UPMC LIFE CHANGING MEDICINE

Patient Name:_____
Identification Number: _____

FAMILY VARIANT OF UNCERTAIN SIGNIFICANCE CO-SEGREGATION PARTICIPANT CONSENT

WHAT IS THE PURPOSE OF TESTING?

The intent is to determine whether a genetic finding is associated with disease or not. This information can serve as an explanation for the cause of disease in the family and it may change medical management for you and/or your relative, depending on the results.

WHAT SPECIFIC INFORMATION IS THE LABORATORY INVESTIGATING?

Everyone has genetic variants or changes in their DNA, most of which are harmless. There are some variants identified in an individual which have not been seen often enough in the general population to know if it is associated with the disease or not. These variants are referred to as "variants of uncertain significance" or VUS. VUS are not medically actionable. Your relative was identified to have a VUS. One possible way to learn more about a VUS is to see whether the variant corresponds with disease in other family members.

IF I CHOOSE TO PARTICIPATE, WHAT'S NEXT?

You would give permission to your healthcare provider to release your medical and family history information. You would also need to provide a blood or saliva sample. Your healthcare provider can order a saliva kit or arrange the blood draw to be done locally.

IS THERE A COST FOR THIS TEST?

There is no additional charge to participate in the VUS SEGREGATION ANALYSIS PROGRAM.

WHAT SHOULD I EXPECT IN TERMS OF RESULTS?

- If the analysis of your DNA leads to a change in the classification of the family variant from a VUS to disease-causing, then you and your relative will receive a written report.
- If the analysis of your DNA leads to a change in the classification of the family variant from a VUS to likely benign or benign, then your relative will receive an updated written report acknowledging your sample and change in variant classification. You will not receive a report.
- If the analysis of your DNA does not change the classification, then your relatives' updated report will acknowledge receipt of your sample. You will not receive a report. - However, if the laboratory learns through new evidence that the VUS is reclassified to be disease-causing, both you and relative will receive a report. The report will be submitted to the healthcare provider who coordinated the initial study for dissemination to family members as long as that provider can still be contacted.

GENETIC TESTING INFORMATION

This information can be obtained from the original family member tested.

PCGL DNA# of Proband: _____Relationship to Proband: _____

INFORMED CONSENT

UPMC LIFE CHANGING MEDICINE

Patient Name:_____
Identification Number: _____

FAMILY VARIANT OF UNCERTAIN SIGNIFICANCE CO-SEGREGATION PARTICIPANT CONSENT

My signature below indicates I have received information about the VUS SEGREGATION ANALYSIS PROGRAM, and that I have read and understood the material in this document. I have been given the full opportunity to ask questions regarding my participation.

_____

Participant's Name (please print) Participant's signature Date

INTERPRETER'S STATEMENT

Execute if an interpreter is provided to assist the individual in understanding this informed consent form:

I have translated the information and advice presented orally to the individual to be treated by the person obtaining this consent.

In addition, I have sight translated the consent form (read it aloud in his/her language). To the best of my knowledge and belief he/she understood this explanation.

_____

Cyracom ID (if applicable)

_____

Print Name

_____

Signature (Not required if a Cyracom Interpreter Was Used)