Association of Iodine Organization Defect and Ectopy of the Thyroid Gland in a Patient with Primary Congenital Hypothyroidism

We describe a neonate with primary congenital hypothyroidism associated with a positive perchlorate discharge test suggestive of a partial iodine organization defect. To our knowledge, such an association has not been described previously. The implication of such an association is discussed.

Neonatal screening programs for congenital hypothyroidism (CH) have revealed an incidence of approximately one in 2500–4500 live births. Primary congenital hypothyroidism is characterized by an absent or hypoplastic thyroid gland in 35% of cases or an inborn error of thyroid hormone metabolism in 22%.

We present a case of primary CH with an ectopic thyroid gland which was associated with a positive perchlorate discharge test suggestive of a partial iodine organization defect. To our knowledge, such a case has not been reported previously. The association of such an association is discussed.
an enlarged gland in a sublingual location with a normal uptake (Fig. 1), thyroid uptake and scintigram following 100 μCi of 131I was performed and showed a sublingual thyroid with high radioactive iodine uptake of 19% and 24% at 6 and 24 h respectively. Percloharate discharge test was performed after oral administration of 100 μCi of radioactive iodine (131I) using a scintillation probe and scaler, 1 and 2 h thyroid uptake was determined followed by 0.5 g oral dose of potassium perchlorate. Thyroid uptake was subsequently measured every 15 min for 1 h and 30 min for an additional 1 h. Percloharate discharge test revealed 32% discharge which indicates partial organization defect. Both parents and siblings were clinically and biochemically euthyroid and in particular there was no evidence of goitre (Table 1). On day 12, the baby was started on l-thyroxine 50 μg daily (11.5 μg/kg/day). At 33 days of age he was clinically and biochemically euthyroid, T3 2.7 nmol/l, T4 213.8 nmol/l, and TSH 6.6 mIU/l (normal range for age, < 10 mU/l).

At the time of writing this child was almost 11 months old. It would be interesting to repeat the perchlorate discharge test to see if the organization defect was transient. However, we believe that it would be unsafe to discontinue therapy for a period of 4–6 weeks in order to carry out this procedure as long as the patient is less than 3 years old. Such an interruption in therapy might affect brain growth. There are other methods of investigating the same problem, for example, by performing a 131I scan and perchlorate discharge test after TSH administration whilst the patient remains on therapy. Unfortunately, we do not have the facilities to carry out this test.

Table 1
Thyroid function tests in parents and siblings of the patient

<table>
<thead>
<tr>
<th>Subject</th>
<th>Age (years)</th>
<th>TSH mIU/l</th>
<th>T3 nmol/l (normal)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>65–160 adults, 1.8–3 adults, 90–200 children, 1.5–4 children</td>
</tr>
<tr>
<td>Father</td>
<td>34</td>
<td>1.1</td>
<td>154</td>
</tr>
<tr>
<td>Mother</td>
<td>26</td>
<td>1.4</td>
<td>140</td>
</tr>
<tr>
<td>Sibling (1)</td>
<td>16</td>
<td>1.1</td>
<td>172</td>
</tr>
<tr>
<td>Sibling (2)</td>
<td>24</td>
<td>0.9</td>
<td>165</td>
</tr>
</tbody>
</table>
Discussion
The presence and location of thyroid tissue in a hypothyroid child has direct bearing on genetic counselling and prognosis. Athyrosis and thyroid ectopy recur very rarely in the same family, whereas hypothyroidism due to defects in thyroxine synthesis or release are inherited as autosomal recessive traits with an estimated frequency of 25% in siblings. The aetiology of the thyroid ectopy is unknown and it may be multifactorial.

The diagnosis of primary hypothyroidism was established in this infant on the basis of high TSH and low T₄ and T₃ values, while ectopy and organification defect were explained on the basis of Tc⁹⁹m, ¹²³I thyroid uptake and scintigraphy, and perchlorate discharge test. The finding of a normal level of serum thyroglobulin in this patient reflects the presence of a significant amount of functioning thyroid tissue. We did not examine urinary iodine excretion in our patient, and the mother received iodine supplements.

hormones are known to play an essential role in the development and growth of the brain, and findings of normal thyroid functions with no clinical evidence of goitre in parents and siblings would not confirm the nature of this association. Finally, endocrinologists and paediatricians taking care of patients with congenital hypothyroidism should consider such a case which might contribute to our understanding of the pathogenesis of congenital hypothyroidism.

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References