

Mozaic Turner Syndrome and Precocious Puberty Association: A Three-Year-Old Patient

Özlem Sangün¹, Pınar Kiper Mısırlıoğlu²,
Tülün Savaş³

¹Başkent University Faculty of Medicine, Department of
Pediatric Endocrinology, Ankara, Turkey

²Başkent University Faculty of Medicine, Department of
Pediatrics, Ankara, Turkey

³Başkent University Faculty of Medicine, Department of
Pediatric Neurology, Ankara, Turkey

Turner syndrome is chromosome abnormality seen in 1/2500 female live births and it is mostly seen as a cause of hypergonadotropic hypogonadism and puberty delay.

A 26-month-old female patient had vomiting attacks since she was 2.5 months old, mild dysmorphism, and a dead sibling history. Her karyotype analysis indicated mosaic Turner syndrome [cytogenetic analysis' result: Mos 45,X(13)/46,Xr(X)(27),lshr(X)(Wepx+)] and thus she was referred to our clinic.

In her physical examination, her nose bridge and

forehead were shallow; she had a mild hypertelorism; nipples were wild apart, and she had a mild pectus excavatus at thorax. Her bone age was consistent with 1.5-2 years while she was 18 months. On her pelvic ultrasonography, streak gonad was reported and it was confirmed with an magnetic resonance imaging. On her first evaluation, follicle-stimulating hormone (FSH) was 50.22 mIU/mL, luteinizing hormone (LH) was 2.45 mIU/mL, estradiol (E2) was <10.00, and 4 months later, the results were similar (FSH: 54.09 mIU/mL, LH: 2.58 mIU/mL, E2: 147.85 pg/mL). It was double-checked and confirmed. Her pelvic ultrasound imaging was performed again and it showed that bilateral ovary volumes have increased.

Turner syndrome is generally a cause of ovarian deficiency, but in rare mosaic cases, high gonadotropin levels stimulate the healthy germ cells and cause precocious puberty. Noticing these patients earlier and treating them would make the patients' life standards better, especially regarding their low height potentials.

Key words: Turner syndrome, precocious puberty