Elucigene®

Clarity without complexity

Molecular Diagnosis of Male Factor Infertility

A single workflow solution for the molecular diagnosis of Male Factor Infertility

- **Y-chromosome microdeletion analysis**
- **Extension analysis of microdeletions**
- **Sex chromosome aneuploidy analysis**
- **Investigation of Cystic Fibrosis related CBAVD**

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CE

Male Factor Infertility

Identifying the genetic causes of male infertility can result in more effective clinical management of patients. Statistically, 10-15% of couples experience difficulty in conceiving, with male related factors e.g. low sperm count, thought to be an underlying issue in approximately 50% of cases. Although in the majority of incidences the causes of male infertility are unknown, studies have shown that sex chromosome aneuploidy and microdeletions of specific regions of the Y-chromosome can play a role.

Klinefelter syndrome is the most common sex chromosome aneuploidy associated with male infertility. This syndrome has a live birth incidence of between 1:500 and 1:650 males with the most common cause being an additional copy of the X chromosome (47, XXY karyotype).

Y-chromosome microdeletions are the next most common genetic cause of male infertility, with microdeletions occurring in three regions (AZFa, AZFb, AZFc) detected in up to 7% of oligospermia (low sperm count) and 13% of nonobstructive azoospermia cases. These microdeletions occur due to homologous recombination of repetitive sequences in these regions and the exact molecular mechanisms and recombination events underlying these changes have been elucidated. These regions are located at chromosome Yq11 and although the AZFa microdeletion region is distinct, there is a significant degree of overlap between the regions affected by AZFb and AZFc microdeletion.

Recommended analytic steps for Y-chromosome microdeletions

The European Academy of Andrology (EAA) and European Molecular Genetics Quality Network (EMQN) have published a series of best practice guidelines for Y-chromosome microdeletion testing. In 2013, these guidelines were amended to include extension analysis which provided further characterisation and sizing of detected AZF region microdeletions using a separate defined set of markers.

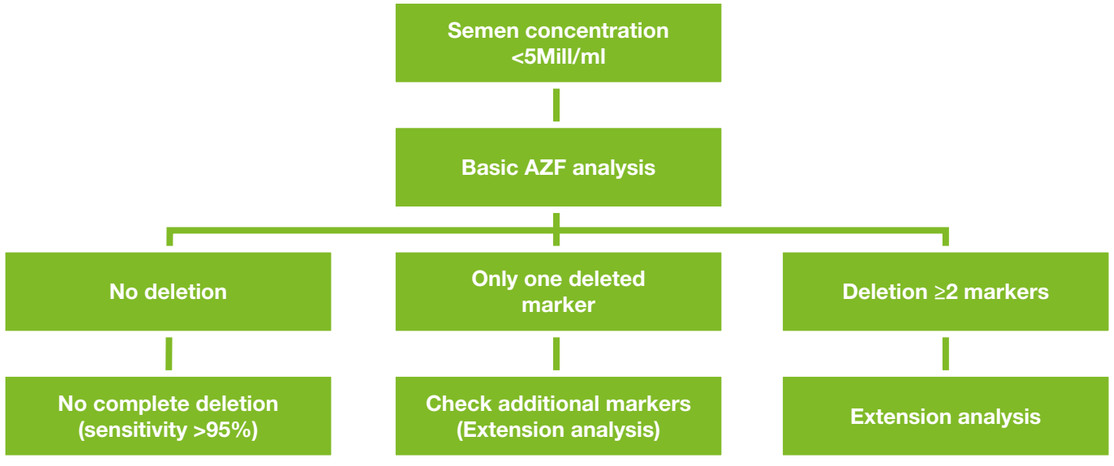

Figure 1: Summary of flowchart showing recommended diagnostic steps for Y-chromosome microdeletion analysis (Krausz et al., 2014).

Male Factor Infertility Kit

The Male Factor Infertility Kit detects both sex chromosome aneuploidy and Y-chromosome microdeletions in a single tube using QF-PCR (Quantitative Fluorescence-Polymerase Chain Reaction) technique and employing a CE Genetic Analyzer platform. The Male Factor Infertility Kit uses EAA/EMQN prescribed markers and primers to detect Y-chromosome microdeletions affecting the AZFa, AZFb and AZFc regions in accordance with best practice guidelines.

