

Supplement 3: Request form completed by the family to select the types of incidental findings that they wish to have report from whole genome sequencing (WGS)

SAMPLE SUBMISSION FORM
Request Concerning Incidental Findings Found By Whole Genome Sequencing (WGS)

PATIENT INFORMATION (Required)

Last Name

First Name

Date of Birth (mm/dd/yy)

It is recommended that the patient/family receive genetic counseling regarding whole genome sequencing (WGS) before and after the test. My signature below indicates that I have been informed of the following facts about the whole genome sequencing test and that I have had the opportunity to have any questions answered.

This test will be performed to find the genetic basis of my/my child's disorder.

1. I may learn that one or more DNA differences called variants in one or more genes are likely to explain the cause of the disorder in me/my child.
2. I may learn that no specific DNA variants were detected that may explain my/my child's disorder. This outcome does not mean that I/my child do not have a genetic disorder.
3. I may learn that one or more DNA variants were identified that may cause medical conditions that are unrelated to my/my child's disorder. These are referred to as Incidental Findings. Efforts will be made to limit the number of Incidental Findings.

This test examines the entire human genome for DNA variants. We classify the test results into two types:

1. Primary Results (DNA variants likely to be responsible for the disorder under investigation in my/my child's case) Primary Results will always be included in the laboratory report.
2. Incidental Findings (DNA variants that are not likely to be responsible for the disorder under investigation in my/my child's case but were seen in the process of finding the Primary Result). Incidental Findings that cause a disorder where medical intervention can prevent or decrease the effect of a childhood onset disease will always be included in the laboratory report.

Indicate the types of Incidental Findings you wish to have reported in addition to the required results (check boxes below):

No Other Incidental Findings Retain data for future analysis

OR

- Untreatable Childhood Disorders (eg. Tay-Sachs disease)
 Treatable Adulthood Disorders (eg. Hereditary Colon Cancer)
 Untreatable Adulthood Disorders (eg. Alzheimers Disease)
 Carrier of a Disorder (eg. Phenylketonuria)

Signatures (Sign here to provide consent and print name following signature)

Patient (If a minor, assent if appropriate)

Date (mm/dd/yy)

Genetic Counselor

Date (mm/dd/yy)

Ordering Physician

Date (mm/dd/yy)

Parent or Guardian (If patient is a minor)

Date (mm/dd/yy)

Parent or Guardian (If patient is a minor)

Date (mm/dd/yy)