

SKELETAL MANIFESTATIONS IN AN ADULT WITH TURNER'S SYNDROME

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ABSTRACT

Turner's syndrome (TS) is a well-defined sex chromosomal disorder associated with short stature and various skeletal anomalies. Several factors have been reported to be responsible for the development of these skeletal anomalies. These factors are mainly due to premature fusion of the epiphysis as seen in the short 4th metacarpals, premature fusion of the medial half of the distal radial growth plates resulting in Madelung's deformity and oestrogen deficiency resulting in osteoporosis and delayed skeletal maturation. The presence of multiple skeletal features in one patient as seen in this present case is rarely reported. This case is hereby reported to highlight the classical skeletal manifestations of Turner's syndrome.

Key words: Turner, Syndrome, Skeletal, Adult

Introduction

Turner's syndrome is the most common chromosomal abnormality in females.¹ It affects 1 in 2 of 745 live female births in Nigeria.² It is a result of absence of an X chromosome or the presence of a structurally abnormal X chromosome.^{1,3}

It is characterized by a female phenotype with short stature, webbed neck, ovarian dysgenesis, cubitus valgus, and congenital heart disease (CHD).³ Skeletal abnormalities are known features of Turner's syndrome.⁴ The incidence of skeletal abnormalities is nearly 100% for short stature and 35-60% for other skeletal anomalies such as short metacarpals, cubitus valgus, high-arched palate, micrognathia, and short neck.^{1,5} In addition, Le'ri-Weill dyschondrosteosis (LWD) characterized by Madelung's deformity and mesomelia occasionally occurs in Turner's syndrome.⁵ Bilateral prominence of the medial femoral condyles with depressed medial tibial plateau resulting in genu valgum may also be present.¹

Case Presentation

B.O. is a 33-year-old woman who presented with history of inability to menstruate. She had never menstruated before. Her mother also noticed absent secondary sexual characteristics. There was no history of head injury in the past or childhood illness. There was no associated history of headache or reduced vision. In addition, there was no history of anterior neck swelling or history suggestive of cold intolerance or hoarseness of voice.

She is not a known diabetic, hypertensive or sickle cell disease patient. She had not been admitted in the hospital before and had not undergone any surgery. Her pregnancy and delivery were uneventful. However, she had delayed developmental milestones;

- Neck control at 3 months
- Crawling at 9 months
- Standing with support at 3 years
- Walking at 4 years

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Physical examination revealed a stunted young lady, not pale, anicteric, acyanosed with no palpably enlarged lymph nodes, sparse axillary hair, no finger clubbing nor pedal edema.

The anthropometric parameters showed; height = 1.44m, weight = 35.75kg and body mass index (BMI) = 13.24kg/m². (Height for age indicates stunted growth while the BMI is underweight).

Head and neck examination showed normal posterior hair line and normal positioned ears. She has high archedpalate but no webbed neck.

Chest was bilaterally symmetrical and moved with respiration. No breast development was seen bilaterally. The nipples were widely spaced. The respiratory rate was 18 cycles per minute.

Cardiovascular system showed regular, synchronous, full volume pulse. There was no radio-femoral delay, no thickened arterial wall nor collapsing pulse. No locomotor brachialis was also seen. Pulse rate was 80 bpm while the BP was 100/60mmHg. The Jugular Venous Pressure (JVP) was not raised. Precordium was not hyperactive and no thrill was noted. Heart sounds S1 and S2 were heard. No murmur was picked.

External genitalia showed no pubic hair, underdeveloped labia majora, nil labia minora, slightly enlarged clitoris and infantile vagina orifice.

Musculoskeletal system showed wrist deformities with bilateral prominent styloid processes, cubitus valgus, bilateral ulna deviation, mild genu valgum deformity and overlapping 4th toe in the right foot Central nervous system revealed loss of olfactory nerve sensation. The gait, tone, reflex and power were normal. Sensation to pain, touch and pressure were also normal.

Full blood count, fasting blood sugar and urinalysis were normal. Echocardiography showed sinus tachycardia.

Abdominopelvic ultrasound showed normal abdominal visceral, hypoplastic uterus (4 mm in widest AP diameter). The ovaries were not visualized.

No bone abnormality was found on chest radiograph. Other skeletal series revealed shortening of the left 4th metacarpal bone (metacarpal sign), coarse reticular pattern of the carpal bones, crowding of the carpal bones as well as narrowing of scapho-lunate angle (Fig. 1). Others include: bilateral deformity of the medial aspect of the distal radial bone, subluxation



Figure 1: Plain radiograph of the left hand (anteroposterior view) showing short 4th metacarpal. Coarse reticular pattern of the carpal bones and defective development of the inner third of the radius and crowding of the carpal bones as well as narrowing of scapho-lunate angle are also demonstrated. These features are consistent with Madelung's deformity.

of the distal ulna, forward projection of the carpal bones and hand (Madelung's deformity) (Fig. 2), mild bilateral genu-valgum deformity of the knee, and



Figure 2: Plain radiograph of both hands (lateral view) showing lateral and dorsal bowing of the radius as well as subluxation of the distal ulna giving a bayonet-like appearance suggestive of Madelung's deformity.

depressed medial tibial plateau (Fig. 3). Diagnosis of Turner's syndrome was made based on these findings.

