Congenital Hypothyroidism in Algeria: The Emergency of National Neonatal Screening

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Background:
Congenital hypothyroidism (CH) is the commonest congenital endocrine disorder and the primary cause of treatable mental retardation. In low-income countries lacking newborn screening programs, CH remains a serious public health problem.

Objective:
To investigate the characteristics at diagnosis and clinical outcome of patients with CH in Algeria; and determine factors related to psychomotor development.

Methods:
A retrospective study of patients referred for elevated TSH between 2007 and 2017 was conducted in two pediatric clinics in Algiers. Age at diagnosis, clinical characteristics and initial dose of treatment were recorded. Patients were classified as having transient or permanent CH upon imaging and/or re-evaluation at 3 years. Severity was assessed according to venous free thyroxine (FT4) and thyroid stimulating hormone (TSH) at diagnosis. Neurocognitive assessment was based on intelligence quotient (IQ) evaluation using Weschler Preschool and Primary Scale of Intelligence III. Severe cognitive delay was defined as IQ below 70.

Discussion:
Diagnosis of CH remains late in Algeria, with a correspondingly poor neurodevelopmental outcome. These data reinforce the need to view newborn screening as an emergency in our country and in other African countries.

Results:
Between 2007 and 2018, 139 patients (75 girls, 64 boys) were followed for a mean ± SD (range) duration of 3.5±2.5 [0.1-9] years. Mean age at diagnosis and start of treatment was 7.2± 16.3[0.1-138] months. Consanguinity was present in 20(16%) of cases, with a family history of thyroid disease in 47 (34%) and there were 7 familial cases. The most common clinical feature was jaundice, seen in 49 (41%). Delayed development was found in 10 (7%) and short stature in 9(6%) patients, both associated with late diagnosis (>3 months of age). Of the 30 cases re-evaluated at 3 years, 13 (43%) had permanent hypothyroidism. TSH was higher in permanent compared with transient CH, median TSH 125.6 vs 18.4 mIU/l (p< 0.001). Overall IQ in 43 children evaluated aged 6.6±2.2 [2.5-10.8] years was 80.95±17.8 [34-104], IQ >80 was correlated with an earlier mean age at diagnosis: 2.4±1.4 vs 13.3 ±18.5 months p<0.0001.

![Figure 1: Signs and symptoms of CH recorded prior to initiation of treatment](image)

![Figure 2: WPPSI-R: Wechsler Intelligence Scale for Children-Revised](image)

### Table 1: Data for 43 infants with CH, with Global WISCR IQ and relative risk criteria versus age at diagnosis, degree of TSH elevation, and levo-thyroxine (L-T4) dosage

<table>
<thead>
<tr>
<th></th>
<th>Global N= 43</th>
<th>IQ&lt;80 N=21</th>
<th>IQ&gt;80 N=22</th>
<th>p</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age at diagnosis, mean (range)</td>
<td>7,96(0,16-69)</td>
<td>13,3(0,16-69)</td>
<td>2,4(0,23-9)</td>
<td>&lt;0,001</td>
</tr>
<tr>
<td>Serum TSH mIU/l, median (range)</td>
<td>124,7 (11,4-500)</td>
<td>92 (60-100)</td>
<td>151 (11,4-500)</td>
<td>0,001</td>
</tr>
<tr>
<td>L-T4µg/kg/d, mean (range)</td>
<td>7,1 (3-12,5)</td>
<td>7,8 (3,6-12,5)</td>
<td>8,5 (5-10)</td>
<td>0,143</td>
</tr>
<tr>
<td>IQ, mean (range)</td>
<td>80,95(34-104)</td>
<td>61,62(34-78)</td>
<td>92,53(80-104)</td>
<td>&lt;0,001</td>
</tr>
</tbody>
</table>

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