**Background:** Pediatric healthcare is critically dependent on the availability of accurate and precise reference intervals to allow appropriate clinical interpretation.

**Objective and hypotheses:** To obtain reference intervals for TSH, T4, fT4, T3 and T-Uptake in a pediatric population from Córdoba, Argentina.

**Methods:** Serum samples of 807 healthy neonates and infants (age range 2 to 365 days) were analyzed using electrochemiluminescent immunoassay (cobas e 601).

**Results:** No significant differences were observed between the sexes. The percentile 2.5th, 50th and 97.5th were calculated for all reference groups. The TSH, T4, fT4, T3 and T-Uptake levels are shown in table 1.

<table>
<thead>
<tr>
<th></th>
<th>TSH (uIU/mL)</th>
<th>T4 (µg/dL)</th>
<th>fT4 (ng/dL)</th>
<th>T3 (ng/dL)</th>
<th>T-Uptake</th>
</tr>
</thead>
<tbody>
<tr>
<td>Days</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>2-14</td>
<td>77</td>
<td>2.5th - 97.5th</td>
<td>2.5th - 97.5th</td>
<td>2.5th - 97.5th</td>
<td>2.5th - 97.5th</td>
</tr>
<tr>
<td>15-29</td>
<td>96</td>
<td>1.15 - 7.61</td>
<td>7.3 - 16.4</td>
<td>1.1 - 2.0</td>
<td>124.4 - 314.6</td>
</tr>
<tr>
<td>30-89</td>
<td>419</td>
<td>0.85 - 7.79</td>
<td>7.3 - 17.7</td>
<td>1.0 - 2.1</td>
<td>141.9 - 356.5</td>
</tr>
<tr>
<td>90-365</td>
<td>213</td>
<td>0.80 - 7.17</td>
<td>7.1 - 17.0</td>
<td>0.9 - 2.1</td>
<td>142.1 - 370.6</td>
</tr>
</tbody>
</table>

**Table 1:** TSH, T4, fT4, T3 and T-Uptake percentiles

**Conclusions:** We report pediatric reference intervals for TSH, T4, fT4, T3 and T-Uptake. It should assist pediatricians in interpreting these hormonal results more accurately and thereby lead to improved diagnosis of childhood thyroid diseases. Our results reveal that physiological behavior of TSH, T4 and fT4 levels is similar in the four age groups, showing a tendency to decrease at one year of life, whereas T3 values are slightly higher than in the normal period and remain high. This behavior can be the result of different mechanisms, including an increased tissue activity of type I deiodinase and the "reset" of the hypothalamic hypophyseal "set point" for the control of TSH.

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**Familial non-autoimmune hyperthyroidism: activating mutation of thyrotropin receptor gene discovered after three generations of a family**

*Song Hai Lim*¹,²; *Johari Mohd Ali*³; *Loo Ling Wu*¹

¹National University of Malaysia, Department of Paediatrics, Faculty of Medicine, Kuala Lumpur, Malaysia. ²Putrajaya Hospital, Department of Paediatrics, Putrajaya, Malaysia. ³University of Malaya, Department of Molecular Medicine, Faculty of Medicine, Kuala Lumpur, Malaysia

**Background:** Familial Non-autoimmune Hyperthyroidism (FNAH) is a rare etiology for congenital hyperthyroidism due to activating mutation of thyrotropin receptor (TSHR). Recommended treatment is total ablative thyroid tissue by total thyroidectomy followed by radioactive iodine administration.

**Objective and hypotheses:** To describe a family with three generations of members affected by this condition. Index case was a Chinese girl who had been having hyperthyroidism since seven years old and was treated as Graves' disease. Her course of illness was prolonged and difficult to be controlled with anti-thyroid medication. Her mother also suffered from hyperthyroidism and had undergone two thyroid surgeries. The patient delivered a newborn girl at the age of twenty, who was hyperthyroid since birth and remained so by fifteen months old. Thyroid receptor antibody was found to be negative for the patient and her daughter, and subsequently their blood samples were sent to investigate for mutation in TSHR gene.

**Methods:** DNA were isolated from peripheral blood lymphocytes and all TSHR coding exons were amplified, purified and sent for direct DNA sequencing.

**Results:** The index case and her daughter were heterozygous for p.L629F (TTG>TTC) activating mutation in TSHR gene.

**Conclusions:** Diagnosis of FNAH can sometimes be difficult as similar familial predisposition can also be found in autoimmune hyperthyroidism. Genetic confirmation is important to predict outcome and provide most appropriate treatment.

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**Clinical utility of thyroid scans in mild neonatal hyperthyrotropinaemia**

**P2-d3-1164 Thyroid 6**

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**P2-d3-1165 Thyroid 6**

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