Quiz

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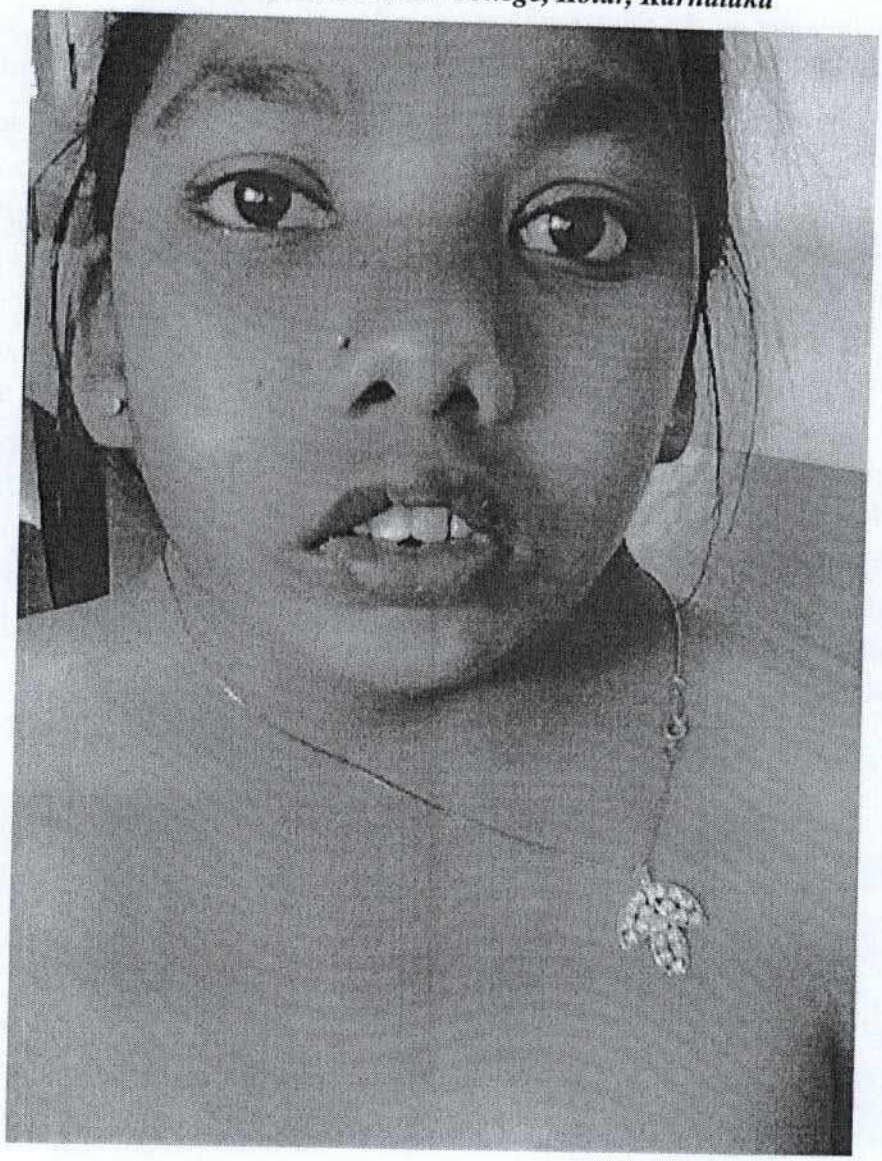

Patient aged 15 years, presented with primary amenorrhoea and cubitus valgus.

