INCIDENCE AND ETIOLOGY OF PRIMARY CONGENITAL HYPOTHYROIDISM IN FLANDERS

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INTRODUCTION

In several European countries, the incidence of primary congenital hypothyroidism (PCH) seems to increase in the last decade, especially in countries that have lowered the newborn screening TSH cutoff.

In Flanders, two centers manage since 2012 the newborn screening for PCH and these maintained the same TSH assay and cutoff (15 mU/L).

AIM

We studied in Flanders:
- Current incidence of PCH
- Etiology of PCH
- Clinical characteristics of infants with PCH
- Initiation of thyroxine treatment

METHOD

Inclusion: All screened infants between 2012 and 2017

Single center (VCBMA), covering about half of the newborns in Flanders.

Collected data when diagnosed with PCH:
- Birth data
- Results of hormonal measurements at screening and at recall
- Scintigraphy and ultrasound results at initial evaluation
- Data on thyroxine treatment initiation

RESULTS

Table 1: Incidence of CHT

<table>
<thead>
<tr>
<th>Year</th>
<th>Screened children</th>
<th>Children with CHT</th>
<th>Incidence of CHT</th>
</tr>
</thead>
<tbody>
<tr>
<td>2012</td>
<td>28511</td>
<td>4</td>
<td>1/7128</td>
</tr>
<tr>
<td>2013</td>
<td>28571</td>
<td>5</td>
<td>1/5714</td>
</tr>
<tr>
<td>2014</td>
<td>28384</td>
<td>3</td>
<td>1/9461</td>
</tr>
<tr>
<td>2015</td>
<td>27535</td>
<td>13</td>
<td>1/2118</td>
</tr>
<tr>
<td>2016</td>
<td>27904</td>
<td>5</td>
<td>1/5581</td>
</tr>
<tr>
<td>2017</td>
<td>26748</td>
<td>10</td>
<td>1/2675</td>
</tr>
<tr>
<td>Total</td>
<td>167653</td>
<td>40</td>
<td>1/4191</td>
</tr>
</tbody>
</table>

During the study period, 167,653 live newborns were screened for PCH, and thyroxine treatment was started in 40 (22 females [F] and 18 males [M]) newborns after confirmed PCH (serum TSH > 20 mU/L at recall).

The yearly number of treated newborns varied greatly (between 3 and 15 per year), but without a clear increase.

A low serum FT4 (< 15 pmol/L) was present in 36 newborns.

Figure 1: Etiology of CHT

Table 2: Clinical characteristics and treatment of CHT

<table>
<thead>
<tr>
<th>Number of cases</th>
<th>CHT median (min-max)</th>
<th>Starting dose median (min-max)</th>
</tr>
</thead>
<tbody>
<tr>
<td>40</td>
<td>11.8 µg/kg/dag (7.7-31.6)</td>
<td></td>
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<td>40</td>
<td>11.8 µg/kg/dag (7.7-31.6)</td>
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<tr>
<td>20</td>
<td>5.1 µg/kg/dag (3.5-6.3)</td>
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<td>20</td>
<td>5.1 µg/kg/dag (3.5-6.3)</td>
<td></td>
</tr>
<tr>
<td>20</td>
<td>2.0 µg/kg/dag (1.3-2.8)</td>
<td></td>
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<tr>
<td>20</td>
<td>2.0 µg/kg/dag (1.3-2.8)</td>
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</tbody>
</table>

Based on scintigraphy (n = 29) and/or ultrasound (n = 32), a diagnosis of gland in situ, ectopy, aplasia and hypoplasia and no imaging was made in respectively 12 (5F), 15 (11F), 10 (5F) and 2 (0F) and 1 (0F) newborns.

The overall incidence of PCH is 1 in 4191 live newborns.

Abnormalities in thyroid morphogenesis are diagnosed in 69%.

No fetal growth difference is observed between the different etiologies of PCH, but a female gender excess is present in the newborns with ectopy.

Newborns with apparent thyroid aplasia receive the highest dose. Thyroxine treatment is initiated before the age of 14 days in 87.5% of the newborns.

REFERENCES


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CONTACT INFORMATION

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