Fimlab

INFORMED CONSENT FOR EXOME SEQUENCING

Information about exome sequencing is available in Fimlab's test instruction manual through our Extranet service.

By signing this form, I give my consent to exome sequencing study. I have received information about the nature of the test, and I am aware that:

- The test aims to identify a genetic change in the fetus that explains the structural abnormalities observed. The result may confirm a hereditary disease in the fetus. The test may also reveal other heredity related information, such as familial relationships.
- The result may have significance for other family members.
- Detected genetic variants are interpreted in the context of the provided clinical information and current scientific knowledge. Interpretation may change or be supplemented as new research emerges. Only pathogenic or likely pathogenic variants that, based on current knowledge, are considered likely to explain the observed structural abnormalities will be reported. Variants of uncertain significance are not reported.
- A negative result does not exclude the possibility of a hereditary disease or genetic predisposition.
- The result may be inconclusive or require further studies.
- The analysis is performed at Fimlab's subcontracting laboratory in Germany (Medicover Genetics / MVZ Martinsried), where samples and relevant clinical information from the referral form are sent for analysis and interpretation. Information is handled confidentially in accordance with the General Data Protection Regulation (GDPR) and the data protection agreement between the laboratories.
- The sample may be used as a positive control, for example in genetic testing of relatives, for internal laboratory quality assurance, or for method development.
- Individual genetic variants detected in the test may be reported to national or international databases without personally identifiable information.

- It is possible that an incidental (secondary) finding may be detected in a parent's sample that is unrelated to the fetal findings. The American College of Medical Genetics (ACMG SF v3.2) has listed genes for which reporting of secondary findings is recommended in connection with broad sequencing studies (https://www.sciencedirect.com/science/article/pii/S1098360023008791?via%3Dihub). These relate to conditions or predispositions where monitoring or treatment may have health significance. Secondary findings are handled according to the signed consent of the person being tested. ACMG listed secondary findings are not reported from fetal samples.

Fetal sample information

Fetus of (name of the expectant person): _____

Date of birth: _____

(If request is submitted under fetus's own personal ID, the sections above are completed with his/her details.)

Signature: _____ **Date**: _____

(If signed by a person other than above, please add name clarification and relationship to the fetus.)

Name of the treating / ordering clinician: _____

The requesting physician or healthcare unit submits the signed form to Fimlab Genetics either by fax (09 425 782 83) or by encrypted email (genetiikka@fimlab.fi). Alternatively, it may be agreed with the patient that they bring the completed and signed form to the sample collection unit, from where it is forwarded to Fimlab together with the sample tube.

Family members in trio analysis:

Samples to exome sequencing from other family members. Analysis is requested with code B -KontR-D.

Person 1:

Name: _____ Date of birth: _____

Relationship: ☐ mother ☐ father ☐ sibling ☐ other _____

Secondary findings reporting

☐ I wish to receive information about possible secondary findings.

☐ I do NOT wish to receive information about possible secondary findings.

Signature: _____ **Date:** _____

Person 2:

Name: _____ Date of birth: _____

Relationship: ☐ mother ☐ father ☐ sibling ☐ other _____

Secondary findings reporting

☐ I wish to receive information about possible secondary findings.

☐ I do NOT wish to receive information about possible secondary findings.

Signature: _____ **Date:** _____

In trio exome testing, samples requested under code B -Kontr-D from parents or other family members are used to support diagnosis and to assess the significance of variants found in the fetus. Reports for parents or other family members include the parental result relevant to the finding reported in the fetus, and—if consent has been given—any secondary findings.

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