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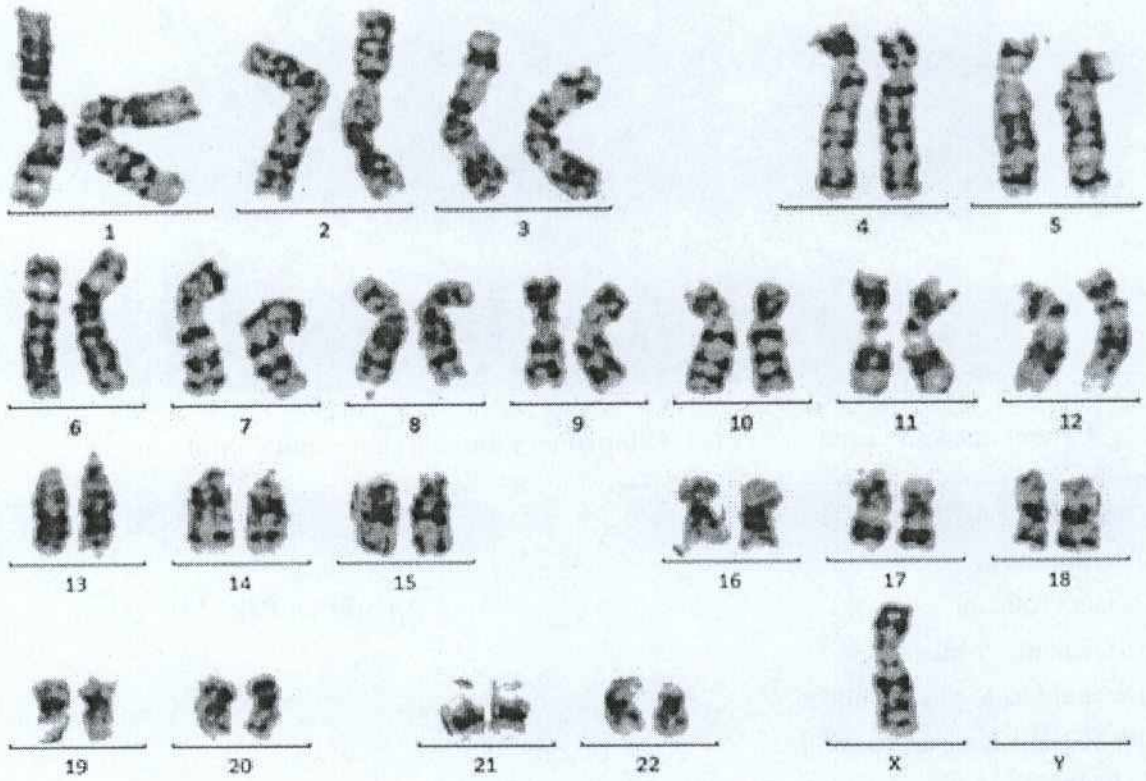
TURNER SYNDROME

Turner syndrome is one of the most common monosomies in live born humans. This is associated with monosomy or partial monosomy of the X chromosome, including mosaicism. Embryos with a 45 X, karyotype are prone to spontaneous abortion. More than 99% of 45 X conceptuses abort spontaneously, accounting for one-fifth of all spontaneous abortions. The phenotype of Turner syndrome is female. Many patients with Turner syndrome have some or none of the somatic abnormalities, while 50% of cases have characteristic features. Non-pitting oedema of the hands and feet is present only in newborns. Most cases present as short stature and absence of secondary sexual characters at adolescence. As short stature may be the only manifestation, chromosomal analysis of prepubertal girls with short stature is indicated.

Clinical features of Turner syndrome are, short stature, short webbed neck, cubitus valgus, primary amenorrhoea, Lack of secondary sex characters, Presence of epicanthic folds, hypertelorism ,high-arched palate, low posterior hair line , shield chest with widely spaced nipples, coarctation of the aorta, horse-shoe shaped kidney and other renal anomalies, multiple naevi, lymphoedema of the hands and feet in newborns, nail hypoplasia and short 4th metacarpal and metatarsal bones. IQ may be lower than that in normal siblings. Specific deficits in perceptual and spatial thinking, mathematics and language skills may occur. There is an increased risk for autoimmune



thyroiditis and essential hypertension by teenage in these patients.

Management includes Growth hormone therapy should be started in early childhood, as soon as the child with Turner syndrome starts falling below the 5th percentile for normal, and must be continued till growth is complete. Oestrogen can give rise to adequate development of the breasts and pubic hair. Initially conjugated oestrogen or ethinyl oestradiol is started in a low dosage when a bone age of 15 years is achieved or earlier if the patient is keen on the development of secondary sexual characters. The dose is increased every six months. After 1-2 years or whenever breakthrough bleeding occurs, cyclical progesterone is added to simulate normal cycles. Regular menstruation can be achieved with this treatment. Assisted reproduction using donor ova is possible. Other associated problems such as visual defects, strabismus, congenital heart, renal defects, hypertension and hypothyroidism should be investigated and treated as required. Screening for diabetes mellitus should be carried out during adulthood, especially in obese patients.

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