Resistance to thyroid hormone (RTH) is a rare genetic disease due to thyroid hormone receptor defect and is usually inherited in an autosomal dominant fashion. We describe a case of RTH in a Southasian family of Bangladeshi extraction.

Presentation
A 32 year old female, normally fit and well, initially referred to ENT surgeon due to goitre. She was clinically euthyroid and not on any medications.

Investigations
Ultrasound thyroid: Multinodular goitre with benign features; THY2 on FNA
Thyroid function test:
- TSH: 1.35 miu/l (normal range 0.4-4 miu/l)
- FT4: 29.2 pmol/l (12-22 pmol/l)
- FT3: T3 7.1 pmol/l (2.80 – 7 pmol/l)

Differential Diagnoses
1. TSHoma
2. RTH (resistance to thyroid hormone)
3. Assay interference

Further Investigations
- Alpha subunit: <0.30 IU/L (0-1)
- SHBG: 27 nmol/l (20 – 130)
- Anterior pituitary function: normal
- Assay interference ruled out.

Family history
Her father and brother have similar thyroid function test results (normal TSH, raised FT4 and FT3).

Diagnosis
RTH confirmed by genetic analysis: heterozygous for thyroid hormone receptor beta mutation due to single base change at c.1378G>A in exon 10 of TRbeta gene resulting in abnormal TRbeta protein (p.Glu460Lys).

Management
1. Conservative management of goitre
2. Euthyroid clinically – no treatment
3. Further family screening and genetic testing underway

<table>
<thead>
<tr>
<th>Parameter</th>
<th>RTH</th>
<th>TSHoma</th>
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</thead>
<tbody>
<tr>
<td>Family history</td>
<td>Positive</td>
<td>Negative</td>
</tr>
<tr>
<td>SHBG and subunit</td>
<td>Normal</td>
<td>Increased</td>
</tr>
<tr>
<td>T3 suppression test</td>
<td>TSH suppressed</td>
<td>TSH not suppressed</td>
</tr>
<tr>
<td>TRH test</td>
<td>positive</td>
<td>Negative</td>
</tr>
<tr>
<td>Pituitary MRI</td>
<td>Negative</td>
<td>Positive(tumour)</td>
</tr>
<tr>
<td>Molecular study</td>
<td>TR mutation</td>
<td>No Mutation</td>
</tr>
</tbody>
</table>

Conclusion
1. RTH has been described in multiple ethnicities.
2. It is rare but important diagnosis to make
3. Wrong diagnosis can have significant repercussions.
4. TSHoma is an important differential diagnosis (see table above).
5. Family screening and genetic testing is vital.
6. It has implications for inheritance.
7. Management of RTH in pregnancy can be challenging.

References:

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