

DETECTION OF AZF REGIONS MICRODELETIONS of Y CHROMOSOME

AMPLI-SET Y Chromosome UE

Cat. n.1.501

Y Chromosome microdeletions of AZF regions (AZoospermia Factor) are observed in 10-15% of azoospermic men and 5-10% 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 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) amplifies the "sequence tagged sites" (STS) assessed by the Guide Lines of the European Academy of Andrology (3); in this way the detection of almost 100% of the deletion having a clinical significance is possible. 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, always producing 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 Assay: A) extraction of genomic DNA B) amplification C) detection on agarose gel.

Applicability: On extracted and purified genomic DNA from whole blood samples.

Numbers of Tests: 24

REAGENTS and STORAGE

AMPLIFICATION	
M-PCR mix A	-20°C
M-PCR mix B	-20°C
H ₂ O sterile	-20°C
Taq Polymerase (5U/μl)	-20°C
Male Genomic DNA Positive Control	-20°C

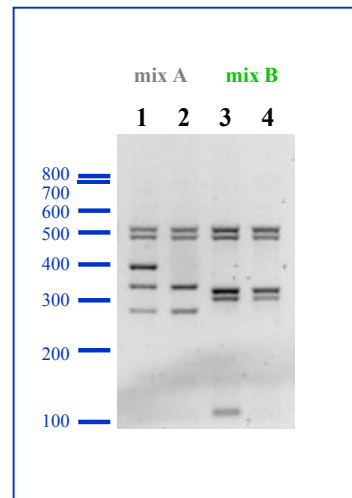
Stability: over 12 months if correctly stored (Agarose gels, if protected by light, can be stored 1 year at room temperature).



	STS	M-PCR Mix
AZF_a	sY84 sY86 ZFX/ZFY SRY	A B
AZF_b	sY127 sY134 ZFX/ZFY SRY	A B
AZF_c	sY254 sY255 ZFX/ZFY SRY	A B

ANALYSIS OF RESULTS

PCR products 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

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.