Hypoplastic uterus, fallopian tube and ovaries were found during diagnostic laparoscopy. Cytogenetic study (karyotype) showed 45 XO confirming Turner's

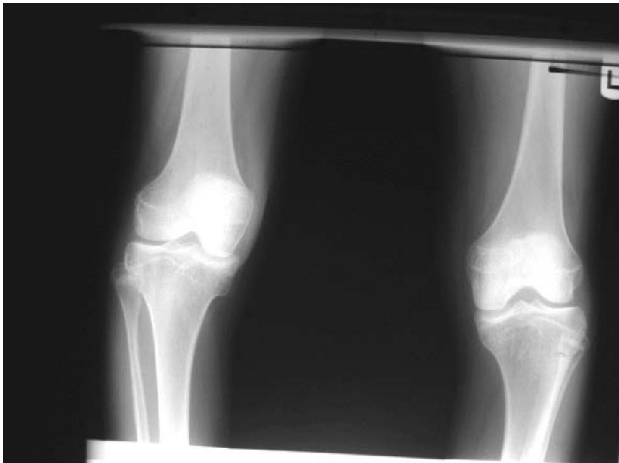


Figure 3: Plain radiograph of both knees (AP view) showing prominence of the medial femoral condyles with depressed medial tibial plateau bilaterally and mild genu valgum.

syndrome.

The family physicians had several counselling sessions with the patient and her family members discussing the patient's medical and psychosocial problems. The patient was later lost to follow up.

Discussion

Turner's syndrome (TS) is a well-defined sex chromosomal disorder associated with short stature and various skeletal anomalies, together with non-skeletal stigmata.¹

However, the syndrome was named after Henry Turner, an American endocrinologist, who in 1938 described seven women with characteristic phenotypic features of the syndrome.⁶ He emphasized the presence of gonadal dysgenesis and was the first to initiate estrogen replacement therapy.

TS is associated with increased overall mortality and reduced life expectancy.¹ Even after exclusion of deaths from congenital heart disease, the mortality rates remain excessive, particularly in women with 45X monosomy.¹

The incidence of skeletal abnormalities is nearly 100% for short stature and 35-60% for other skeletal anomalies such as short metacarpals, cubitus valgus, high-arched palate, micrognathia, and short neck.^{1,5} In addition, Le´ri-Weill dyschondrosteosis (LWD) characterized by Madelung's deformity and mesomelia occasionally occurs in TS.⁵

About half of females with TS have cubitus valgus, or a wide carrying angle, as a result of a developmental defect of the head of the ulna.¹ Short metacarpals and metatarsals are found in a proportion of women with TS.¹ In addition, short 4th metacarpal is radiologically evaluated as positive, borderline, or negative when a tip of the 4th metacarpal is below, on, or above a straight line drawn between tips of the 3rd and 5th metacarpals respectively.⁵ It is positive in this case.

"Madelung's deformity" (also known as bayonet deformity) of the wrists may be present as a result of lateral and dorsal bowing of the radius and subluxation of the distal ulna.¹ It is associated with TS in 10% of cases.⁷ This feature was classically demonstrated in this case.

Osteoporosis, crowding and coarse reticular pattern of the carpal bones may be present as seen in this case.^{1,8} Bercu et al⁸ reported that nine out of ten patients with Turner's syndrome have a coarse reticular pattern of the carpal bones and therefore concluded that this new sign is more reliable and specific than "metacarpal sign" which may also be found in patients with short stature due to other etiologies.

In the knee, prominence of the medial femoral condyles with depressed medial tibial plateau resulting in a genu valgum may also be present.¹ These findings were also seen in this present case.

Adults with TS continue to show evidence of a reduced bone mass,¹ and this has also been shown to be associated with an increased risk of fractures.⁹ Davies et al reported a fracture frequency of 45% in women with TS. No fracture was found in this patient. This reduction in bone mass is partly as a result of delayed skeletal maturation¹ or oestrogen deficiency⁹ but the prevalence of osteoporosis and bone fractures does not show significant increase in women with Turner's syndrome who are treated with standard oestrogen therapy.¹⁰

The characteristic facies of a female with TS is also primarily due to skeletal malformations.¹ These result in micrognathia, a downward droop of the outer corner of the eyes and epicanthic folds, a high arched palate, and low-set ears.¹ Only arched palate was present in this case.

Scoliosis may be present in approximately 10% of females, and it may or may not be associated with

vertebral abnormalities.¹ Scoliosis was not demonstrated in this present case. Sternal abnormalities on a lateral chest roentgenogram are common skeletal abnormalities associated with Turner's syndrome and are independent of associated congenital heart disease (CHD).⁴ No sternal abnormality or congenital heart disease was present in this case.

Several factors have been reported to be responsible for the development of these skeletal anomalies.⁵ These factors are mainly due to premature fusion of the epiphysis as seen in the short 4th metacarpals, premature fusion of the medial half of the distal radial growth plates resulting in Madelung's deformity and oestrogen deficiency resulting in osteoporosis.⁵ Osteoporosis may result in arthritis and pathological fracture.¹¹ The arthritis may also result from congenital developmental dysplasia of the hip joint - a disorder seen commonly in Turner's syndrome and contributing to the development of arthritis in such patients.¹¹ However, arthritis and pathologic fracture are not demonstrated in this patient.

The diagnosis of TS may be delayed until adulthood in up to 10% of women. This is especially likely in females who enter puberty spontaneously and subsequently present with amenorrhea (primary or secondary) or infertility.¹ The present patient presented on account of primary amenorrhea. The definitive diagnosis is made on the basis of a chromosomal analysis.¹ A peripheral lymphocyte karyotype is routinely analysed and is diagnostic in the majority of cases. In rare instances, this karyotype is normal in females with TS mosaicism.¹

In conclusion, this case of a 33-year-old woman with TS is presented because of its myriads of classical radiographic features.

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