



Aim:

The 45,X/46,XY karyotype is rare (1/15.000)

- It represents from Turner females to phenotypically normal males with varying degrees of genital ambiguity.
- Although, high gonadotropin levels have been described in 0-5 years old girls with Turner syndrome, high FSH level is not well known finding in prepubertal girls older than 6 years.

Case:

- A 6 y 8 m girl presented with lipomastia
- She was born 3720 gr via section and her background was normal

Physical examination:

Height: 116.4 cm (-0.57 SD)

Weight: 26.8 kg (+1.17 SD)

BMI: 19.7 kg/m² (+1.73 SD) (95.6 p)

MPH: 170 cm (+1.05 SD)

BP: 110/70 mmHg,

Prominent eyes, prolonged palpebral fissur

Other systems were normal, Breast 1 (lipomastia), Pb 1

CA: 6^{8/12}

PAH: 154.9 – 148.8 cm

BA: 6^{10/12} - 7^{10/12}

MPH: 170 cm

- FSH: 16.7 mIU/mL
- LH: <0.1 mIU/mL
- E₂: <5 pg/mL
- TSH: 2.5 IU/mL
- fT4: 1.57 ng/dL
- Pelvic USG: Uterus 29x5x10 mm
RO 13x4x7 mm
LO 10x5x6 mm

- **Karyotype:** 45X0/46XY (22/82) FSH: 10 mIU/mL

- Buccal mucoza: 45X0/46XY (82/18) FSH: 8.2 mIU/mL

- ECHO and Urinary USG were normal

- No hearing loss

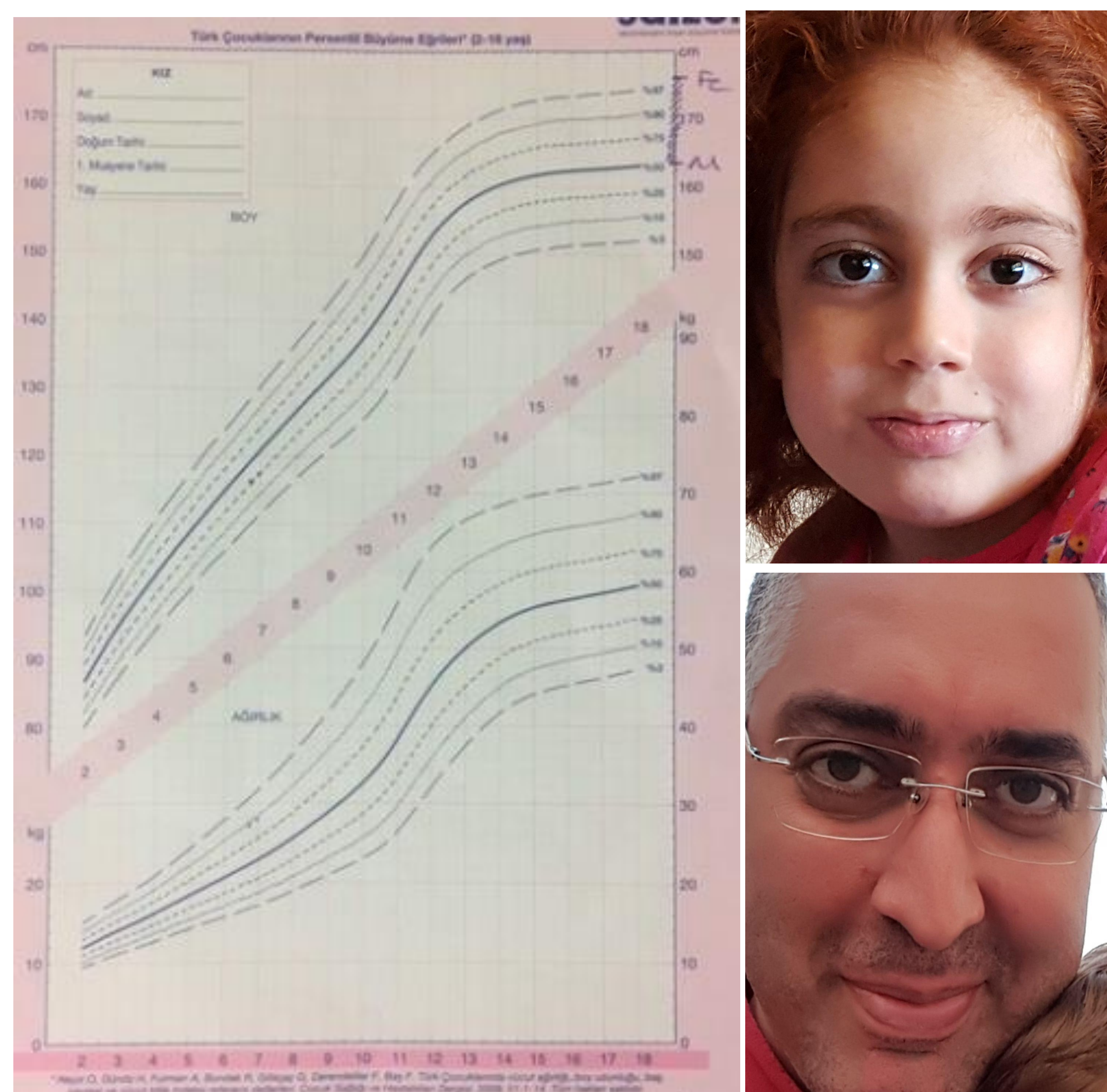
- Glucose: 82 mg/dl, insulin: 7.8 µIU/mL, HbA1c: %5

- AST:25 U/L, ALT: 14 U/L, tissueTransgl IgA negative,

- Normal Lipids

- Negative Anti-TPO

- IGF1: 144 ng/mL, DHEAS: 99 µg/dL (2.8-85)



Conclusion:

Despite of normal height, Turner syndrome should be suspected in the presence of

- Prepubertal FSH elevation or
- Lower height SDS than MPH SDS

Elevated FSH level expects in 0-5 years old children with 45X0 Turner syndrome, but it can be also seen in prepubertal girls.