

# THREE SIBLINGS WITH GONADAL DYSGENESIS

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## ABSTRACT

Gonadal dysgenesis in female is defined as absent or insufficient development of ovaries. The patient with gonadal dysgenesis presents with primary amenorrhea and lack of development of secondary sexual characteristics due to inability of ovaries to produce sex steroids. The karyotype in patients with gonadal dysgenesis can be 46XX, 45XO, mosaicism or deletion of a certain part of X chromosome. Most cases are unexplained but thought to be autosomal recessive.

## CASE REPORTS

**Case 1:** A fourteen and half year-old-girl, presented with delayed puberty. She was born from consanguineous parents after a full-term pregnancy. Pelvic ultrasonography revealed streak ovaries and a hormone profile consistent with hypergonadotropic hypogonadism (Table). Thyroid, adrenal and growth hormone functions were normal. Karyotype was that of a normal female, 46 XX..

**Case 2:** We subsequently examined the proband's sister who was then 11 years old. Her laboratory and imaging results were similar to those of the proband (Table).

**Case 3:** 22 year old girl, sibling of the first patient was investigated because of primary amenorrhea (Table). She was receiving oral contraceptives therapy for primary amenorrhea of unknown cause.

Table: Clinic and laboratory findings of our patients

	Patient 1	Patient 2	Patient 3
Body weight (kg)	47(10-25 <sup>th</sup> p)	29 (10 <sup>th</sup> p)	52 (25 <sup>th</sup> )
Height (cm)	150 (3 <sup>th</sup> p)	133(3 <sup>th</sup> p)	168 (75 <sup>th</sup> )
Pubic hair (Tanner)	Stage 2	Stage 2	Stage 3
Breast (Tanner)	Stage 1	Stage 1	Stage 1
LH (mIU/ml)	17.11	14,4	16
FSH(mIU/ml)	79.9	43	112
Estradiol (ng/ml)	<10	<10	
Thyroid function	Normal	Normal	Normal
PRL	11.8	15.1	18
Standart synacten test	Normal	Normal	
Pelvic US	Hipoplastic Uterus , ovaries were not visible	Hipoplastic Uterus , ovaries were not visible	Hipoplastic Uterus , ovaries were not visible
Bone mineral density(z score)	-5.4	-0.3	-
Karyotype	46 XX	46 XX	46 XX

## CONCLUSION

Hypergonadotropic complete ovarian insufficiency with normal karyotype, also known as XX female gonadal dysgenesis is a rare , genetically heterogeneous disorder. Known causes of isolated XX-gonadal dysgenesis include recessive mutations in the FSH receptor gene, severe X-linked recessive mutations in the growth and differentiation factor BMP15, and both recessive and dominant mutations in the NR5A1/SF-1 transcriptional regulator.

Mutation analyse of the cases for SF-1 are pending.

A rare disease of gonadal dysgenesis in three siblings is presented.