

CAG repeat polymorphism of androgen receptor gene and X-chromosome inactivation in daughters of women with polycystic ovary syndrome (PCOS): Relationship with endocrine and metabolic parameters

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Background: The polycystic ovary syndrome (PCOS) is a hyperandrogenic disorder that arise from a combination of genetic and environmental factors. **Aim:** To assess the role of the androgen receptor (AR) CAG repeat polymorphism in the metabolic and reproductive features in daughters of women with PCOS (PCOSd). **Methods:** Sixty-seven PCOSd and 60 daughters of control women (Cd) were studied in early stages of sexual development. Sex steroids, glucose, insulin and lipids were determined. The AR CAG repeat sizes and X-chromosome inactivation (XCI) were analyzed. **Results:** PCOSd and Cd had similar mean number of CAG repeats and XCI pattern. In PCOSd and Cd, methylation-weighted biallelic means CAGn (mwCAGn) was not associated with androgen levels. In infants and pubertal PCOSd, mwCAGn was associated with a low concentration of HDL-cholesterol. **Conclusions:** AR CAG repeat polymorphism appears to be unrelated with serum androgen levels. However, the short mwCAGn variant may have a possible impact o