# A RARE CAUSE OF SRY (-) 46, XX DSD: AROMATASE DEFICIENCY

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## Introduction

Aromatase deficiency is a rare cause of autosomal recessive 46, XX disorders of sex development (DSD) due to *CYP19A1* gene mutations. The affected patients cannot have a normal estrogen biosynthesis. It is characterized by low serum estrogen, increased gonadotropins, and ovarian cysts. Herein, we report a new case with aromatase deficiency.

**Table 1. Hormonal Analyzes**

| | 28 days | 3 months | 6 months |
|---|---|---|---|
| **FSH (mIU/ml)** | 72,3 | 83,2 | 77,5 |
| **LH (mIU/ml)** | 34,8 | 9,1 | 20,7 |
| **$E_2$ (pg/ml)** | <5 | <5 | <5 |
| **Total Testosteron (ng/dl)** | 107 | <20 | 109 |
| **17-OH Progesterone (ng/ml)** | 3,8 | | |
| **ACTH (pg/ml)** | 44,56 | | |
| **Cortisol (mcg/dl)** | 8,83 | | |

## Case Report

A 1-month-old girl was referred due to cliteromegaly. Her mother developed acne, voice change and hirsutism during pregnancy. She was born with a birthweight of 2990 gr at the 37th week of gestation. The parents were nonconsaguineous. The physical examination was unremarkable except a cliteromegaly of 1.3 cm and posteriorly fused labia minora. Initial evaluations excluded virilizing congenital adrenal hyperplasia. In hormonal analyzes FSH, LH and testosterone levels were high and E2 level was very low (Table-1). Pelvic ultrasonography revealed a normal uterus and multiple ovarian cysts. Karyotype was 46, XX and SRY was negative. Aromatase deficiency was considered due to the presence of maternal virilization, detection of hypergonadotropic hypogonadism during mini-puberty and low estradiol levels despite elevated total testosterone levels. A previously identified homozygote mutation in *CYP19A1* (c.628G>A, p.Glu210Lys) was found. During the follow-up, the fusion at the posterior of the labium minus was surgically corrected.

## Conclusion

Aromatase deficiency should be kept in mind in patients with SRY (-) 46,XX DSD cases particulary whenever there is a history of maternal virilization during pregnancy.