Aims

1. To determine the outcome including final diagnosis of patients screening positive for congenital hypothyroidism (CHT) in the North East and Cumbria region.
2. To establish the overall incidence of patients with CHT in the North East and North Cumbria region.
3. To determine the outcomes of infants with bloodspot screening TSH levels of 6-10 mU/L
4. To feedback our regional findings to the National Screening Programme centre (NHS England).

Method

Full Caldicott approval was obtained. All patients screened by the service born between 1st April 2005 and 1st January 2011 were included (and hence older than 3 years at the time of audit). Mean bloodspot TSH greater than 20 mU/L on first screen or greater than 6 mU/L in those subject to repeat testing constituted a positive result. Electronic records of patients identified at the regional screening centre were reviewed and the responsible local paediatrician contacted to establish:

- Whether they were started on thyroxine therapy
- Whether they were still on thyroxine therapy
- Whether they underwent radiological investigation (as per national recommendation).

Results

107 patients screened positive on first or repeat testing. We obtained results for 86.9% (n=93) patients. 75.2% (n=70) patients receiving thyroxine at 3 years of age

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- 25.8% (n=24) of patients underwent thyroid imaging of which 83.3% (n=20) revealed a radiologically normal thyroid gland. Abnormalities were identified in

- 16.7% (n=4) patients of which three patients had thyroid agenesis and one which was not treated.

The estimated incidence of CHT in the North East and North Cumbria is 1 in 3000.

Most Infants with a TSH >6 mU/L do not have classical CHT. NHS Screening programmes: A laboratory guide to newborn screening in the UK for congenital hypothyroidism. February 2014. p.2

Conclusions

The North East and North Cumbria incidence of CHT is similar to previously reported national figures. 8.6% of infants with CHT identified in the North East and North Cumbria Region would not have been identified if the recommended national cut-off (10 mU/L) were used.

The positive predictive value of a borderline TSH value is considerably lower than values over 20 mU/L.

Recommendations

- Continue to audit the regional Newborn CHT screening programme data and develop a more efficient system to feedback patient outcomes to the National screening centre.
- Promote thyroid imaging as a means of helping to determine the diagnosis and predicting which patients require lifelong thyroxine therapy.
- Collaborate to collect national data regarding patients identified with CHT with previously a negative screening test (false-negatives).
- Develop our understanding of the cause of a marginal increase in TSH concentrations and the implications of treating or not treating these patients.

Acknowledgements

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Reference


Table 1 shows positive predictive value of bloodspot TSH results (≥20 mU/L on first or second bloodspot and 6–20 mU/L on second bloodspot).

<table>
<thead>
<tr>
<th>TSH mU/L</th>
<th>Blood Spot TSH</th>
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<tbody>
<tr>
<td>6–10</td>
<td>37.5</td>
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<tr>
<td>10–20</td>
<td>42.8</td>
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<tr>
<td>&gt;20</td>
<td>78.5</td>
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