

Evaluation of the Enteroinsular Axis in Cystic Fibrosis (Aim 3 Genetics)

Sponsor: Cooperative study between CHOP and Upenn, sponsored by the National Institutes of Health

Brief Description of Genetics Phase:

- The purpose of this research study is to learn about the genetics of diabetes in cystic fibrosis
- **Type 1 and Type 2 diabetes**-result when either the body does not make enough insulin or the body does not respond correctly to this insulin.
- **Insulin**-a hormone which is made by cells in the pancreas and helps carry glucose (sugar) from the food we eat to the cells of the body for energy.
- In recent years, diabetes has emerged as one of the most significant co-diseases that many cystic fibrosis (CF) patients develop. While CFRD has many features similar to both Type 1 and Type 2 diabetes, it is very different; therefore, treatment and care of CFRD should not be the same.
- This study looks at how genes associated with Type 2 diabetes are related to diabetes and insulin secretion that occurs in cystic fibrosis
- Your participation in this study will include 1 approximately 30-minute study visit to the Hospital of The University of Pennsylvania (UPenn).
- You will be compensated \$10 in cash for your time

Contact Information:

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Study Procedures:

Blood/Saliva sample:

- A blood sample will be drawn by needle stick for testing a specific gene that has been linked with diabetes.
- This study will look at how common this gene is in CF, along with other genes that are related to Type 2 diabetes.
- About 1 teaspoon of blood will be drawn. This test can be done at the time of a clinical draw or can be done specifically for this study—the option is yours.
- A blood specimen is preferred; however, if that is not possible then we will collect a sample of your saliva

Follow-Up/Phenotyping Phase:

- The results of the genetic testing will not be shared with you—largely because the meaningfulness of the genes in CFRD has yet to be determined.
- Based on your results you may be invited to participate in a follow-up study. Involvement in this second study is completely voluntarily.
- At the time of this invitation, we can share what information we have regarding the genetics of CFRD and can share with you the results of your studies.

What are the possible risks or discomforts?

Risks associated with blood draw:

- There is a small and rare risk of infection and fainting. There may be some pain or bruising of the skin at the site where the blood is taken.
- For children in the study, the blood samples will be drawn by a health professional skilled in drawing blood in children

Risks associated with DNA-Genetic testing:

- There may be a risk that if people other than the researchers have access to your genetic information they could misuse it. We think the chance of this ever happening to you is very small because your personal information will not be attached to the blood used for research.
- The laboratory personnel that will analyze your DNA will not know your personal information. The genetic results will be stored under a code number without any information that may identify you, in a secure, password-protected database. The researchers who view this database will not have access to your name or identifying information.
- The Genetic Information Nondiscrimination Act is a Federal law that makes it illegal for some groups to discriminate against you based on genetic information. This law applies to all health insurance companies, group health plans, and employers with 15 or more employees. This law does not apply to companies that sell life insurance, disability insurance, or long-term care insurance.