# A Rare Variant of Turner Syndrome: Clinical Report from Kuwait

**Dr Kholoud Salem (1), Dr Dalia Al-Abdulrazzaq (2)**

(1) Department of Pediatrics, Mubarak Al-Kabeer Hospital, Ministry of Health, Kuwait

(2) Department of Pediatrics, Faculty of Medicine, Kuwait University, Kuwait

## Introduction:

- Turner Syndrome (TS) is characterized cytogenetically by X chromosome monosomy, the presence of an abnormal X chromosome, or mosaicism of a 45,X or have an abnormal sex chromosome rearrangement (1) .
- Girls with variant TS show no features, fewer or milder features of TS.
- We present the first clinical report of a girl with a rare variant of TS (46, X, i(X) (q10)) from Kuwait.

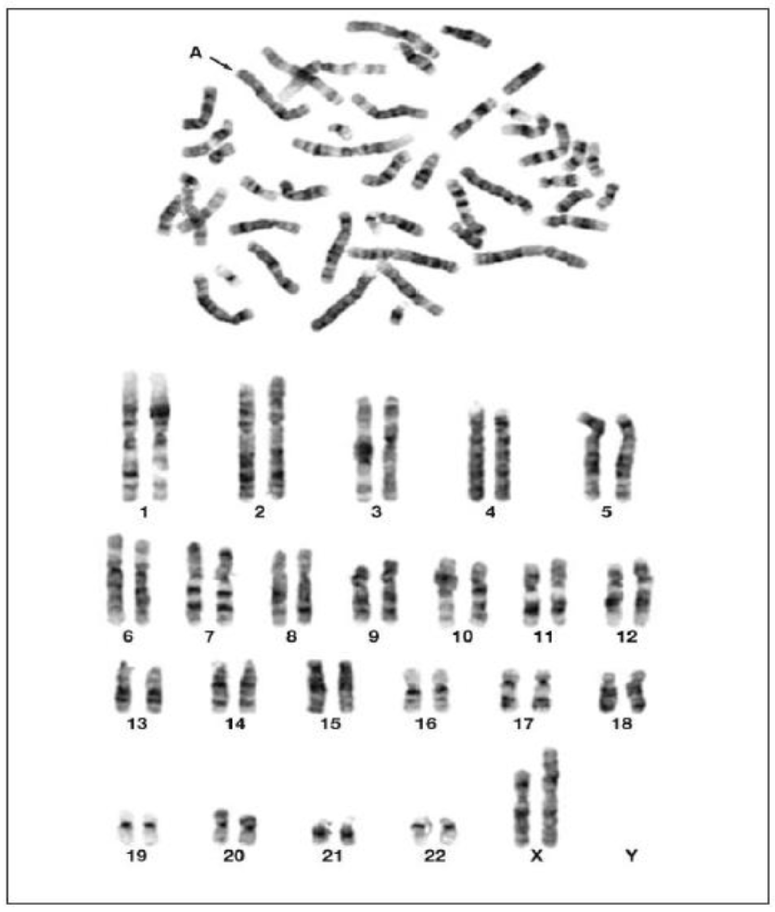


**Figure (1) Chromosomic photograph of similar case from the literature (1). Isochromosome X (Arrow)**

## Material and methods:

- A 12 year old Kuwaiti girl was referred to Endocrine clinic for short stature (already started on growth hormone therapy).
- She is known to have primary hypothyroidism.
- Full assessment and completion of the workup for short stature and hypothyroidism was started at the Endocrine clinic.

## Results:

- Height was at -4 SD and prepubertal.
- No other physical features of TS.
- Chromosomal analysis revealed 46, X, i(X) (q10) (Figure 1).
- Investigational workup showed in Table (1).
- ENT examination showed secretory otitis media (OM).
- She is currently under treatment with growth hormone and thyroid replacement.

## Conclusion:

- Our case demonstrated similar reported features namely, short stature, hypothyroidism, and gonadal dysfunction. However, different from other cases did not have cardiac or renal anomalies (2).
- This case emphasizes the basic principle in managing girls with short stature by performing genetic studies even without overt clinical features of TS.

| Investigation | Result |
|---|---|
| TSH, fT4 on TT | Normal |
| Anti-TPO AB | Negative |
| US abdomen and Pelvis | small uterus for her age and non-visualized ovaries with no renal anomalies |
| Echocardiography | Normal |

**Table (1)**

(1) Akbas E et al "Rare Types of Turner Syndrome: Clinical Presentation and Cytogenetics in Five Cases". (2012) LabMedicine, 43, 197-204.
(2) Sybert V, McCauley E. "Turner's Syndrome". (2004) N Engl J Med;351:1227-38.

ESPE 2015 1245--P3
Misc 1
mohamed kholoud
DOI: 10.3252/pso.eu.54espe.2015
Poster presented at: ESPE 2015 BARCELONA