



ampli set Y Chromosome Extension^{CE IVD} 24 tests cat 1502

confirmation and analysis of extension of AZF regions microdeletions of Y Chromosome

Y Chromosome microdeletions of AZF regions (AZoospermia Factor) are observed in 10-15% of azoospermic men and 5-7 % of oligospermic patients. Many genes in every AZF region have been identified (DBY, USP9Y RBMY1, eIF1AY, DAZ, GOLG, BPY2 etc), but it isn't clear which of them is involved in spermatogenesis. Deletions of regions of the long arm of Y Chromosome can occur and partial microdeletions or deletions of single genes are rare (1,2).

Ampli set Y Chromosome Extension completes the **ampli set Y Chromosome UE** since allows to confirm, using the PCR technique, microdeletions identified by the first step kit and to analyze the entire deleted region. **Ampli set Y Chromosome extension** kit is made of two mix PCR: 1) "Confirmation Mix" and 2)"Extension Definition Mix" of the microdeletions. The first group includes three mix-PCR, each allows the amplification of 1 STS inside one of the AZF regions (for AZFa: sY84: for AZFb: sY127; for AZFc: sY254). As a deletion is showed by the absence of PCR product, the mix PCR in the kit contains a primers pair specific for ZFX/ZFY genes that always produces an amplification product (internal PCR product). The purpose of this First Step PCR is the confirmation of the deletion detected with the **ampli set Y Chromosome UE**. The second group of mix PCR (Extension Mix PCR) allows the amplification of STS in the proximal and distal border of every AZF region. As the clinical significance of the deletion extensions (4,5), this second amplification, performed on the deleted samples obtained by the Confirmation PCR, allows to assess if a deletion is *complete* or *partial*. The choice of the markers is based on the directions of European Academy of Andrology Guide Lines (1) and on the data of recent literature.(2,6)

Principle of method: A) extraction of genomic DNA; B) amplification; C) detection on agarose gel
Applicability: on extracted and purified genomic DNA from whole blood samples or buccal cells.

ANALYSIS OF RESULTS

PCR products, fragments of 126-495 bp of the two PCR, can be separated on agarose gel electrophoresis. The absence of PCR products of specific regions of Y chromosome shows the presence of a microdeletion of the sequence.



	Confirmation mix	Extension Mix
AZFa	sY84 ZFX/ZFY	sY82 sY83 sY87 sY88
AZFb	sY127 ZFX/ZFY	sY105 sY114 sY143 sY1197 sY152
AZFc	sY254 ZFX/ZFY	sY143 sY158 sY160

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