Auditing the Congenital Hypothyroidism Screening Programme in the North East and Cumbria Region

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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 6-10mU/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 and over therefore had permanent CHT. 16.1% (n=15) had transient hypothyroidism. 6.4% (n=6) patients had normal thyroid function (untreated). 1 patient had thyroid hormone resistance due to a variant in the TSH receptor and the remaining patient had hyperthyrotropinaemia which was not treated.

The estimated incidence of CHT in the North East and North Cumbria is 1 in 3000. This is in keeping with the national incidence as estimated by newborn screening programme of 1 in 3000[1].

Only 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 demonstrated thyroid dysgenesis.

Of those with a TSH 6-9.9 mU/l 57% (n=8) did not have permanent CHT compared to only 8.3% (n=5) of those with a TSH greater than 20 mU/l. (Figure 1).

The initial mean bloodspot TSH correlated with confirmatory serum TSH (Figure 2). The Confirmatory TSH also noted to correlate to the mean T4

Figure 1 -Outcome of Patients with a Positive CHT Newborn Screen and Mean TSH. Normal is defined as a patient with normal serum thyroid function on testing by local paediatric team. Transient represents patients who required thyroxine but was discontinued by 3 years of age.

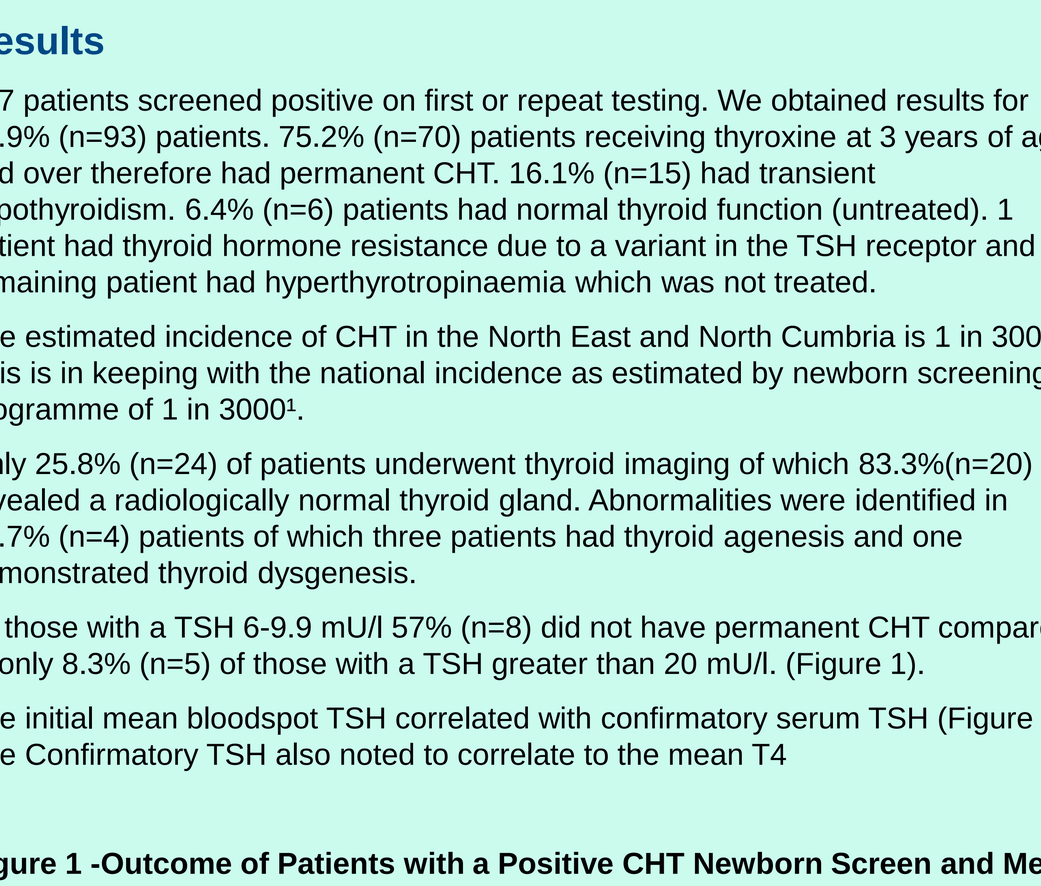

Figure 2. Correlation Between Mean Screening TSH and Confirmatory TSH in mU/L

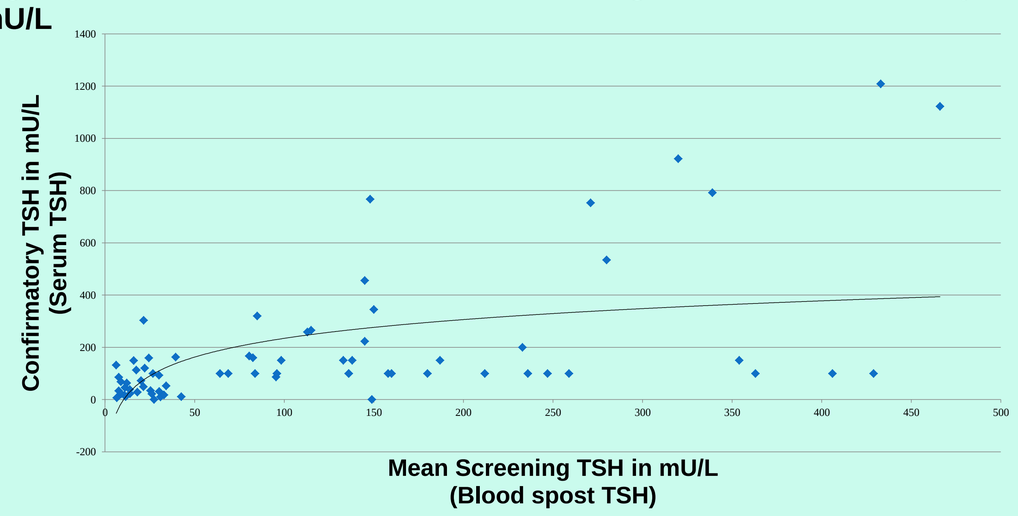

Table 1 shows positive predictive value of bloodspot TSH results (≥ 20 mU/l on first or second bloodspot and 6 – 20mU/l on second blood spot).

Table 1. Estimated Positive Predictive Value of an initial TSH

TSH m/UL	Blood Spot TSH
6-10	37.5
10-20	42.8
>20	78.5

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.

Most Infants with a TSH 6-10 mU/L do not have classical CHT (dysgenesis) and do not have permanent CHT; we do not know whether they benefited from intervention.

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

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Reference

1-- Adapted from NHS Screening programmes: A laboratory guide to newborn screening in the UK for congenital hypothyroidism. February 2014 4 – Newborn Blood Spot Screening Programme – CHT. NHS Newborn Screening [Accessed 20th October 2014] http://newbornbloodspot.screening.nhs.uk/cht.