A RARE CAUSE FOR 46,XX OVARIAN DYSGENESIS: PERRAULT SYNDROME

Gülay Karagüzel  Ayşenur Ökten

Department of Pediatric Endocrinology, Karadeniz Technical University, School of Medicine, Trabzon, Turkey
INTRODUCTION

• In 1951, Perrault reported the association of gonadal dysgenesis and deafness, now referred to as Perrault syndrome (PS)

• PS is a rare autosomal recessive condition affecting both females and males, only females have gonadal dysgenesis associated with sensorineural deafness which is present in both sexes
The most commonly reported additional manifestations are neurological, including mental retardation, cerebellar hypoplasia, and neuropathy.

PS is a genetically and clinically heterogeneous disorder.
INTRODUCTION

• Sensorineural hearing impairment and ovarian dysgenesis are the cardinal signs of PS in females
• Its pathogenetic basis is still unclear
• We present a case of PS in a girl
12-year-old girl

- She referred to our department with thyroid dysfunction
  - She was the first child of non-consanguineous parents
- Height: 139 cm (<3.p), Weight: 31 kg (3.p), Height SDS -2.93
- Mental retardation, hearing loss
- Normal external genitalia, prepubertal
- Hemogram, blood glucose, renal and liver function tests were normal
12-year-old girl

- Serum free-thyroxine: 1.29 ng/dL, TSH: 8.4 uIU/mL
  - subclinical hypothyroidism
- Thyroid ultrasound: Normal
- Metabolic screening: Normal

Estradiol: 5.7 pg/mL,
LH: 34.8 mIU/mL, FSH: 119.7 mIU/mL
- primary ovarian insufficiency
Pelvic ultrasound: Atrophic uterus and ovaries were not visualised
Pelvic MRI: ovaries were not visualised

Karyotype: 46, XX
"ovarian dysgenesis"

Audiometry: Sensorineural deafness
Brain MRI: Vermis hypoplasia
Audiometry: Sensorineural deafness

Perrault Syndrome
Perrault Syndrome

- Levothyroxine
- Hormone replacement therapy
- Vitamin D, Calcium

- Laparoscopy
  - September 22, 2014
CONCLUSIONS

• PS is a rare cause of ovarian dysgenesis, but should be considered in a girl with deafness

• It has been identified mutations in **CLPP, HARS2, and LARS2** genes, but no definitive gene mutation for PS and further studies are needed to establish of the underlying molecular defect
CONCLUSIONS

• **Type 1**: static, without neurological illness
• **Type 2**: with progressive neurological disease

• PS can be diagnosed by a careful clinical evaluation