DETECTION OF THE CAG TRINUCLEOTIDE REPEAT POLYMORPHISM IN THE ANDROGEN RECEPTOR (AR) GENE

AMPLI-SET-AR Cat. n. 1.503

The pathogenesis of many male fertility disturbances has not yet been elucidated. It has been estimated that genetic alterations may be involved in a significant percentage of men with severe impairment of spermatogenesis. Efficient spermatogenesis requires high levels of intratesticular testosterone and a functional androgen receptor. The androgen receptor (AR), encoded by a single-copy gene on the X chromosome, is a transcription factor that mediates the action of androgens in target cells. The androgen receptor has three main functional domains: the amino terminal transactivation domain, the DNA-binding domain, and the steroid-binding domain. The transactivation domain contains a polymorphic trinucleotide repeat segment (CAG)n, that encodes a polyglutamine tract.
The increase in CAG repeat sequence, more than 40 CAG, has been found to lead to spinal and bulbar muscular atrophy (Kennedy disease). More than 50% of patients with this disease have clinical signs of infertility associated with severe oligospermia or azoospermia. In contrast, a relatively short CAG repeat has been associated to an increased risk of prostate cancer, an androgen-dependent tumor., Thus, short CAG repeats increase androgen receptor androgenicity, resulting in abnormally high stimulation of prostatic tissue. At the other end of the spectrum, preliminary data indicate that long CAG repeats, while still within the polymorphic range, can reduce receptor androgenicity and lead to male infertility in some but not all populations.
The Kit AMPLI-set-AR enables the detection of CAG repeat expansion in exon 1 of the androgen receptor gene by polymerase chain reaction (PCR) using primers designed at opposite sides of the CAG repeat.

Principle of Assay: A) extraction of genomic DNA B) amplification C) detection on polyacrylamide gel.
Applicability: On extracted and purified genomic DNA from peripheral blood samples.
Numbers of Tests: 24

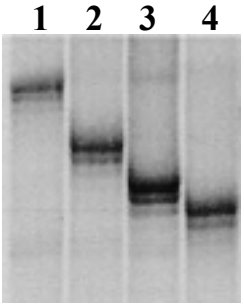
REAGENTS and STORAGE

AMPLIFICATION	
PCR mix	-20°C
Water DNase-RNase free	-20°C
Taq Polymerase (5U/µl)	-20°C
Control DNA	-20°C

Stability: over 12 months if correctly stored.

ANALYSIS OF RESULTS

Representative denaturing polyacrylamide gel electrophoresis of polymerase chain reaction products spanning the CAG repeat segment in exon 1 of androgen receptor gene (AR).



1) PCR product: 306 bp contain **28** CAG repeats
2) PCR product: 300 bp contain **26** CAG repeats
3) PCR product: 297 bp contain **25** CAG repeats
4) PCR product: 294 bp contain **24** CAG repeats

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