

INFORMED CONSENT FOR DNA ANALYSIS (Next generation sequencing - NGS)

Parent Name: _____
Date of birth: _____ Phone: _____
e-mail: _____
Address: _____

1. Child Name: _____
Date of birth: _____ Gender: male/female
2. Child Name: _____
Date of birth: _____ Gender: male/female

I have had genetic counselling about the genetic basis and the molecular genetic analysis regarding

I have had the opportunity to ask all questions I had and I received and understood the answers given in a detailed discussion.

I consent to a peripheral blood sample being taken for DNA extraction and for molecular genetic analysis. I was informed about the minimal risks during the biological sample taking and the possibility that a second sample may be required.

I will be informed in writing about the results obtained by the molecular testing.

I consent to the remaining DNA material being preserved in a DNA bank and used for research purposes.

I consent to the use of the results obtained during the analysis for research purposes and to be published in research journals after complete anonymization of my personal data.

Genetic analysis information:

1. The analysis includes sequencing of the target genes coding regions, including the intron-exon junctions that could result in gene splicing defects.
2. Analysis includes target genes associated with the clinical symptoms of the patient.
3. Due to methodological limitations, next generation sequencing does not give full coverage for all genes and all coding regions.
4. Large deletions/duplications, expansions and etc. cannot be detected by this method.
5. The raw data may include potential sequencing artefacts specific for the sequencing technology.
6. If necessary, regions with unsatisfactory coverage can be reanalysed by an alternative method such as Sanger sequencing, which has to be additionally paid.
7. The next generation sequencing is a screening method. All pathogenic variants detected by NGS have to be verified by an alternative method.
8. Clinical exome sequencing includes sequencing of 4813 genes with clear phenotype correlation.
9. Whole exome sequencing covers ~ 20 000 genes including such with unclear clinical significance.
10. If no disease-causing genetic variant is found, genetic changes responsible for the disease may still exist.
11. Sometimes gene variants are proven but their clinical significance is unclear. A comprehensive explanation of all possible causes of diseases due to genetic reasons is not possible. It is also not possible to exclude every disease risk utilizing genetic analysis.
12. If need be reanalysing the genes as a result of new clinical information about the patient, additional payment is required.

I hereby give Laboratory “Genica” my consent to perform the analysis in collaboration with another laboratory: _____.

I hereby give Laboratory “Genica” my consent to operate with my personal data in relation to the diagnostic test.

Date: _____

Patient signature: _____