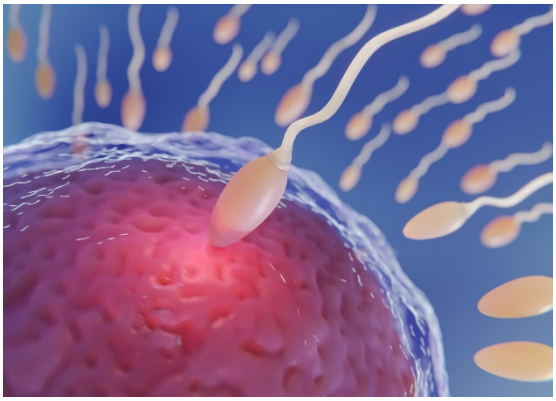
DNK s.r.o.
diagnostické centrum

Laboratory diagnosis of genetic factors of male infertility

Diagnostic kit for detection of deletions of Y-chromosome AZF locus by real-time PCR

Microdeletions in the long arm of the Y chromosome, along with Klinefelter syndrome, are the most common genetic causes of severe forms of male infertility, such as oligozoospermia and non-obstructive azoospermia.

On average, in men with an unknown cause of infertility, the detection rate of microdeletions on the Y chromosome is about 2%. At the same time, in men with azoospermia, the proportion of microdeletions is higher (10% or more) than in men with oligospermia.



Y chromosome deletions

There are three areas on the Y chromosome in which deletions are associated with male infertility: AZFa, AZFb, AZFc (AZF — azoospermia factor). Deletions of AZF areas differ in frequency of occurrence and lead to various disorders and their manifestations:

- **AZFa deletions** – *Sertoli cell only syndrome* (SCOS), complete azoospermia (detection rate – 3%);
- **AZFb deletions** – cessation of spermatogenesis, complete azoospermia (detection rate — 9%);
- **AZFc deletions** – hypospermatogenesis and varying degrees of oligospermia (detection rate – 79 %).

The European Association of Andrologists (EAA) and the European Molecular Genetic Research Quality Network (EMQN) have developed guidelines "Practical guidelines for the molecular diagnosis of microdeletions in the Y-chromosome" (2004).

It is recommended to use highly conserved unique DNA sequences – STS markers – to detect deletions.
For each region, 2 markers are determined (AZFa - markers sY84 and sY86; AZFb - sY127, sY133 or sY134; AZFc - sY254 and sY255). In the case of deletion, both markers should not be detected.
The control region of the Y chromosome (Sex-determining region Y - SRY) and the region present in both sexes (control of sufficient amount of human DNA for analysis – HMBS gene) were also determined.

Indications for molecular-genetic testing

EAA recommends that all men with azoospermia and severe oligospermia should be tested for Y microdeletion (sperm concentration less than 5 million/ml):

- when examining an infertile couple
- when choosing how to eliminate infertility and assessing the need for invasive procedures
- when assessing the risk of fertility disorders in sons

Diagnostic kit RealBest-Genetics AZF microdeletions (Cat. No. 3814)

It was developed taking into account the recommendations of the "Practical Guide for the Molecular Diagnosis of Microdeletions in the Y Chromosome" (2013) and allows to identify all the necessary markers: 2 STS markers for the AZFa, AZFb and AZFc regions, as well as the control region of the Y chromosome and the HMBS gene as a check for the presence of human DNA.

Microdeletion detection method – multiplex real-time PCR with fluorescence detection

The kit is designed for the analysis of 48 samples including control and is intended for use with devices CFX96 (Bio-Rad, USA), DT-96 a DTprime (DNA-Technology, Russia), Gentier 96/R (Xi' TianLong, Science and Technology Co., Ltd., China)

Features and advantages of the diagnostic kit

- Ready Master Mix for PCR: Simplification of analysis procedures and high stability of test quality
- Multiplex: use of only two tubes of RMM per patient to test for all markers
- Specimens: whole blood or buccal epithelium
- High stability of the set: storage at a temperature of 2–8 ° C; transport up to 26 °C for not more than 10 days

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