

Title (en)

ESTROGEN RECEPTOR ALPHA VARIANTS AND METHODS OF DETECTION THEREOF

Title (de)

ÖSTROGENREZEPTOR ALPHA VARIANTEN UND VERFAHREN ZUR DETEKTION DERSELBEN

Title (fr)

VARIANTS DE RECEPTEURS OESTROGENIQUES ALPHA ET PROCEDE DE DETECTION DESDITS VARIANTS

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Application

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Abstract (en)

[origin: WO0162969A2] The present invention is based on sequencing genomic DNA from human chromosome 6 and cDNAs to define the genomic structure of estrogen receptor alpha genes and novel polymorphism/haplotypes in the estrogen receptor gene/protein. Such polymorphism/haplotypes can lead to a variety of disorders that are mediated/modulated by a variant estrogen receptor, such as a susceptibility to cancer, osteoporosis, cardiovascular disorder, etc. Based on this sequencing approach, the present invention provides genomic nucleotide sequences, cDNA sequences, amino acid sequences and sequence polymorphism/haplotypes in the ESR-alpha genes, methods of detecting these sequences/polymorphism/haplotypes in a sample, methods of determining a risk of having or developing a disorder mediated by a variant estrogen receptor and methods of screening for compounds used to treat disorders mediated by a variant estrogen receptor.

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