عنوان مقاله:

A female with 46,XY Disorder of Sexual Development with normal SRY gene sequence: A case report

محل انتشار:

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خلاصه مقاله:

Background: Disorders of sex development (DSD) are a medical condition that affects the normal process of sexual Various of the genes needed for gonad development have been identified by investigation of patients with disorders sex development (DSD).Phenotypes of patients with 46,XY DSD range from atonalism in female phenotype with complete external genitalia to male phenotype with testicular regression. Individuals with 46,XYagonadiam show a wide range of clinical features and in some cases, there is not a clear diagnosis for this patients. We presented the clinical and molecular study a patient with 46,XY female without gonadal tissue. Case presentation: A 27-year-old female was attended to our center because of primary amenorrhea. Ultrasonography did not show gonadal tissue including Mullerian structures, uterus, and Wolffian structures. Also, the patient had not streak gonad. We performed cytogenetic study and molecular analysis, including automated sequencing of the entire coding region of SRY gene, in the patient with agonadism. Our result showed 46,XY karyotype. Also, we noticed that molecular mutations in SRY are not identified as a cause of DSD female without a gonadal tissue. Laboratory examination showed that this case is a unique patient with 46,XYfemale agonadism that has no association with previously described. Conclusions: The present case was a patient with 46, XYagonadism without hormonal or kidney defect and we did not detect mutation in SRY gene. To our knowledge, this case is a unique patient with 46,XYagonadism that has no association with previously described. So this case would be helpful for clinicians to assess 46,XY female patients without gonadal .tissue

کلمات کلیدی:

Disorder of sexual development, XY female agonadism, SRY gene