Request with a declaration of informed consent to the PARENTO reproductive risk genetic test

SOP NZOZ GENOMED-04, zał. 14 v 02.2

To be filled in by the lab	Date (dd/mm/yy): ..... /..... /..... time: ..... of sampling Sample received by:	*Code (given by the lab)*

PATIENT'S PERSONAL DATA

Name and surname:		PESEL number:
Address:		DOB: D D / M M / Y Y Y Y
E-mail: (**Can we use this e-mail to send test results?** ☐ **YES**)		Sex: ☐ male ☐ female
Partner's name and surname: (if a couple gets tested together)		Phone No.:

TEST DESCRIPTION

PARENTO OPTIMUM and PARENTO MAXIMUM are screening tests for genetic disease carrier status. They have been developed by Genomed experts for future parents who would like to get to know their individual and common reproductive risk, or likelihood of having a child with a hereditary genetic disease. With this knowledge at hand, they can calmly and responsibly plan to become parents in line with their personal beliefs.

The PARENTO test is performed on the basis of data obtained via Next-Generation Sequencing (NGS). The test has two options:

- **PARENTO OPTIMUM** – analyses a sequence of 191 (*women*) or 170 (*men*) genes; the test detects 90 most frequent genetic diseases in the Polish or general population (carried by ≥1/200) and >50 other serious genetic diseases.
- **PARENTO MAXIMUM –** analyses a sequence of around 1700 (*women*) or 1500 (*men*) genes obtained via Whole-Exome Sequencing (WES); the test covers all known genes responsible for recessive and sex-linked diseases that can be reliably evaluated via NGS.

Both options include frequent diseases that are impossible to detect by NGS alone:
spinal muscular atrophy (SMA) (both sexes), congenital adrenal hyperplasia (CAH) (both sexes), and fragile X syndrome (FXS) (women).

Full information on both PARENTO options is available at parento.pl.

I acknowledge that:

- The diagnostic report will provide information on discovered genetic variants that constitute reproductive risk, i.e. pathogenic or likely pathogenic variants that cause recessive or sex-linked genetic diseases.
- The report **will not** provide information on genetic variants of uncertain clinical significance or on benign variants.
- If both partners are tested, reproductive risk will be presented in relation to the partner's result, but for rare diseases it may be impossible to provide precise reproductive risk if frequency of occurrence, pathogenic variant statistics and/or test clinical sensitivity are unknown.
- The test does not offer absolute guarantee that the diseases it covers would be ruled out, hence a negative test result nevertheless has some residual risk resulting from the constraints of the methods applied and/or the current state of medical knowledge.
- The recessive disease carrier status usually gives no symptoms, but sometimes it can be a predisposition to develop, for instance, cancer or a neurodegenerative disease. It is not the purpose of the PARENTO test to obtain such information, but if the test is to be conducted correctly, information on such variants will be obtained regardless. Aware of the above:

☐ I DON'T WANT ☐ I WANT

the result to include an analysis of the risk of detected variants to my own health.

- The PARENTO test **may not be the best choice** for people from families aware of high reproductive risk. In such case, they should consider a targeted genetic test.
- 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.
- 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 (PARENTO OPTIMUM) or in up to 12 weeks (PARENTO MAXIMUM) from the date when the Genomed lab receives your sample.

TEST OPTION SELECTION

Parento Optimum	Parento Maximum
☐ **Couple**	☐ **Couple**
☐ **Individual**	☐ **Individual**

☐ Only **common risks** of both partners

If both partners decide to check **only their common** reproductive risk, individual carrier status data will not be included or analysed in the test report.

Parento

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Genomed

ADDITIONAL INFORMATION AND DECLARATIONS

1. I acknowledge that Next-Generation Sequencing (NGS) will be used to analyse coding region (exon) sequences of selected 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. The test report will cover known pathogenic variants detected by NGS as well as pathogenic and likely pathogenic variants according to ACMG 2015 and ACGS 2020 guidelines.
2. I acknowledge that sequencing does not detect extensive gene deletions, duplications, or rearrangements. Such defects also constitute reproductive risk. Subject to data availability and quality, the PARENTO test will cover screening NGS data analysis for such alterations/structural alterations. The genes responsible for spinal muscular atrophy and congenital adrenal hyperplasia will be tested using MLPA, and the gene responsible for the fragile X syndrome will be tested using QF-PCR.
3. I acknowledge that the Parento test does not serve to evaluate the risk of future children having conditions that result from inherited or new chromosomal aberrations (such as the Down syndrome). Regardless of the test result, I am aware of the need to conduct prenatal screening tests for such conditions.
4. I agree that my 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 my 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).

☐I declare that before I agreed to have 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 *Patient's signature*

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

Name:		VAT ID:
Address:		Phone No.:
		E-mail:

CLINICALLY SIGNIFICANT INFORMATION

Are you aware of any genetic disease(s) in your family? ☐ YES ☐ NO

If the answer is YES, please name the disease(s) and degree of kinship with the person(s) who had them.

……

……

Have you had a bone-marrow transplant? ☐ YES ☐ NO

A bone-marrow transplant makes it impossible to test your blood and saliva.

Did you 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)

☐ venous blood ☐ saliva ☐ other ……