INTRODUCTION

46, XY gonadal dysgenesis is a disorder of sex development that presents the following characteristics:

- Chromosomal pattern: 46, XY
- Normal external female genitalia
- Streak gonads
- Presence of uterus and Fallopian tubes

DIAGNOSIS: At the age of puberty

Absence of thelarche and primary amenorrhea

As the phenotype during childhood is of normal female, the patients are raised as females, and once the diagnosis is established, hormonal replacement is initiated in order to induce pubertal development.

The gonad forms as a bipotential structure during early embryogenesis and differentiates into a testis in case of a 46,XY karyotype with an intact SRY gene. The testicular production of testosterone and AMH leads to the development of male internal and external genitalia.

In case of gonadal dysgenesis in 46,XY subjects, testis development is impaired, and the absent or reduced production of testosterone and AMH will lead to the formation of female external genitalia or ambiguous genitalia.

PURPOSE

The description of the rare case of two sisters affected of 46, XY complete gonadal dysgenesis and gonadoblastoma, in whom SRY mutations were identified.

PATIENTS AND METHODS

An 11-year-old girl was referred after she palpated an abdominal mass. She was born from non consanguineous parents and her past medical history was unremarkable.

Physical exam: B I- PH II- normal external female genitalia, FSH:126.3 IU/L - LH: 30.25 IU/L

KARYOTYPE: 46, XY

Left ovarectomy was performed and pathology also revealed gonadoblastoma.

She was treated with chemotherapy, and she is currently in good health, receiving estrogen/progesterone.

Family chromosomal pattern:
- Mother: 46,XX
- Father: 46,XY
- Sister: 46, XY

Molecular analysis of the Y chromosome: deficit of the HMG region of the SRY gene

CONCLUSION

46, XY complete gonadal dysgenesis is a disorder of sex development characterized by streak gonads, external female genitalia, presence of uterus and fallopian tubes but absence of menstruation and secondary sex characteristics.

The first candidate gene that is evaluated is usually SRY because mutations in the other genes typically cause defects in additional organ systems. However, SRY mutations are identified in only 15% of patients.

There is a high probability of gonadal malignancy thus bilateral gonadectomy is strongly recommended, however there is no consensus regarding the optimal timing.

- (J Clin Endocrinol Metab, August 2007, 92(8):3305-3313)

All authors declare no financial or other conflict of interest.