



INFORMED CONSENT FOR EXOME ANALYSIS

Patient's name _____

Personal identification number _____

I hereby confirm that I have received and understood the information regarding **exome analysis** and have been informed about the meaning of the following facts:

1. I am aware of the fact that this analysis searches for genetic findings that are associated with a hereditary disease or with an increased risk for developing a hereditary disorder. I understand that this analysis could also identify further hereditary information, for example paternity and kinship.
2. I am aware that this analysis may identify genetic variants that are classified as clinically uncertain variants, so that the association with the patient's particular disorder remains unclear. The laboratory team reports only genetic variants that are classified as unclear variants (variant of uncertain significance, VUS). The variant classification is based on the American College of Medical Genetics and Genomics (ACMG) guidelines. I am aware that my physician may recommend further analyses or genetic counseling to clarify the significance of the research result.
3. I am aware that TYKS Genomics laboratory generates a gene sequencing result of my probe, analyzes and interprets the findings, and provides a report of the result(s). The genetic sequence obtained from the probe will be further analyzed via the Sophia DMM software and the genetic sequence will be stored anonymously in the Sophia Genetics analysis software for a time-period of 5 years. The genetic sequence will be confidentially transferred and personal information will never be shared with the software program.
4. I am aware that results of this analysis may be possibly presented anonymously in a research publication and /or a medical congress. Personal information will never be shared.
5. My DNA sample will be stored in the laboratory for any potential further analysis in the future (an additional investigation will be only carried out after receiving a request from the treating physician with the patient's consent). I am aware that I can request my sample to be disposed upon sending a written request to the laboratory after the completion of this analysis at any time.
6. It is possible that this genetic analysis identifies a variant that is not associated with patient's specific disorder (secondary or incidental finding). The American College of Medical Genetics and Genomics (ACMG) has reported recommendations for the list of secondary findings in an international publication. These recommendations include likely pathogenic and pathogenic variants that are associated with secondary findings unrelated to the patient's disorder and it has been recommended to include the investigation of these particular genes when performing an exome analysis. It is also possible that this analysis may reveal likely pathogenic or pathogenic genetic variants that are of clinical relevance for the patient. As an example, secondary findings may be associated with a high malignancy risk, so that the patient could benefit from adequate surveillance after the identification of such a variant.
7. I am aware that in the trio-exome study, the medical records of the subject's parents in the patient information system are reviewed for the referral.

By signing this consent form I hereby confirm that I have read and understood the content of this informed consent. I have had an opportunity to ask further questions about my consent and my questions have been answered.

- I wish to be informed about incidental findings
- I do not wish to be informed about incidental findings

Patient´s name:	Personal identification number:
Signature (if not the patient):	Relationship to patient (parent, guardian, trustee):
Signature (patient):	Date:

- I wish to be informed about incidental findings
- I do not wish to be informed about incidental findings

Examinee´s name:	Personal identification number:
Relationship to the patient (parent, guardian, trustee):	
Signature:	Date:

- I wish to be informed about incidental findings
- I do not wish to be informed about incidental findings

Examinee´s name:	Personal identification number:
Relationship to the patient (parent, guardian, trustee):	
Signature:	Date: