A Novel Mutation in Human Androgen Receptor Gene Causing Partial Androgen Insensitivity Syndrome in a Patient Presenting with Gynecomastia at Puberty

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Introduction

Partial androgen insensitivity syndrome (PAIS) typically presents with micropenis, perineoscrotal hypospadias, and bifid scrotum with descending or undescending testes and gynecomastia at puberty. It is an X-linked recessive disorder resulting from mutations in androgen receptor (*AR*) gene. In approximately 50% of cases a mutation in *AR* gene cannot be detected.

Here, we aim to present clinical characteristics of a novel mutation in the *AR* gene in an adolescent boy with PAIS who presented with gynecomastia at puberty.

Case Report

A 16-year-old boy,

Complaints;
- gynecomastia
- sparse facial hair

On physical examination;
Weight: 94 kg (2.84 SD)
Height: 170 cm (-0.56 SD)
Phenotypically male
Gynecomastia (Tanner's stage IV)
Normal axillary hair

External genitalia;
- Stretched penis lenght 8 cm and diameter 2.5 cm
- Penoscrotal hypospadias and bifid scrotum (Figure 1).
- Testes were palpable in the scrotum bilaterally as 2 mL
- Pubic hair Tanner stage IV

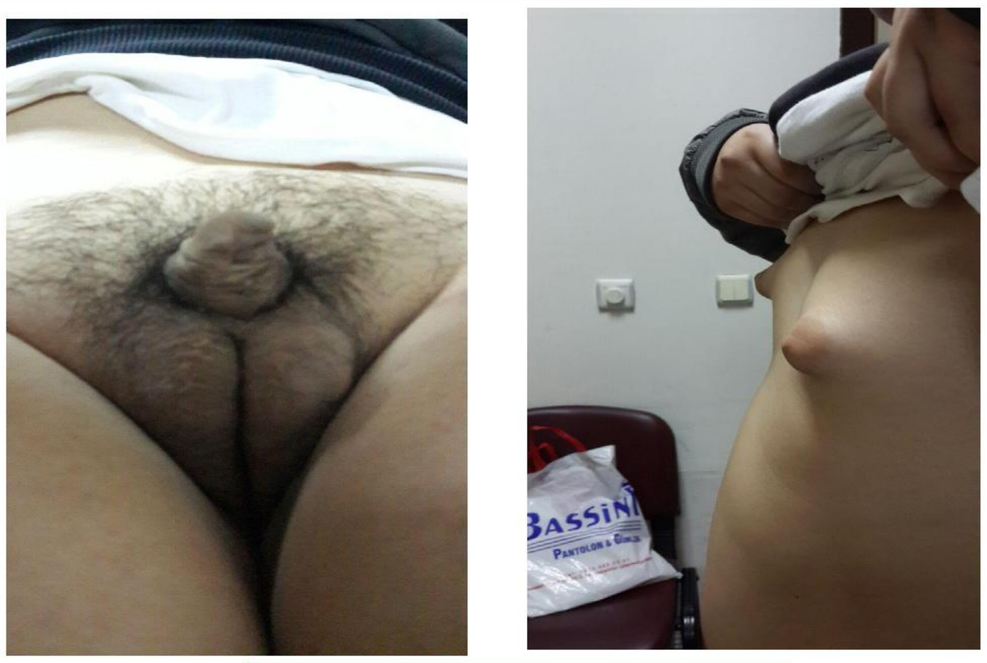

Figure 1. Patient pictures

Family history revealed male relatives from maternal side with similar clinical phenotype (Figure 2).

