UPMC LIFE CHANGING MEDICINE

Patient Name:_____
Identification Number: _____

APPLICATION FOR FAMILY VARIANT OF UNCERTAIN SIGNIFICANCE CO-SEGREGATION TESTING

Family studies on variants of uncertain significance may help elucidate the significance of a variant identified in the proband. Family studies are considered on a case-by-case and are authorized under the discretion of the director.

POINTS CONSIDERED FOR ELIGIBLITY

- **Penetrance of the gene**: Segregation studies are less likely to be informative for a VUS in genes associated with low or moderate penetrance and most likely informative where penetrance is high. Additional affected and unaffected individuals must be available in the family.
- **Phenotypic spectrum associated with the gene**: Family studies are most informative for genes with a well-defined, highly specific phenotype that is strongly associated with the specific gene or group of genes and less informative for genes associated with highly variable non-specific phenotypes.
- **The availability of documentation to support family history reports:** Documentation is particularly important for genes associated with specific histologic or molecular features. Family studies will only be performed if relevant documentation of a relative's diagnosis is available.
- **The likelihood that testing of specific family members will be sufficient**: Family studies in kindreds with a limited number of informative individuals are unlikely to be sufficient for VUS resolution, particularly if the variant is novel or very rare.

In order to be considered, please provide the following information.

Proband's name_____ Proband's Lab number_____

Physician/GC_____

Phone number _____ Fax number _____ Email _____

1. Which DNA variant are you interested in testing?

2. Please provide clinical rationale for the request of the family study.

3. Please list the informative relatives. Attach clinically relevant notes and pedigree if available.

NAME	GENDER	RELATIONSHIP TO PROBAND	HEALTHY	AFFECTED	SYMPTOMS/DISEASE (if affected)	DX AGE (If affected)
			☐	☐		
			☐	☐		
			☐	☐		
			☐	☐		
			☐	☐		

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If the family member(s) are considered eligible to participate in the program, the healthcare provider will be notified. The family member(s) would need to sign the PARTICIPANT CONSENT FORM. Results will take approximately 4-6 weeks upon receipt of all participating family members.

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)