

Relationship between androgen receptor gene structure and protein function. Structural gene defects that promote androgen insensitivity in man Estructura del gen del receptor de andrógeno y función de la proteína. Defectos estructurales del gen que determ

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The human androgen receptor is a member of the superfamily of steroid hormone receptors and contains three functional domains: an amino-terminal region involved in the expression of androgen regulated genes, a central cystein-rich DNA binding region and a carboxy-terminal hormone binding region. Proper functioning of this protein is a prerequisite for normal male sexual differentiation and development. Androgen action is currently studied in vitro, using fibroblasts culture from genital skin and complementary DNA of the androgen receptor gene has been recently cloned and sequenced. During recent years a substantial progress has been made elucidating the structure-function relationship of the androgen receptor and the characterization of the molecular defects associated with androgen insensitivity syndromes. There appears to be a broad correlation between the degree of receptor dysfunction caused by the mutation and the patient phenotype.