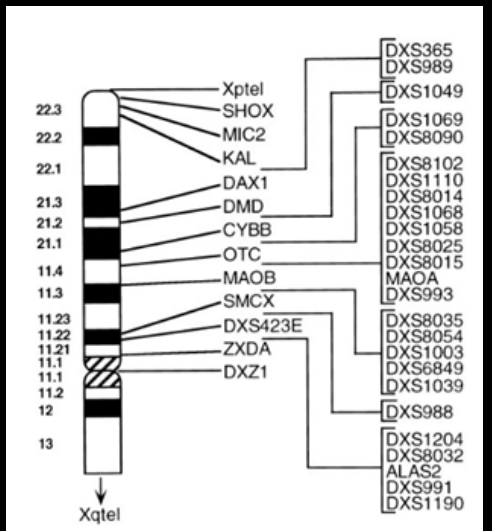
DISCUSSION

Turner syndrome (TS) is a chromosomal condition affecting 1 in 2000 females characterized by partial or complete deletion of one of the X chromosomes [1]. About 50% of affected cases are Monosomy for X chromosome (45,X) and tend to present with short stature/skeletal changes, a webbed neck, cardiovascular and renal abnormalities, gonadal dysgenesis and/or ovarian failure. For mosaic TS, the variability of the phenotype depends on the degree of mosaicism. Patients may experience normal pubertal development and can conceive spontaneously [2,3].

Patients with deletion of the short arm of the X chromosome (Xp deletions) present with short stature, with or without other somatic traits common to TS. Due to preservation of fertility, mother-to-daughter transmission of terminal Xp deletion TS is possible and is considered to be familial TS [4,5]. Our patient presents with Xp deletion along with extra copy of same deleted X chromosome (Three copies of Xq).



SHOX gene located on the short arm of X chromosome.

There are multiple chromosome abnormalities associated with TS, including mosaicism as 45,X/46,X + ring, 45,X/46,XX/47,XXX, 46X,Xq (interstitial long-arm deletions) and 46,X,Xp(short arm deletion). The prevalence of Xp deletion among TS patients has been reported to be ~2% [4]. Patients with partial Xp deletion have variable phenotypes including short stature and generally preserved ovarian function along with some other somatic features of TS. The short stature and skeletal features can be explained by a loss of function (haploinsufficiency) of the homeobox gene, SHOX gene in the pseudoautosomal region of Xp.

46,X,psu idic(X)(p22.3)] is a very rare sex chromosome abnormality by the presence of one normal X and one derivative X with the of deletion of the short arm of X chromosome along with extra copy of remaining X chromosome. In this case TS features are present because of deletion of the short arm of X chromosome, which is carries many important gene like SHOX.

Our case presents a girl with a de novo Xp deletion with PSU isodicentric (Xp) having short stature and delayed puberty.