Request with a declaration of informed consent

to INFANO preventive test

SOP NZOZ GENOMED-04, zał. 13 v 02.2

To be filled in by the lab	Date (dd/mm/yy): ...../...../..... time of acceptance: ..... Accepted by:	Code (given by the lab)

PATIENT'S DETAILS (to be filled in by the parent/legal guardian)

Name and surname of the Patient:

Statutory representative (name and surname):

Residence address of the Patient/statutory representative:

E-mail:

(**Can we use this e-mail to send test results?** □ **YES**)

PESEL No:

Date of birth D D / M M / Y Y Y Y

Phone No.

Sex: □ male □ female

TEST DESCRIPTION

INFANO is a genetic screening test for childhood onset diseases of genetic origin. The purpose of the test is to detect diseases before their clinical symptoms appear. It allows starting treatment before changes and damages are irreversible and preventing serious complications due to late diagnosis. We selected the diseases and genes covered by the INFANO test on the basis of data on genetic causes of diseases from up-to-date recommendations, databases, and scientific papers, with well-documented diagnostic significance and considerable prevention and/or treatment potential.

I have been advised that a diagnostic report will be drafted on the basis of my child's genetic data analysis. The report will feature the following:

- The result of analysis for pathogenic genotypes of 78 diseases covered by the INFANO test, based mainly on data obtained using NGS, but also using other essential methods, as in the case of spinal muscular atrophy. In particular, the result covers 21 groups of diseases displaying incomplete penetrance, selected and reported according to the recommendations of the American College of Medical Genetics and Genomics: ACMG SF v3.1 (Miller et al., Genet Med. 2022), as they may directly affect my child's health and life.
- An optional result of MLPA for one disease not covered by NGS, which meets the preventive assumptions of the INFANO test. The analysis will be carried out if I express my consent herein and agree to bear additional costs.

Full information on the INFANO test options is available at: infanotest.pl.

I acknowledge that:

1. The diagnostic report will feature information on detected pathogenic genotypes, i.e.:
   - Detection of two pathogenic or likely pathogenic variants in the same gene in the case of a recessive disease;
   - Detection of a pathogenic or likely pathogenic variant in a gene responsible for a dominant disease.
2. The report will not provide information on genetic variants of uncertain clinical significance or on variants that do not give symptoms.
3. Diseases listed in ACMG SF v.3.1 are serious, but a positive genetic test result is not tantamount to developing them. I have considered the burden such information on my child will place on me.
4. When I receive a negative test result but any of the diseases it covers or some additional symptoms are suspected, I can request Genomed to extend the analysis and interpretation of the result data. It will require a doctor's referral stating the grounds on which analysis should be extended beyond the preventive purpose of the INFANO test.
5. The INFANO test is not intended for children whose symptoms indicate a specific genetic disease or a chromosomal syndrome. In such case, a targeted diagnostic test is recommended.
6. The lab may suggest a comparative genetic test for the Patient's biological parents and in some cases also for the Patient's siblings to be able to interpret the result correctly.
7. If the currently collected sample does not meet the quality criteria, it may be necessary to re-sample or collect a different kind of sample.
8. The final test report is the diagnostic test result as defined by the Polish Act on laboratory testing. The report will be prepared in up to 8 weeks from the date when the Genomed lab receives the sample. I will receive information on a positive test result immediately, even before the diagnostic report is drafted.

CONSENT TO AN ADDITIONAL TEST (It requires an additional payment, beyond the INFANO test price.)

☐ **In addition** to the INFANO test, please test the Patient for congenital adrenal hyperplasia (gene *CYP21A2*) using MLPA.

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ADDITIONAL INFORMATION AND DECLARATIONS

1. I acknowledge that Next-Generation Sequencing (NGS) will be used to analyse coding region (exon) sequences of selected 139 genes together with intron-exon splicing regions (+/- 20 nucleotides away from the exon) and selected variants of well-documented clinical significance found in the gene non-coding regions. If NGS reveals pathogenic genotypes in the genes under analysis, the presence of pathogenic and likely pathogenic variants will be verified using a reference method (Sanger sequencing).
2. I acknowledge that sequencing does not detect extensive gene deletions, duplications, or rearrangements. Such defects may also cause genetic diseases. The genes responsible for spinal muscular atrophy and congenital adrenal hyperplasia (the latter ordered separately) will be tested using MLPA.
3. I am aware that correct interpretation of genetic test results requires correct information on kinship.
4. I agree that the test results and any biological material left can be anonymously used for the purposes of research, diagnostic test development, statistical analyses, and scientific publications.
5. I agree that Genomed can keep the Patient's genetic material once tests are completed.
6. I declare I have been informed that I can withdraw the consents from sections 4 and 5 above at any time and I can demand the data to be deleted as from the date Genomed receives my request to that effect.
7. The physician who ordered the test has explained its essence and its diagnostic significance to me (pursuant to Article 9(2) of the Act of 6 November 2008 on patient rights and the Patient Ombudsman) OR

☐ I declare that before I agreed to the test I exercised the right resulting from Article 9(4) of the said Act and made an informed decision not to request the information referred to in Article 9(2) of the said Act on the diagnostic significance of the planned genetic test and the essence of the disease it can detect from my physician.

…… ……

Place and date *Signature of the parent/statutory representative*

INFORMATION ON THE REFERRING CENTRE (WHERE TO SEND THE TEST RESULT)

Name:		NIP (VAT) number:	
Address:		Phone No.:	
		E-mail:	

CLINICALLY SIGNIFICANT INFORMATION (to be filled in by the referring physician/centre)

Was the Patient's newborn screening test result positive? ☐ YES ☐ NO
If the answer is YES, what was the disease and the test result? (Please attach it.)
……

Has the Patient had any genetic tests done? ☐ YES ☐ NO
If the answer is YES, what was the disease and the test result? (Please attach it.)
…… ……

Are you aware of any genetic disease(s) in the Patient's family? ☐ YES ☐ NO
If the answer is YES, please name the disease(s) and degree of the Patient's kinship with the person(s) who had them:
……

Has the Patient had a bone-marrow transplant? . ☐ YES ☐ NO
* A bone-marrow transplant makes it impossible to test blood and saliva

Did the Patient have a blood transfusion in the last 3 months? ☐ YES ☐ NO
* Genetic tests can be performed 3 months after a transfusion. Otherwise the test results could be false.

Other important clinical data:
……

Signature and stamp of the referring physician		Sample collected by	
Place and date		Date and time of sample collection	

SAMPLE TYPE (please tick)

☐ Oral cavity swab ☐ Saliva ☐ Umbilical cord blood ☐ Venous blood ☐ Other ……