Y Chromosome Microdeletion

REFERRAL REASON

Genetic screening test for Y chromosome microdeletion.

TEST RESULTS

Y chromosome microdeletion not detected in this individual.

INTERPRETATION

Molecular investigation results show that Y chromosome microdeletion (AZFa, AZFb and AZFc region) is absent in this individual.

RECOMMENDATIONS

1. Genetic counseling
2. Further correlation of the test results with clinical symptoms, family history and other laboratory findings. Additional testing is required in case of strong suspicion.

METHOD

Polymerase chain reaction (PCR) followed by agarose gel electrophoresis.

BACKGROUND

Most commonly detected abnormalities in men with azoospermia (complete absence of sperm) and severe oligospermia (sperm count less than 5 million/ml) are Y chromosome microdeletions. Three common regions AZFa, AZFb, and AZFc on the long arm of Y chromosome are frequently micro deleted in infertile men. Y chromosome STS markers tested include: AZFa: sY84, sY86; AZFb: sY127, sY134 and AZFc: sY254, sY255 (EAA/EMQN guidelines, 2004)1.

- **AZFa deletion:** result in spermatogenic failure (Sertoli-cell-only syndrome, SCOS).
- **AZFb deletion:** result in azoospermia/spermatogenetic arrest.
- **AZFc deletion:** most commonly found microdeletion with variable phenotype ranging from oligo spermiato azoospermia and SCOS.

LIMITATIONS

1. Errors in our interpretation of results may occur if information given is inaccurate or incomplete.

2. This testing method does not exclude the possibility of other genetic causes of infertility
3. Breakpoints of identified microdeletions will not be determined
4. All laboratory investigations are performed to support diagnosis in patient care and they carry some limitations due to Sensitivity and specificity of assay procedures and also depend on the quality of the sample received by the laboratory. Results should be correlated with clinical history, life style and medication given to the individual.
5. *It is presumed that the specimen used to perform the test belongs to the patient specified above, such verification having been carried out at the collection level of sample.

REFERENCE

Simoni M, Bakker E, Krausz C (2004) EAA/EMQN best practice guidelines for molecular diagnosis of Y-chromosomal microdeletions. State of the art 2004. Int J Androl 27: 240–249.

*** End Of Report ***