RĪGA STRADIŅŠ UNIVERSITY

***Genetic test for simultaneous detection of genetic variations important for reproduction***

Reproduction problems including infertility are important social health issues in many countries of the world. Data suggests that infertility affects around 15% of all couples. Of which ~15% of male infertility has roots in genetic problems and ~10% of female infertility are genetic (Foresta et al., 2002). In Latvia, ~1100 in-vitro fertilization (IVF) procedures are performed every year. In 2009, 537 463 medicinal fertilization procedures were performed in Europe, and this number is still growing, therefore reproductive medicine is an industrial branch with a high export potential.

Choosing effective strategies to treat infertility depends on the results of the tests performed on the patient – including genetic test results. Since reproduction problems are caused by many genetic factors, it is very important to elucidate the exact causes of infertility. Genetic testing is especially important in in choosing the best method of fertilization and to improve the chance of the procedure's success. It is also important when the parents, or germ cells donors, have a genetic pathology, and so to avoid passing it on to offspring – this means the birth of a healthy child.

Today, because there are no tests to assay for the set of the most important genetic variations associated with infertility, genetic testing in infertility is done in a stepwise fashion. Therefore, multiple tests are usually done, and often using several different laboratory machines. This increases the costs and time spent on testing, because these tests can often not be performed at the same laboratory. These high costs and technological limitations can lead to incomplete genetic testing; so many infertility patients risk not finding out the exact cause of their infertility and may lack an appropriate treatment strategy.

The use of diagnostic tests currently available in the market is limited, because the effectiveness of these tests is low – each test can detect only a few genetic variations that are involved in reproduction. High costs are a concern when it comes to detecting all known genetic variations that are involved in reproduction – to detect these variations, multiple tests have to be purchased, as no single test covers all or even the majority of these genetic variations.

Researchers at the RSU Scientific Laboratory of Molecular Genetics have developed a new method – a genetic test:

- which simultaneously detects 33 different genetic variations, thus diagnosing the most frequent and most important genetic causes of infertility and providing information for making decisions for an optimal infertility treatment method;
- which detects genetic variations for both male and female caused failure of reproduction;
- which is used in any standardized molecular biology laboratory allowing to perform the test at a laboratory during a single day.

Our test combines the most frequent genetic markers for male and female infertility, the most frequent cystic fibrosis mutations, as well as sex-specific markers (which allow detection of changes in sex chromosome count – e.g., Klinefelter syndrome), and genetic variations that cause inherited thrombophilia, which is an important cause of negative pregnancy outcomes, and other genetic variations that may affect fertility.

*Current stage*

A new method – a genetic test has been developed, that:

1) has been partially validated and approbated by repeated testing of the results for 100 individuals, using various thermocyclers and capillary electrophoresis machine models;
2) has been tested via a number of external quality assurance schemes – results have been obtained, including certificates from organisations issuing various external quality assurance schemes);
3) has been compared with other similar commercially available genetic tests;

The target market is persons with reproduction problems, which would require genetic testing. In accordance with newest data, there are 15 000-20 000 infertile couples in Latvia, and of these, 15-20%[1] of all cases have no known cause of infertility, whereas the worldwide estimate of infertile couple count is ~48,5 million (WHO data, 2012).

[1] http://polsis.mk.gov.lv/view.do?id=3583