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Introduction

17 β -hydroxysteroid dehydrogenase type 3 (17 β HSD3) enzyme deficiency is a rare cause of 46 XY disorder of sexual development. It is autosomal recessively inherited. Clinical phenotype is highly heterogeneous and depends on the mutation severity. Conversion of androstenedione into testosterone and estradiol into estrone deteriorates due to lack of the enzyme.

Objective

In this case report, we present a newborn who had complete female genitalia and was assigned as male after the early diagnosis of 17 β HSD3 deficiency.

The Karyotype:

- 46, XY

Genetic Study:

- Molecular analysis showed homozygous pathogenic variant p.R80Q (c.239G>A) in the *HSD17B3* gene.

Case

A 3 days old newborn was referred because of bilateral masses in the inguinal region. The patient was born by cesarean section at 38 weeks of gestation with 3100 g. The mother had type 2 diabetes mellitus. There was no disorder in the course of pregnancy. Parents were not consanguineous and her two siblings were dead by 3 and 4 months of age.

On physical examination;

- Weight: 3.3 kg (50 p)
- Height: 52 cm (75-90p)
- Head circumference : 34 cm (25-50p),
- Genital examination revealed normal vaginal opening and no cliteromegaly (Sinnecker stage 5) (Figure 1).
- The masses of 1 ml volume (gonad?) in the inguinal regions were palpated.

Laboratory Findings: (10 days old)

- FSH 2.2 IU/mL (N, <0.20-3.98)
- LH 11.3 IU/mL (N, <0.02-3.81)
- Total testosterone 94.2 ng/dl (N,75-400)
- **Androstenedion 143.8 ng/dl (N,0-73)**
- 17 OH progesterone 31 ng/dl (N,2-112)
- Dihydrotestosteron 11.46 ng/dl (N,5-60)
- **Baseline testosterone/androstenedione ratio 0.65 (N>0.8)**
- **Baseline testosterone/dihydrotestosteron ratio 8.2 (N< 8.5)**

Pelvic ultrasonography, showed the presence of testicles in the inguinal canal and no ovaries or uterus was observed.

Clinical follow up;

Multidisciplinary team considered male sex assignment for the patient. He was treated with parenteral testosterone at a dose of 25mg per month for 4 months in order to increase the size of phallus. In the sixth month of treatment, phallus size reached 3.5x1.5 cm (Figure 2). Patient underwent orchiopexy four times in 3, 5, 30 and 33 months of ages; had surgeries of vaginectomy, and correction of hypospadias-chordee at 26 months of age. Figure 3 and 4 show external genital images of the patient at 1.6 and 2.5 years of age.



Figure 1



Figure 2



Figure 3



Figure 4

Neonatal period (Figure 1), 6 months after (T) treatment (Figure 2), 1.6 years of age (Figure 3), 2.5 years of age (Figure 4)

Conclusion

17 β HSD3 deficiency cases which are raised as females are usually diagnosed late in the adolescent period due to virilization or lack of secondary sexual characteristics. In early diagnosed 17 β HSD3 deficiency cases, male sex assignment is crucial to prevent future gender confusion related problems.