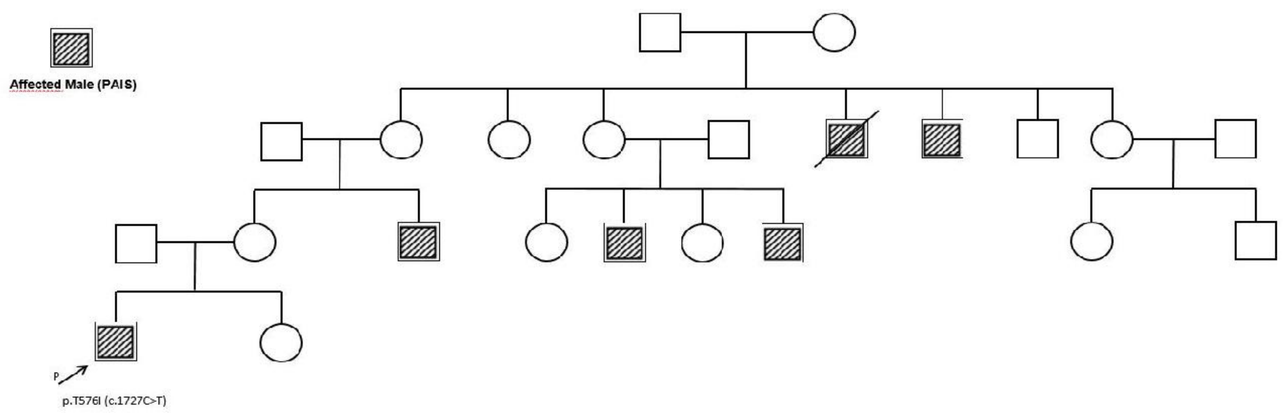

Figure 2. Pedigree

Laboratory results
FSH: 42.8 IU/L,
LH: 37.4 IU/L
T.Testosteron: 419 ng/dL
E2: 30.5 pg/dL

Karyotype: 46, XY

Radiological Evaluation:
Pelvic USG: No mullerian structures are present

Molecular analyses:
DNA sequence analysis revealed a novel mutation hemizygous p.T576I (c.1727C>T) in the *AR* gene.

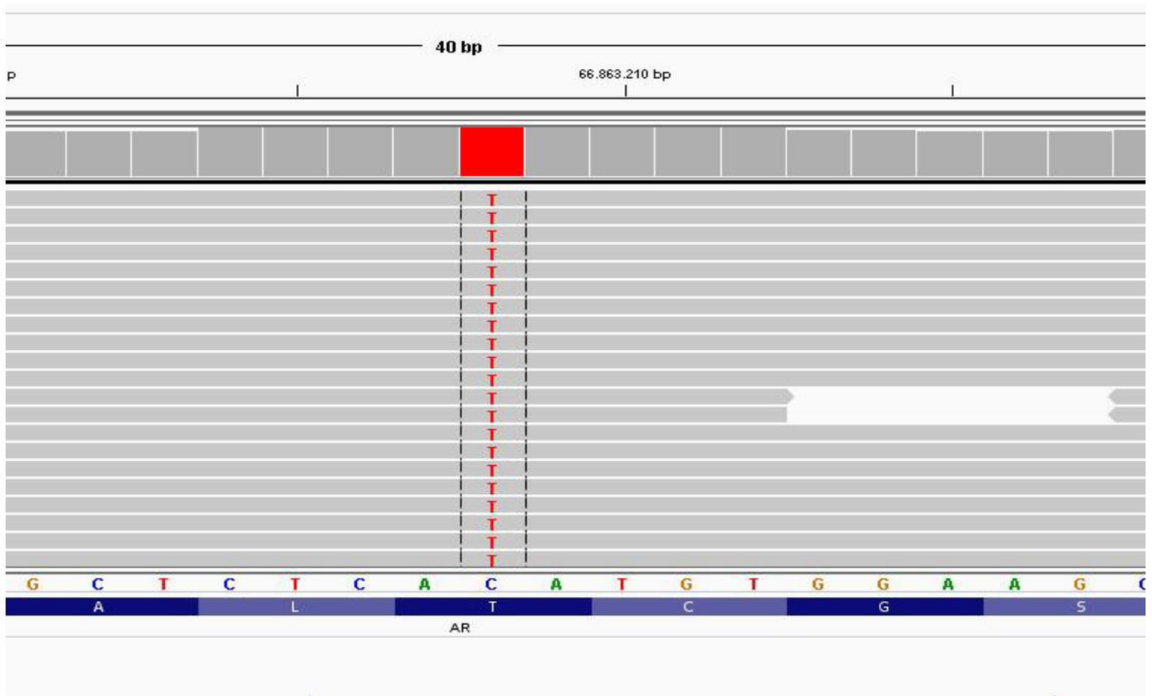

Figure 3. Androgen receptor gene map

Conclusion

The diagnosis of PAIS is based upon clinical phenotype and laboratory findings, and can be confirmed by detection of a defect in the AR gene. An accurate approach including a detailed family history suggesting an X-linked trait is an important clue to arrive at a quick diagnosis.

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