Fimlab

INFORMED CONSENT TO EXOME SEQUENCING

You can find information regarding exome sequencing on patients and family members in Fimlab's laboratory manual Exome sequencing, blood - Fimlab.

By signing this form, I give my consent to large-scale sequencing. I have been informed about the nature of the test and I am aware that:

- The result may confirm a diagnosis or predisposition to a hereditary disease.
- The result may also be relevant to the other members of my family.
- A negative result does not exclude the possibility of a hereditary disease or a predisposition thereof.
- The result may remain unclear or require further testing.
- Any detected alteration will be interpreted based on the current information and data, so it is possible that the interpretation may be changed or complemented due to new data.
- The test will be performed at Fimlab's subcontracted laboratory in Germany (Medicover Genetics / MVZ Martinsread), where my sample and clinical current information and data in the referral form will be sent to complete the test and interpret the results. The information will be processed confidentially in accordance with the General Data Protection Regulation (GDPR) and the data protection agreement between laboratories.
- The sample may be used as a positive control sample, for example, in genetic testing on relatives, internal laboratory quality assurance or method development.
- Individual genetic alterations detected in the test can be reported to national or international databases without any information that would identify the person.

- It is possible that my sample may contain a gene variant that is unrelated to the disease being tested as a secondary finding. The American College of Medical Genetics has listed genes (ACMG SF v3.2), which they recommend to report if detected as secondary findings in large-scale sequencing (https://www.sciencedirect.com/science/article/pii/S1098360023008791?via%3Dihub). Monitoring and treating the diseases or predispositions linked to these genes may have a significant impact on the patient's health. The secondary findings will be processed according to the signed consent of the subject.

Patient information

Patient name: _____ **Date of birth:** _____

Test type

☐ B -ExSeq-D or ☐ B -ExTri-D

Information and consent of the family members can be found on the next page. These are only required in case of a request for B -ExTri-D test.

Reporting secondary findings in the patient

☐ I would like to be informed of any secondary findings. ☐ I do not wish to be informed of any secondary findings.

Signature: _____ **Date:** _____

Name of the signatory (and family relationship to the subject if the signatory is their legal representative):

_____

Name of the treating / commissioning physician: _____

The commissioning physician or healthcare unit will deliver the signed form to Fimlab's Genetics Laboratory either by fax to +358 (0)9 425 782 83 or encrypted email to genetiikka@fimlab.fi. Another option is that the patient will take the completed and signed form with them to sample collection, where it will be sent to Fimlab together with the sample tube.

Information and consent of the other subjects for B -ExTri-D test:

Patient name: _____ **Date of birth:** _____

Other samples from the family for exome sequencing. Note! These tests are ordered under a request for B -Kontr-D.

Person 1:

Name: _____ **Date of birth:** _____

Family relationship: ☐ mother ☐ father ☐ sibling ☐ other _____

Reporting secondary findings

☐ I would like to be informed of any secondary findings.

☐ I do not wish to be informed of any secondary findings.

Signature: _____ **Date:** _____

Person 2:

Name: _____ **Date of birth:** _____

Family relationship: ☐ mother ☐ father ☐ sibling ☐ other _____

Reporting secondary findings

☐ I would like to be informed of any secondary findings.

☐ I do not wish to be informed of any secondary findings.

Signature: _____ **Date:** _____

In trio exome sequencing, samples requested from the parents or other family members under B -Kontr-D will be used to help determine the patient's diagnosis and assess the significance of the genetic alterations detected in the patient. The medical statements of the parents / other family members will indicate the parent's results in relation to the findings reported in the child, as well as any secondary findings if consent has been granted.

The commissioning physician or healthcare unit will deliver the signed form to Fimlab's Genetics Laboratory either by fax to +358 (09) 425 782 83 or encrypted email to genetiikka@fimlab.fi. Another option is that the patient will take the completed and signed form with them to sample collection, where it will be sent to Fimlab together with the sample tube.