The role of early thyroid imaging in infants with congenital hypothyroidism

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Introduction

Congenital hypothyroidism (CH), occurs in 1:4000 to 1:4000 newborns. Most of the cases are sporadic caused by thyroid dysgenesis or dysmorphogenesis and typically require lifelong therapy. However, some of the children diagnosed with CH will have a transient form of the disease. Most of newborns with CH are detected by the routine newborn screening programs and treatment is promptly initiated following confirmatory measurements of serum thyroid stimulating (TSH) and free T4 (FT4) levels. Imaging of the thyroid gland is considered to be essential in determining the underlying etiology of CH. Radiologists scintigraphy and ultrasonography are the most widespread and readily available imaging modalities. Both are not perfect and have diagnostic pitfalls as well as clinical drawbacks. Despite these limitations and the fact that imaging of the gland does not influence the treatment decision or initial dose, it is universally recommended at diagnosis as it may distinguish between permanent and transient CH.

Objectives

- To assess the role of early thyroid imaging in the diagnosis and management of CH.
- To detect early clinical and laboratory factors that will enable a more selective approach for thyroid imaging

Methods

Design
Retrospective study

Setting
Institute for Endocrinology and Diabetes, Schneider Children's Medical Center of Israel (SCMC)

Patients
- Diagnosed with CH at SCMC between the years 2000-2012
- Born at term
- Followed for at least 3 years
- Underwent thyroid imaging

Excluded from the study
- Patients with major congenital malformation or genetic abnormalities
- Treatment with medications that may interfere with thyroid functions

Data collected
- Pregnancy and perinatal history: maternal hypothyroidism or treatment with L-thyroxin during pregnancy
- Anthropometric measurements obtained at diagnosis, 1, 3, 6, 9, 12, and 24 months
- L-thyroxin treatment dose calculated as mcg/kg/day obtained at diagnosis, 1, 3, 6, 9, 12, and 24 months

Imaging studies
thyroid scan and/or thyroid sonography findings:
- Normal size and position gland with normal radiotamide uptake
- Normal gland with increased radiotamide uptake
- Agenesis of gland
- Ectopic gland
- Hypoplastic gland

Infants were categorized into 3 groups:
- Thyroid dysgenesis - infants with agenesis/ectopic thyroid and permanent CH
- Ectopic-permanent - infants with normal sized or hypoplastic thyroid gland and permanent CH
- Ectopic-transient - infants with normal sized or hypoplastic thyroid gland and transient CH

Results

The study cohort

<table>
<thead>
<tr>
<th>Thyroid dysgenesis</th>
<th>Ectopic-permanent</th>
<th>Ectopic-transient</th>
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<tbody>
<tr>
<td>Male (%)</td>
<td>15 (25.8)</td>
<td>39 (69.3)</td>
<td>10 (16.6)</td>
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<tr>
<td>Female (%)</td>
<td>46 (74.2)</td>
<td>16 (27.8)</td>
<td>56 (92.4)</td>
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<td>TSH (mIU/L)</td>
<td>3.55 (1.2)</td>
<td>3.48 (0.6)</td>
<td>3.22 (0.8)</td>
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<tr>
<td>Weight, kg (meanSD)</td>
<td>5.88 (0.8)</td>
<td>6.08 (0.5)</td>
<td>5.94 (0.7)</td>
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<tr>
<td>Gestation (weeks)</td>
<td>39 (39.6)</td>
<td>39 (39.6)</td>
<td>39 (39.6)</td>
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</tbody>
</table>

Overall transient 17/142-11.9%
Average age of L-thyroxin treatment discontinuation (years) 2.3±1.5

Clinical and perinatal characteristics of the study cohort

TSH & FT4 at diagnosis

TSH & FT4 levels during follow-up

Summary & Conclusions

Transient and permanent CH are distinct in TSH levels at diagnosis and thyroxin requirements throughout follow-up.

Early thyroid imaging does not distinguish between permanent and transient CH.

Imaging can be postponed and preformed accordingly to clinical judgment or needs.

A more selective approach for early thyroid imaging in CH is suggested.

DOI: 10.3252/pso.eu.54espe.2015

LT4 dose Eutopic-Permanent vs. Agenesis/Ectopic

LT4 dose Eutopic-Permanent vs. Ectopic-Transient

Transient thyroid imaging requirements
CH: 1.5-3.5 mg/kg/day, FT4: 1.0-1.5 ng/dl