INFORMED CONSENT FOR EXOME SEQUENCING (B -ExSeq-D, B -ExTri-D, Ts-ExSeq-D, Ts-ExTri-D, B -ExKon-D, Ts-ExKon-D)

According to the assessment by the attending physician, an exome sequencing test is useful for determining the cause of the disease. Consent for HUSLAB's diagnostic exome sequencing test is voluntary and you can withdraw your consent at any time before the completion of the test.

By signing this form, I consent to an extensive sequencing test, accept the reporting of the results and the storage of the information in the information systems managed by HUS. I have received information about the test (e.g., an information leaflet, verbally) and had an opportunity to discuss the test with a physician.

- The result may confirm the diagnosis of a hereditary disease or a predisposition to it
- The result may also be significant for other members of my family
- A negative result does not exclude the possibility of a hereditary disease or a predisposition to it
- The result may remain unclear or require further examinations
- The detected variants will be interpreted in light of the provided clinical information and current knowledge and it is possible that the interpretation will change or be supplemented when new research data becomes available
- The physician can issue a new referral later to request a new assessment of the results
- If any of the sample is left over, it will be stored at the HUSLAB genetics laboratory for at least two years, after which it may be destroyed
- The sample may be used as a positive reference sample, for example, in the gene tests of relatives, in the laboratory's internal quality assurance or in the development of methods.
- Any individual genetic variants may be reported in national or international databases without details individualising the person

Patient's details

Disease or symptom tested: _____

Name of the patient tested: _____**Personal identity code**: _____

Type of test

☐ B -ExSeq-D or Ts-ExSeq-D ☐ B -ExTri-D Ts-ExTri-D parents' details and consents on the next page (order parental tests using a B -ExKon-D or Ts-ExKon-D request)

Reporting of the patient's incidental findings

An incidental finding is a disease-causing or a likely disease-causing variant in one of the 81 genes in the so-called ACMG SF v3.2 list (https://www.sciencedirect.com/science/article/pii/S1098360023008791?via%3Dihub). It is not related to the clinical picture or symptom that was the reason for the exome test. The diseases or predispositions related to these genes are such that monitoring or treatment may have a positive effect on the person's health.

☐ I give permission to report incidental findings ☐ I do not give permission to report incidental findings

☐ The significance of incidental findings has not been discussed with a physician / I have not made a decision on reporting incidental findings, therefore they will not be reported

Signature: _____ **Date**: _____

Name of the signatory (and family relationship with the person tested if the signatory is a legal representative):

_____

Signature of the attending physician: _____

Attending physician: _____

Post or fax the signed form to the HUSLAB genetics laboratory

Details and consent of other persons tested in the B -/Ts-ExTri-D set up

Name of the patient: _____ **Personal identity code**: _____

The family's other samples for the exome test (order tests using the code B -ExKon-D or Ts-ExKon-D)

Person 1:

Name: _____ Personal identity code: _____

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

Is the person tested healthy regarding the symptoms of the index patient: ☐ yes ☐ no (provide the details in the electronic request)

Reporting of incidental findings

An incidental finding is a disease-causing or a likely disease-causing variant in one of the 81 genes in the so-called ACMG SF v3.2 list (https://www.sciencedirect.com/science/article/pii/S1098360023008791?via%3Dihub). It is not related to the clinical picture or symptom that was the reason for the exome test. The diseases or predispositions related to these genes are such that monitoring or treatment may have a positive effect on the person's health.

☐ I give permission to report incidental findings ☐ I do not give permission to report incidental findings

☐ The significance of incidental findings has not been discussed with a physician / I have not made a decision on reporting incidental findings, therefore they will not be reported

Signature: _____ Date: _____

Person 2:

Name: _____ Personal identity code: _____

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

Is the person tested healthy regarding the symptoms of the index patient: ☐ yes ☐ no (provide the details in the electronic request)

Reporting of incidental findings

An incidental finding is a disease-causing or a likely disease-causing variant in one of the 81 genes in the so-called ACMG SF v3.2 list (https://www.sciencedirect.com/science/article/pii/S1098360023008791?via%3Dihub). It is not related to the clinical picture or symptom that was the reason for the exome test. The diseases or predispositions related to these genes are such that monitoring or treatment may have a positive effect on the person's health.

☐ I give permission to report incidental findings ☐ I do not give permission to report incidental findings

☐ The significance of incidental findings has not been discussed with a physician / I have not made a decision on reporting incidental findings, therefore they will not be reported

Signature: _____ Date: _____

In the trio exome sequencing test, the parents' samples will be used to help determine the patient's diagnosis and to assess the significance of the genetic variants identified in the patient. The parent's sample will not be analysed comprehensively. In accordance with their consent, clinically significant incidental findings related to the 73 genes with regard to which monitoring or treatment may have a positive effect on the person's health may be reported to the parent. Both parents will receive a separate statement explaining the parent's result in relation to the finding reported for the child and, in accordance with their consent, the situation with any incidental findings.

Post or fax the signed form to the HUSLAB genetics laboratory