

Surveillance of Congenital Hypothyroidism Beginning June 2011

UK surveillance of primary congenital hypothyroidism in children aged five years and under (UKCHT).

Primary congenital hypothyroidism (CHT) is a disorder involving reduced thyroxine production by the thyroid gland. This may be due to an abnormally sited or absent thyroid gland or a failure of hormone production within the gland. Babies with CHT may have feeding difficulties, sleepiness, constipation and jaundice. Infants who do not start treatment with replacement oral thyroxine therapy soon after birth, may have problems with their mental development and growth. Each year, around 200 babies with CHT are born in the UK and most are detected by newborn bloodspot (heel-prick) screening. Although newborn screening for CHT started in 1981, we do not know how successful it is in identifying babies who require lifelong therapy.

This UK study aims to determine how many babies and children up to and including five years of age are found each year to have congenital hypothyroidism, diagnosed either subsequent to a positive newborn screening test or because of clinical manifestations. We will describe their characteristics at diagnosis, diagnostic tests undertaken and initial treatment. We will also collect additional information about each child's health after one and two years, particularly to differentiate persistent from transient cases. In a population covered by newborn screening, this study will enable us to determine the incidence and characteristics of all children diagnosed with primary CHT, the proportion and outcomes of those detected by newborn screening, the performance of the newborn screening programme, as well as variations in clinical management and care.

BPSU surveillance will be undertaken for 13 months, commencing in JUNE 2011.

Case definition:

Any child up to and including five years of age who, during the past month, has been referred EITHER for diagnostic confirmation following a newborn screening test result suggestive of primary CHT, OR has been confirmed with a diagnosis of primary CHT (known or considered likely to be present from birth), based on a serum TSH $\geq 10\text{mU/l}$.

REPORTING INSTRUCTIONS

Please report any child *up to and including 5 years of age* who, during the past month, has

- a. been referred for diagnostic confirmation following a newborn screening test result suggestive of primary CHT,
- OR
- b. been confirmed with a diagnosis of primary CHT (known or considered likely to have been present from birth), based on a serum TSH $\geq 10\text{mU/l}$.

Please do not report children in whom it is confirmed that they have:

- Secondary CHT due to pituitary under-production of TSH;
- Acquired hypothyroidism.

If in doubt, or awaiting further tests, please report the child.

Website: <http://www.rcpch.ac.uk/what-we-do/bpsu/current-studies/CHT>

FUNDING: UK NHS Newborn Screening Programme Centre (Department of Health)

ETHICAL APPROVAL: This study has been approved by Cambridge South REC (Ref: 11/EE/0152) and has been granted Section 251 NIGB permission under reference: ECC 3-04(k)/2011.

FURTHER INFORMATION: If you would like any advice regarding the eligibility of a particular case for inclusion in the study please contact: Juliet Oerton: *email* j.oerton@ich.ucl.ac.uk *telephone* 020 7905 2241