A considerable number of couples spend years suffering from infertility without proper emotional support or cost-effective treatment options. Identifying the cause of infertility opens up access to personalized, informed clinical management, along with professional counselling.

Genetic testing can be the key for a significant number of infertile couples trying to have children. Understanding the reason for infertility facilitates informed decisions and family planning. Counselling of parents can explain any potential to transmit genetic abnormalities that may affect the health of their offspring.

Benefits of genetic testing

CENTOGENE offers three different infertility gene panels designed by our medical experts, encompassing common genetic causes of infertility and based on comprehensive literature reviews. The highest CAP/CLIA certified testing quality and a turnaround time of 25 days are the hallmarks of CENTOGENE’s superior service.

Infertility prevalence

- Infertility affects nearly 15% of couples wishing to conceive and is generally attributed equally to males and females
- In about 10% of infertility cases there is a genetic etiology
- In patients with a combination of symptoms it is very difficult to narrow down hypotheses for specific underlying genetic causes. Performing numerous physical tests is costly and time consuming. More importantly, a delay in diagnosis and treatment has a dramatic negative impact on the patient’s quality of life.

Genetic testing at CENTOGENE

- CENTOGENE pays extraordinary attention to detail in the critical scientific and medical interpretation of genetic test results. Accurate technical processing of patient samples is only the first step.
Genetics of Infertility

In males, genetic abnormalities may cause infertility by affecting sperm production or sperm transport. The most common genetic causes of male infertility are cystic fibrosis gene mutations, chromosomal abnormalities, and Y-chromosome microdeletions.

In females, genetic abnormalities may cause infertility by affecting their reproductive cycle and/or hormonal balance. The most common genetic causes of female infertility are chromosomal abnormalities and mutations in the FMR1 gene.

When should I advice this panel?

We recommend you to advice the infertility panel in the following circumstances:

- Failure to establish a pregnancy after trying to conceive for six months or longer
- Irregular or absent menstruation
- Low sperm count, abnormal form or movement
- Small or irregular genitals and secondary sexual features (hair, breasts, etc.)
- Known fertility problems
- A history of >1 miscarriage

Diagnostic strategy

WHOM TO TEST?

<table>
<thead>
<tr>
<th>Men</th>
<th>Go for testing</th>
</tr>
</thead>
<tbody>
<tr>
<td>Men/Women</td>
<td>Male infertility panel</td>
</tr>
<tr>
<td>Women</td>
<td>Female infertility panel</td>
</tr>
</tbody>
</table>

TESTING STRATEGY

<table>
<thead>
<tr>
<th>Genetic alteration!</th>
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</thead>
<tbody>
<tr>
<td>Go for action</td>
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</tbody>
</table>

MALLE infertility panel

AR, CATSPER1, CFTR, FSHR, LHCGR

Global infertility panel

AR, CATSPER1, CFTR, FSHB, FSHR, HESX1, LHB, LHCGR, NR5A1, POU1F1, SRY

Female infertility panel

BMP15, CYP21A2, FSHR, LHB, LHCGR, ZP1

ACTION

Genetic counseling
Adapt treatment

Turnaround time: 25 days