focus group with other parents. The focus group will be an online video chat, like through Zoom, and will include discussion of topics related to tissue donation. The focus group will take about 1.5 hours. Parents may also be asked to participate in online surveys regarding topics related to tissue donation.

***Can I change my mind about participating in the U-SPEAK study?***

Yes. The Johns Hopkins All Children's Hospital study team will contact you sometime in the future to provide more details regarding participation in the U-SPEAK study and obtain your consent to participate. You will have the opportunity then to ask additional questions regarding the study. Checking yes on this form allows us to provide your contact details to the study team and is not a commitment to participate.

***Do I have to participate in the U-SPEAK study to donate tissue for dGTEEx?***

No. Participation in the U-SPEAK study is voluntary and is not required to donate tissue for dGTEEx. Additionally, donating tissue for dGTEEx is not required to participate in the U-SPEAK study.

***Who can I contact about the U-SPEAK study?***

Please contact NDRI at 800-222-NDRI (63741) for more information about the U-SPEAK study.