Genetic Centre
iVF Riga Holding

EXOME SEQUENCING

HEREDITARY RECESSIVE DISEASE PANEL

Ability to identify carrier status for recessive genetic disorders, allowing individuals or couples to make informed decisions about family planning, reduce the risk of passing on inherited diseases to their children, and access appropriate genetic counselling and reproductive options.

Indications:

- Family history of recessive genetic disorders
- Consanguineous relationships
- Ethnic or population-specific genetic risks
- Unexplained infertility or recurrent pregnancy loss
- Personal or Partner's Medical History

This knowledge empowers individuals to make informed decisions about their reproductive health and family planning.

Early Detection of Carrier Status - Risk Assessment for Baby - Guidance for Genetic Counselling

TEST FOR POOR RESPONDER - THE EARLY MENOPAUSE PANEL

Diagnostic test used to assess ovarian reserve and predict the likelihood of early menopause in women, particularly those who may be experiencing fertility challenges or are at risk of premature ovarian insufficiency

Indications:

- for women over 35 who are not yet planning a pregnancy
- if premature ovarian failure has been diagnosed – to understand how to act correctly for family planning

Early Detection of Ovarian Dysfunction - Assessment of Fertility Potential - Predictive Value for Early Menopause - Personalized Treatment Planning - Preservation of the genetic material

GENETIC PREDISPOSITION OF EMBRYONIC DEVELOPMENT ABNORMALITIES

Changes in the certain genes can be one of the reasons why egg fertilization is unsuccessful, the development of the embryo (blastocyst) stops or is arrested, or pregnancy does not occur, even if the embryo (blastocyst) has developed to the necessary stage for transfer.

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Indications:

- out of several eggs, only a few were fertilized, and the development of embryos (blastocysts) did not take place
- embryos (blastocysts) were growing until the third day, transfer was performed, but pregnancy did not occur
- embryo (blastocyst) development stopped until day 5 and transfer was not possible only a few embryos (blastocysts) from several fertilized eggs developed until the fifth day and the transfer was performed, but pregnancy did not occur

Assessment of Fertility Potential - Personalized Treatment Planning - Guidance for Genetic Counselling

TEST BEFORE TESE PROCEDURE - DISORDERS OF SPERMATOGENESIS PANEL

Panel offers valuable insights into the genetic factors contributing to male infertility

Indications:

- Non-obstructive azoospermia in patients with normal karyotype and negative Y chromosome deletions.
- These patients are usually referred to the TESE procedure with an average sperm retrieval success rate of 50%.

However, before the TESE procedure, an NGS can be suggested - for the targeted genes that are known to be associated with Sertoli cell-only syndrome, or sperm maturation arrest, or other spermatogenesis disorders that will result in negative TESE outcome. Therefore, it will assist to select the patients for whom TESE will not be effective and save them from unnecessary interventions.

Evaluation of Infertility - Personalized Treatment Planning - Risk Assessment for Baby - Guidance for Genetic Counselling

MOST COMMON HEREDITARY CANCER GENE PANEL: 33 GENES

Detects changes in 33 genes associated with hereditary tumour syndromes. The advantage of this panel is its wide diagnostic capabilities, which, when suspected of hereditary tumour syndromes, can not only determine changes in certain syndrome-related genes, but also expand the range of tested genes, including genes, whose changes can occur phenotypically as tumours less typical for the syndrome. This panel is recommended for hereditary breast / ovarian cancer, as well as in the case of various other tumours, such as Lynch syndrome, or hereditary nonpolyposis colorectal cancer, Li-Fraumeni syndrome, Cowden syndrome, Multiple endocrine neoplasia (MEN), various kinds of hereditary intestinal polyposis with high risk of malignancies, Neurofibromatosis type 1 (NF1). In terms of cost-benefit, the analysis is comparable to the complete single gene sequencing, while the information obtained is much broader and more comprehensive.

Blood For standard molecular examinations (ONCO panels, thrombophilia, CFTR, AZF, FRAXA, etc.),	Peripheral blood collected in a vacutainer containing EDTA	2-5 ml	· 1-7 days stored at +2 - +8°C · Storage for an indefinite time period ≤ -15°C · Transportation at +4°C or at room temperature

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