A Rare Variant of Turner Syndrome: Clinical Report from Kuwait

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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.

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.

<table>
<thead>
<tr>
<th>Investigation</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>TSH, FT4 on TT</td>
<td>Normal</td>
</tr>
<tr>
<td>Anti-TPO AB</td>
<td>Negative</td>
</tr>
<tr>
<td>US abdomen and Pelvis</td>
<td>small uterus for her age and non-visualized ovaries with no renal anomalies</td>
</tr>
<tr>
<td>Echocardiography</td>
<td>Normal</td>
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</tbody>
</table>

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

Table (1)

(1) Abbas E et al “Rare Types of Turner Syndrome: Clinical Presentation and Cytogenetics in Five Cases”. (2012) LabMedicine, 43, 197-201.