Single tube assay:

- AMEL, TAF9 and X and Y specific markers for sex chromosome aneuploidy
- Y-chromosome microdeletion markers
- Quantitative analysis on CE Genetic Analyzer

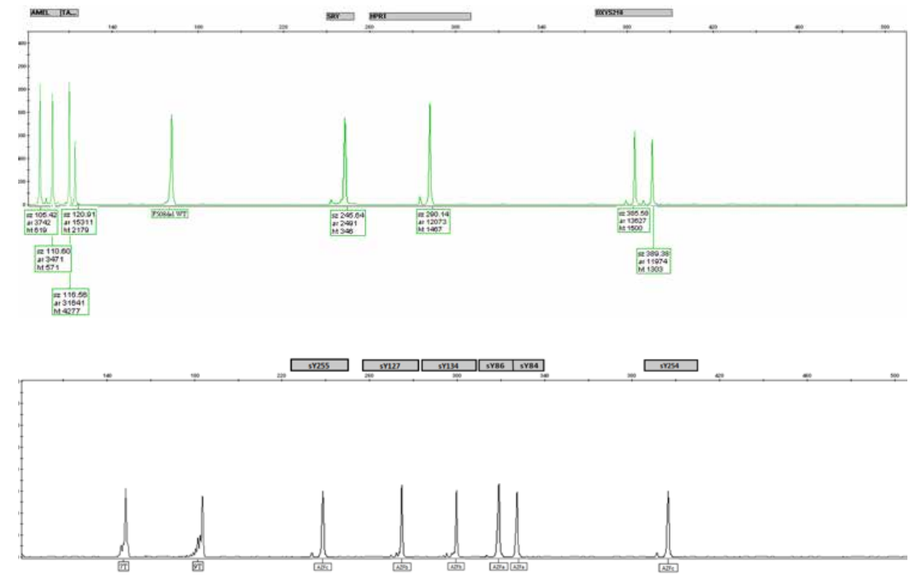

Figure 2: Typical normal Male Factor Infertility Kit result

MFI-Y*plus* Kit

The MFI-Y*plus* Kit is a single tube extension assay used in conjunction with the Male Factor Infertility Kit which contains 11 additional markers for characterisation of Y-chromosome microdeletions in line with published guidelines.

The Elucigene MFI-Y*plus* Kit runs on the same CE Genetic Analyzer platform as the Male Factor Infertility Kit using the same running conditions.

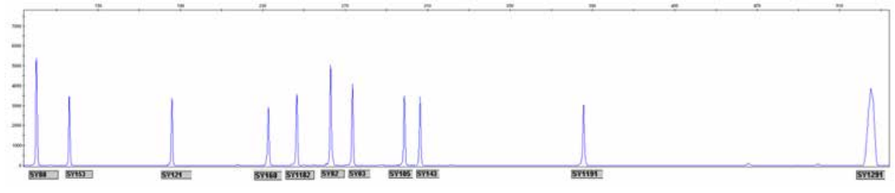

Figure 3: Typical normal MFI-Y*plus* Kit result

CF-EU2v1 50 Mutation Kit

The CF-EU2v1 provides a convenient simple solution for male infertility screening for congenital unilateral absence of vas deferens (CUAVD) and congenital bilateral absence of vas deferens (CBAVD) as part of a Male Factor Infertility screening workflow.

The CF-EU2v1 is the only commercially available pan-European cystic fibrosis testing kit designed specifically to address the most common mutations found across populations of European origin.

The assay identifies 50 mutations in total and also analyses the intron 8 polyT tract with accurate measurement of the adjacent TG repeat. CF-EU2v1 uses quantitative analysis on CE Genetic Analyzer using the same running conditions as the Male Factor Infertility and MFI-Y*plus* Kit.

Ordering information:

Catalogue No.	Product	Product Description	Kit Size
AZFXYB1	Male Factor Infertility Kit	Sex chromosome aneuploidy and Y-chromosome microdeletion analysis	25 Tests
AZFPLBX	MFI-Y*plus*	Extension kit for the characterisation of Y-chromosome microdeletions	10 Tests
CF2EUB2	CF-EU2v1 Kit	Identifies 50 CFTR gene mutations within populations of European origin	50 Tests

Licensee Kits

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