



ampli set Y Chromosome UE^{CE IVD} 24 tests cat 1501

detection of AZF regions microdeletions of Y Chromosome

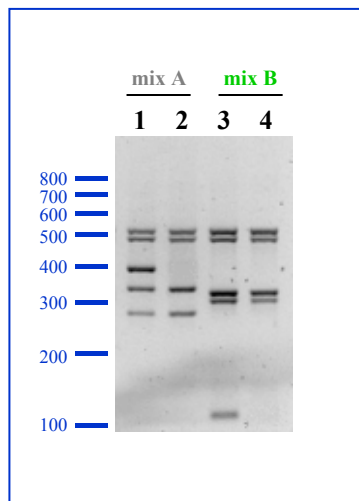
Y Chromosome microdeletions of AZF regions (AZoospermia Factor) are observed in 10-15% of azoospermic men and 5-7 % of oligospermic patients. Many genes in every AZF region have been identified (DBY, USP9Y RBMY1, eIF1AY, DAZ, GOLG, BPY2 etc), but it isn't clear which of them is involved in spermatogenesis.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

The **ampli-set Y Chromosome UE** allows to detect, using the Polymerase Chain Reaction (PCR), Y chromosome microdeletions inside the three AZF regions (AZoospermia Factor) AZFa, AZFb, AZFc. The Multiplex PCR mix (First Step M-PCR) inside the kit allow the amplification of the "sequence tagged sites" (STS) assessed by the Guide Lines of the European Academy of Andrology (3) and let the detection almost 100% of the deletion which have a clinical significance. As a deletion is showed by the absence of PCR product, the mix PCR in the kit contain a primers pair specific for ZFX/ZFY genes that always produce an amplification product (internal PCR product) and a primers pair specific for SRY gene on the short arm of Y chromosome as "testis determining factor" control.

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.



1 - 3 Normal male DNA control
 2- 4 Azoospermic male with deletion of AZFc region



AZFa	sY84 sY86 ZFX/ZFY SRY
AZFb	sY127 sY134 ZFX/ZFY SRY
AZFc	sY254 sY255 ZFX/ZFY SRY

M-PCR mix A

ZFY 495 bp
 SRY 472bp
 sY254 400bp (AZFc)
 sY84 326bp (AZFa)
 sY127 274bp (AZFb)

M-PCR mix B

ZFY 495 bp
 SRY 472bp
 sY86 320bp (AZFa)
 sY134 301bp (AZFb)
 sY255 126bp (AZFc)

REFERENCES

- 1) Kamp C et al. *Hum. Mol. Genet.* 2000 9:2563-72.
- 2) Repping S. et al. *Am. J Hum Genet.* 2002 71:906-22
- 3) Simoni M. *Int J Androl.* 1999 22:292-9.