IDENTIFICATION OF POLYMORPHISMS IVS1 -397 T>C of ESR1 gene and 1730 A>G (*39 A>G) of ESR2 gene

AMPLI-397 T>C ESR1 e *39 A>G ESR2 RT Cat. n.1.508 RT

The estrogens control many cellular processes, the differentiation and the reproductive system functionality. In the females, the main estrogens targets are the ovaries, uterus, vagina and mammary glands. In the male, the estrogen targets are testicles, prosate and epididymis. The estrogen is responsible for the growth and skeleton mainteinance and normal functioning of cardio-vascular and nerve systems. The estrogens function thanks to the receptor (ER).
The estrogen receptor occurs in two different isoforms ER-beta e ER-alpha, coded by two different genes (**ESR2** and **ESR1**) having different abilities in bonding with the ligand and in transducing the regulatory signal for the transcription of the target genes.
The gentotypic combinations of SNP explain the variability of the receptors in qualitative and quantitative terms. In the **ESR1** (6q25) gene different polymorphisms have been described but the most studied is the -397 T/C polymorphism located in the gene intron 1. This polymorphism (rs2234693) is called ***PvuII*** polymorphism because recognized by ***PvuII*** and classified in **P-p**, depending on the presence or absence of restriction site. The T nucleotide is also defined as ***allele p***, while the nucleotide C is defined ***allele P***. the genotype PP (CC) is associated to a receptor dysfunction with a reduced estrogen response.
In the **ESR2**, the polymorphism is located in the **3'UTR** region of the gene, in the 1730 nucleotide (**1730 A→G**) (rs1256049), and it is recognized by the restriction enzyme ***AluI***. This polymorphism is also known as ***39 A→G**. The genotype GG is associated to a reduced estrogen response.
Multilocus association studies have demonstrated a genetic interaction between FSHR gene polymorphisms ESR1 and ESR2 related to the results of controlled ovarian stimulation (COH). The optimization of the in vitro fertilization (IVF) depends on the ovarian response to the exogenous administration of gonadotrophin, like FSH. Some patients, undergoing to multiple induction therapies, are not able to mature a sufficient number of follicles and they are classified as "poor responders". A trigenic profile in homozygosity of receptor polymorphisms for FSH in (680AA) and estrogen receptors, ESR1 (CC) and ESR2 (GG), seem to represent a risk factor because they reduce the response to the FSH stimulation.

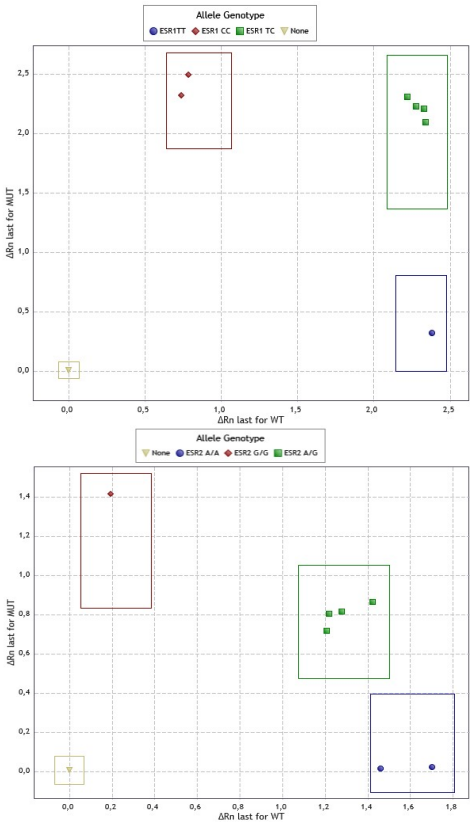
Principle of assay: A) extraction of genomic DNA; B) amplification and detection using real-time PCR equipment;
Applicability: of genomic DNA extracted and purified from whole blood samples and tissue samples from fresh and paraffin.
Numero di Test: 25x2.

REAGENTS and STORAGE

AMPLIFICATION	
PCR mix 2X	+4°C
Primer-probe ESR1 -397 T>C mix 20 X	-20°C in the dark
Primer-probe ESR2 39 A>G mix 20 X	-20°C in the dark
H_20 RNase/DNase-FREE	-20°C
Control A/G ESR2 39 A>G	+4°C
Control GG ESR2 39 A>G	+4°C
Control AA ESR2 39 A>G	+4°C
Control TC ESR1 -397 T>C	+4°C
Control CC ESR1 -397 T>C	+4°C
Control TT ESR1 -397 T>C	+4°C

Stability: over 18 months if correctly stored.

ANALYSIS OF RESULTS



References:
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