Thyroid Disorders of the NEWBORN

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CONGENITAL HYPOTHYROIDISM (CH) – see figure 1

Worldwide, the incidence of congenital hypothyroidism (CH) is about 1 in 4000 babies. Population-based screening programs are vital for the early detection of CH since many affected babies do not have symptoms. The CH screening program in Singapore is based on thyroid stimulating hormone (TSH) levels obtained from umbilical cord blood samples. Diagnosis of primary CH is confirmed from high serum TSH levels and free thyroxine (fT4) levels that are below age-related reference ranges. After diagnosis is confirmed, thyroid antibody assays, radio-nucleide scans and ultrasonography help to determine the aetiology, such as thyroid dysgenesis (85%) due to thyroid aplasia, dysplasia or lingual (ectopic) thyroid, dyshormonogenesis (10%) and other causes (5%). Secondary or hypothalamic-pituitary hypothyroidism is more difficult to diagnose, unless fT4, TSH and thyroxine-binding globulin is simultaneously measured.

Management of CH consists of the rapid normalization of fT4 through supplementation with L-thyroxine, whilst monitoring growth and development. It is important that families caring for CH infants are educated on the need for frequent dose adjustment (during the period of rapid growth) as well as long-term follow-up of cognitive ability. The goal of treatment is to achieve serum fT4 levels in the upper half of the normal reference range. With implementation of a screening program covering practically all newborns in Singapore and early management of affected babies, the incidence of severe intellectual disability from CH has declined over the years.

A majority of preterm babies born at < 30 weeks have lower thyroxine levels than those born at term. The hypo-thyroxinemia appeared transient, may be related to an immature hypothalamic-pituitary axis and was associated with increased post-neonatal morbidity. Ex-preterm infants will need to undergo repeat thyroid function testing, even if they had initially normal cord blood levels. Such repeat tests are usually carried out when they reach a postnatal age equivalent to that of the full-term (by correcting for the degree of prematurity), typically just before hospital discharge. Whether all ex-preterm babies with low thyroxine levels need routine supplementation is uncertain, although it appears prudent to consider treatment.

NEONATAL THYROTOXICOSIS

Newborns with thyrotoxicosis may present with symptoms of excessive metabolism, such as those listed in table 1. In extreme cases, they may develop ventricular tachycardia or supra-ventricular tachycardia, with a risk of heart failure. Diagnosis is confirmed from raised serum fT4 levels, in association with low TSH levels. The management of neonatal thyrotoxicosis consists of antithyroid therapy (either PTU or methimazole), adequate nutrition and monitoring of growth and development. Infants with congestive heart failure may be controlled with propranolol therapy. Long-term follow-up of growth and development is an important part of management. Most thyrotoxic newborns improve rapidly and treatment can be withdrawn over months.

INFANT OF THE MOTHER WITH GRAVES DISEASE

It is not uncommon for babies to be born to mothers with Graves disease who are biochemically euthyroid (normal fT4 and TSH levels) themselves. This is frequently seen because of the close collaboration between the obstetrician and endocrinologist in managing the Graves disease mother. Such collaboration has led to better family planning to defer conception till fT4 levels normalize and use of surgical or radio-nucleide thyroid ablation. Although a Graves disease mother may display no overt manifestation of thyrotoxicosis, circulating thyroid-stimulating immunoglobulins continue to be produced and cross the transplacental barrier, leading to fetal and neonatal thyrotoxicosis in 1% - 5%. Such babies of maternal Graves disease would benefit from repeat thyroid function testing, even if the initial tests were normal at birth, and is best carried out after day 5 when the initial hormone surge has declined to basal values. Most asymptomatic babies with elevated thyroid-stimulating immunoglobulins only need outpatient monitoring of antibody levels, which usually decline over time, reaching basal values by 3 months old. Breastfeeding mothers with Graves disease are best advised to take PTU rather than methimazole because of limited excretion of PTU into breast milk.