PRIMARY AMENORRHEA IN A 46,XY GIRL

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INTRODUCTION

Disorders of sex development (DSD) are medical condition in which the development of chromosomal, gonadal or anatomic sex varies from normal and may be incongruent with each other. Complete and partial androgen insensitivity syndrome (CAIS, PAIS) are associated with an increased risk of gonadal germ cell cancer (GGCC). Recent guidelines recommend gonadectomy in women with CAIS in late adolescence. Optimal protocol of management for phenotypic females with 46, XY gonadal dysgenesis involves prophylactic gonadectomy at diagnosis, due to the high risk of malignancy in these gonads.

CASE REPORT

The patient was referred at the age of 15 years old for primary amenorrhea. She was underweight, weighing 45 kg for a height 158 cm (BMI 18 kg/m2). Her pubertal status based on Tanner scale, was B1, P2, A1. Gynecologic examination revealed an absent vagina and clitoromegaly. Rectal Toucher examination revealed no internal genitalia structure was found. The laboratory data: FSH levels 19.75 IU/ml, Estradiol levels 23.29 pg/ml, testosterone level 271.0 ng/dl. Pelvic Ultrasonography uterus and vagina structure were not visualized and testes structure were identified at left labia mayora. Her karyotype was 46, XY. In our hospital the management of gender selection will be discussed by the team, including gynecological endocrinology, urogynecology, urology, plastic surgery department.

Management of DSD patients should be based on patient-center approach.

Therefore, overall management of DSD patients should follow according to patient's perception regarding to her gender orientation.

CONCLUSION

Chromosomal analysis should become the first line testing in primary amenorrhea. Management of DSD patients should be based on patient-center approach.

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