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

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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

INTRODUCTION

- The most commonly reported additional manifestations are neurologycal, 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

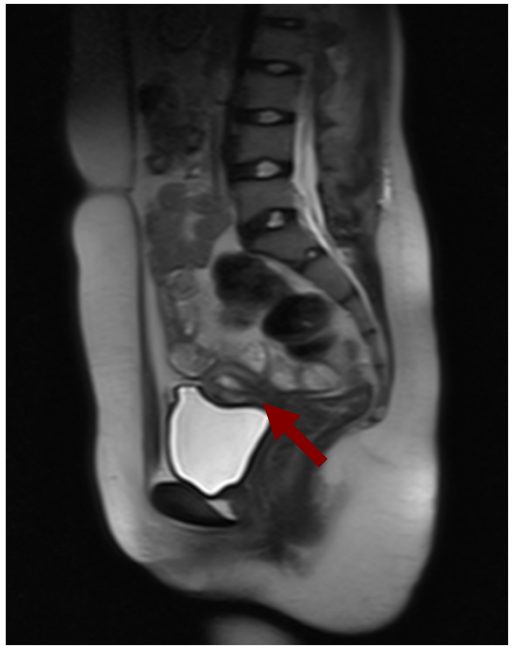
  subclinical hypothyroidism
- Thyroid ultrasound: Normal
- Metabolic screening: Normal

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

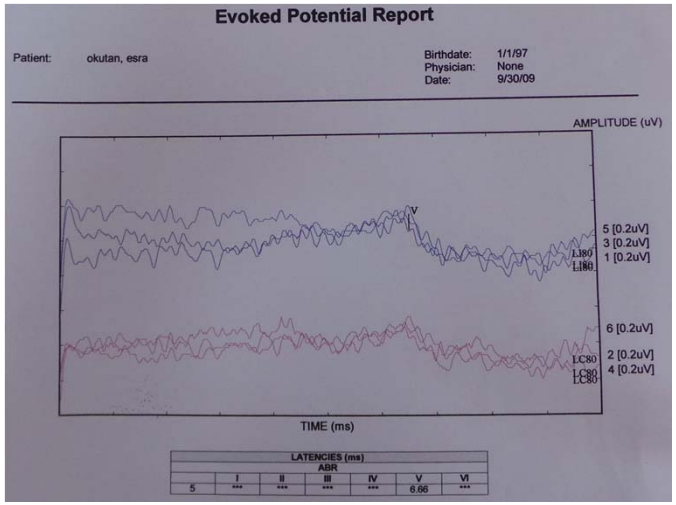
Pelvic MRI: ovaries were not visualised



Karyotype: 46, XX

"ovarian dysgenesis"

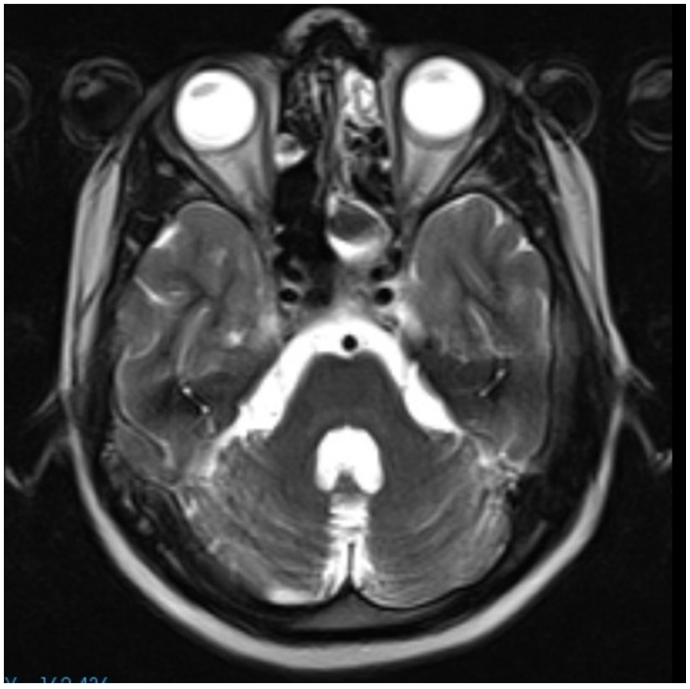
Audiometry: Sensorineural deafness

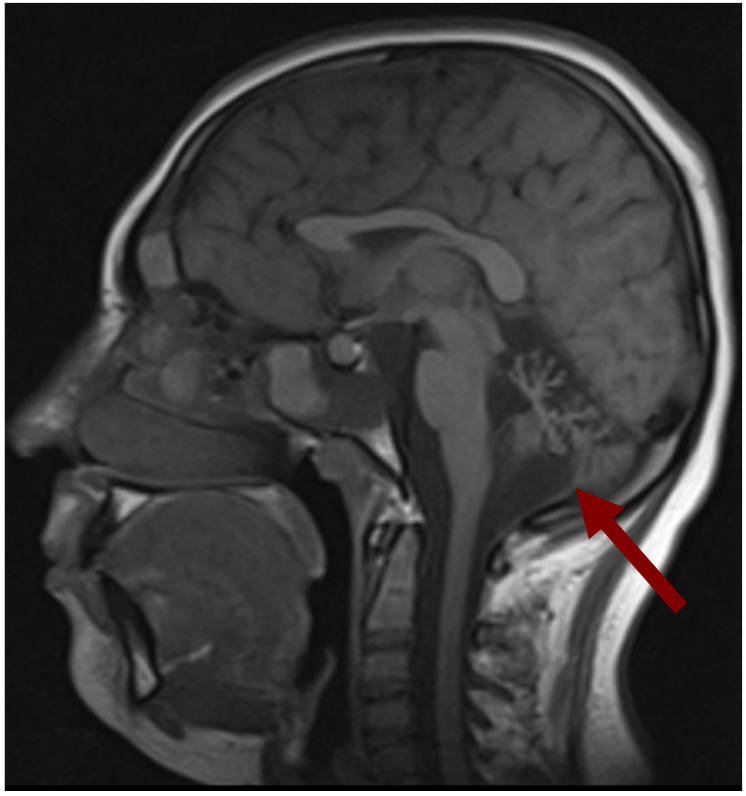


Brain MRI: Vermis hypoplasia

Audiometry: Sensorineural deafness

Perrault Syndrome





Perrault Syndrome

- Levothyroxine
- Hormone replasment 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