Infertility panel
GENETIC TESTING

CENTOGENE
THE RARE DISEASE COMPANY

Infertility prevalence

- Infertility affects nearly 15% of couples wishing to conceive* and is generally attributed equally to males and females
- Infertility is suspected to have a genetic cause in 15% of males and 10% in females.
- 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.

Benefits of genetic testing

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 children.

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 a year 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

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.

Diagnostic strategy

WHOM TO TEST?	TESTING STRATEGY	ACTION
Men Men/Women Women	**Infertility Panel** *ANOS1, AR, ARL13B, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BMP15, C8ORF37, CATSPER1, CC2D2A, CCDC28B, CEP164, CEP290, CFTR, CHD7, DUSP6, ENPP1, FEZF1, FGF17, FGF8, FGFR1, FLRT3, FMR1, FSHB, FSHR, GNRH1, GNRHR, HESX1, HEXA, HFE, HS6ST1, IFT172, IFT27, IL17RD, INPP5E, KIF7, KISS1, KISS1R, LEP, LEPR, LHB, LHCGR, LHX3, LHX4, LZTFL1, MKKS, MKS1, MYO7A, NPHP1, NPHP3, NR0B1, NR0B2, NR5A1, NSMF, OFD1, PCSK1, PHF6, PNPLA6, POLR3B, POMC, POU1F1, PPARG, PROK2, PROKR2, PROM1, PROP1, PRPH2, RDH5, RHO, RLBP1, RPGRIP1L, SDCCAG8, SEMA3A, SOX10, SOX2, SOX3, SPRY4, SRY, TAC3, TACR3, TMEM67, TRIM32, TTC21B, TTC8, TUBB8, WDPCP, WDR11, ZP1* Complementary assays: Repeat expansion analysis for *AR, FMR1* and *MLPA* for aneuploidy and *AZF* region.	Genetic counseling Adapt treatment

Turnaround time: **25 days**

*Foresta, C., Ferlin, A., Gianaroli, L. et al. Guidelines for the appropriate use of genetic tests in infertile couples. Eur J Hum Genet 10, 303–312 (2002). https://doi.org/10.1038/sj.ejhg.5200805

CONTACT AND CUSTOMER SERVICE

Phone: +49 (0)381 80 113-416
Fax: +49 (0)381 80 113-401
Email: customer.support@centogene.com
www.centogene.com

CLIA #99D2049715

CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

ISO 15189 CAP ACCREDITED COLLEGE of AMERICAN PATHOLOGISTS

1.8eng_November2020