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A Case of Turner Syndrome Presenting as Short Stature

Sugha Varuna[1], Bharti Sapna[2]

[1]In charge, Pediatrics Unit, Civil Hospital, Bhawarna, Distt Kangra (HP)
[2]In charge, OBG Unit, Civil Hospital, Bhawarna, Distt Kangra (HP)

Corresponding Author: Bharti Sapna

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ABSTRACT

Turner syndrome has an incidence of 1 in 2500 female births. Clinical signs such as short stature, lymphoedema and delayed puberty, are common reasons for screening for Turner syndrome. We present a 10-year-old female with short stature. The diagnosis was confirmed by the patient's karyotype (45, XO). Physicians must be able to suspect Turner syndrome in order to make an early diagnosis, refer, and manage affected children for optimal growth and development.

Keywords: Turner syndrome, short stature, Growth hormone

CASE DESCRIPTION

A 10 years old girl presented in OPD for evaluation of short stature. The child was not gaining proper height as per her age since early infancy. Her birth and perinatal history were uneventful. She started to sit without support at 14 months and started to walk without support at 22 months. Other developmental domains were normal. Findings on her physical examination included: Short stature with a height of 112 cm (less than 3rd percentile on IAP growth chart). Her mid parental height was 154 cm which lies on 25th percentile (Figure 1).

On physical examination the child had high arched palate, Frontal bossing, flat nasal bridge, low hairline and low set ears, webbed neck, cubitus valgus and broad chest with widely spaced nipples(Figure 2). Her blood pressure in right upper limb was 112/78 mmHg, left upper limb was 110/80 mmHg, right lower limb 112/80 mmHg, left lower limb 114/78 mmHg with pulse of 86/min. On CVS examination, S1 and S2 were normal. The SMR was prepubertal.

Workup for short stature was done. The blood counts, electrolytes, CRP, ESR was normal. Thyroid function test, blood gas analysis and celiac screen was normal. Echo was done which was normal. Ultrasound abdomen and pelvis revealed bilateral streak gonads. Turner syndrome was confirmed by band karyotyping.

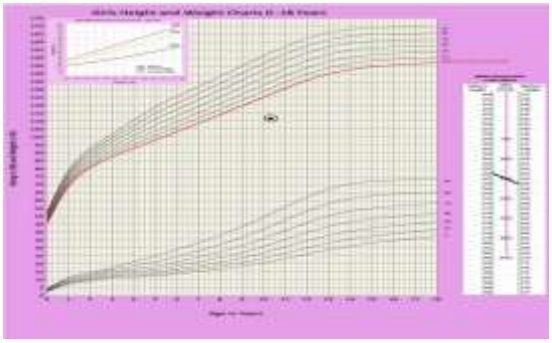

Figure 1 The chart (IAP) shows height below 3rd centile and mid parental height (MPH) at 25th centile

Figure 2 The child has short stature, low hairline, frontal bossing, flat nasal bridge widely placed nipples and cubitus valgus (Picture taken with consent)

DISCUSSION

Turner syndrome (TS) results from the partial or complete loss of X-chromosome in phenotypic females. It has a prevalence of 1 in 2000 to 2500 live births. 45 X is the karyotype typically seen in patients with Turners syndrome, other sex chromosome anomalies such as isochromosome Xq, ring X, deletion Xp, or an abnormal Y chromosome can also cause this condition.(1)

The most common causes for short stature are constitutional and genetic. In the index case because of the presence of features like high arched palate, low hair line, widely spaced nipple, and cubitus valgus along with short stature, Turner syndrome was suspected. These features had been missed in her previous hospital visits.

According to a study the diagnosis of Turner syndrome is made on an average of seven years when patient presents with short stature.(2) In a case series, 4 percent of girls referred for genetic evaluation of isolated short stature were diagnosed with Turner's syndrome (3).Cardiac anomalies are detected in 20% most common being coarctation of aorta. 40% patients have hypertension.(4) 75% patients have recurrent bilateral otitis media. High arched palate results in recurrent infection as fluid drainage from middle ear is impaired. Sensorineural hearing loss is seen. (5) Developmental delay is present but intelligence is normal in most cases . Our patient also had delay in gross motor milestones.

Recombinant growth hormone therapy has been shown to improve adult height in patients with TS by 5–8 cm (6) but the efficacy depends on multiple factors including mid-parental height, age at initiation of GH therapy, duration and dose of GH therapy and baseline height prior to initiation of GH therapy.(7,8) GH therapy is recommended at 4–6 years of age or sooner in the presence of growth failure.(9) The latest guidelines recommend starting at 45–50 µg/kg/day and possibly increasing up to a dose of 68 µg/kg/day administered subcutaneously seven days a week, preferably at night.(9) Height should be monitored every 3–4 months in the first year of therapy and every 4–6 months thereafter and GH can be discontinued after linear growth is complete (bone age of approximately 13.5 to 14 years; height velocity <2 cm/year). (9)

In the index child due hypergonadotropic hypogonadism from gonadal dysgenesis, she would then need hormone replacement therapy, initially for induction of puberty, and later on in life, for maintaining secondary sexual characteristics and possibly, pregnancy.(10)

CONCLUSION

Most of the turner syndrome cases are diagnosed in puberty due to stunted growth and failure to attain menarche. It is therefore important to clinically identify these cases early period to start hormonal therapy for development, growth, and pubertal induction. Screening for congenital heart disease, horseshoe kidney, and hypothyroidism which are commonly associated with turner syndrome is important. There should be focus on patients' mental, reproductive, and physical growth to minimize complications associated with Turner syndrome.

Declaration by Authors

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Conflict of Interest: The authors declare no conflict of interest.

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