

Management of Congenital Hypothyroidism
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Abstract

Congenital hypothyroidism affects about 1 in 2500 newborns in India. This condition can be so subtle that it is often missed in the first few months of life. Neonatal thyroid screening programmes are necessary to diagnose this condition early. When detected at this age and treated effectively the prognosis for intellectual outcome is good. However late detection and treatment leads to irreversible brain damage.

Introduction

The unique and vital actions of thyroid hormone on growth and development are manifest in the first two decades of life and their actual crucial role in foetal and early postnatal development is well established. Disorders of the thyroid gland are the most common endocrine disorders of childhood. The incidence of congenital hypothyroidism in India is about 1 in 2500 live births, whereas world wide it is about 1 in 4000 live births.

Congenital hypothyroidism is one of the very few causes of potentially preventable mental retardation if detected and treated in the first month of life. Because of the subtlety of the clinical manifestations of congenital hypothyroidism in the neonatal period it is often missed. By the time the late manifestations of the disease become evident it is already too late to prevent the residual brain damage. It is therefore of paramount importance that this condition is detected early in the neonatal period and treatment is started immediately.

In the developed world neonatal thyroid screening programmes are in place for well over 2-3 decades now! Unfortunately in India we do not have a uniform congenital hypothyroid screening programme in place and hence most of our congenital hypothyroid babies are not detected early and suffer some permanent residual brain damage.

Diagnosis

A high index of clinical suspicion is necessary to diagnose congenital hypothyroidism early.

Early Clinical Features

(All or none of these may be present in the first month)

- Umbilical hernia
- Pallor and hypothermia
- Enlarged tongue
- Hypotonia
- Prolonged jaundice
- Rough, dry skin
- Open posterior fontanelle
- Relative constipation
- Mild post-maturity
- Heavy baby

Early Clinical Features (1 month plus)

- Umbilical hernia
- Pallor & hypothermia
- Large tongue
- Rough, dry skin
- Constipation

- Facial puffiness
- Growth retardation
- Poor development
- Myxoedema

Laboratory Tests

Laboratory estimation of T_3 , T_4 and TSH should be done on day 5 to 7 of postnatal life in full term neonate because before this TSH is higher and can lead to a false positive diagnosis. Estimations of TSH measured on cord blood can also be misleading unless special standardization for the given population is used. Most babies with congenital hypothyroidism have TSH values greater than 20 mIU/L. 99% of congenital hypothyroidism is due to primary hypothyroidism (Primary defect is in the thyroid gland) and that too due to congenital agenesis or dysgenesis of the gland in most cases. T_3 and T_4 values are low. In rare situations where the congenital hypothyroidism is due to pituitary or hypothalamic defect T_3 , T_4 and TSH are all low and will be missed by the thyroid screening programmes that measures only TSH. But in these cases other pituitary hormone deficiencies frequently coexist leading to clinical abnormalities such as micropenis, underscended testes or diabetes insipidus, thus helping the clinician to make the diagnosis by providing a clinical clue.

Transient Thyroid Dysfunction

All premature neonates have some degree of hypothyroxinaemia which is transient and does not require any treatment.

Transient primary hypothyroidism exists and it can not be distinguished from the permanent condition. Accordingly, all babies with low T_4 and high TSH values should be treated. At a later date (18 months to 3 years), the dose of thyroxine is dropped below replacement level to determine the continuing need demonstrated by a rising TSH concentration.

Transient hyperthyrotropinemia (high TSH and normal T_4) has emerged as a condition because of neonatal screening programmes. This condition does not require treatment but needs follow up with regular T_4 , TSH estimations to exclude ectopic thyroid and dysmorphogenesis. The TSH concentrations gradually fall over a period of 3-6 months.

Treatments

The goal of the therapy is to maintain circulating T_4 level in the upper normal range and normalize the elevated TSH. It is very important that the treatment is started as soon as the diagnosis is made to prevent further brain damage. L-thyroxine is started in a dose of 10-15 mcg/kg/day and they tapered to 5-7 mcg/kg/day to maintain fT_4 at the higher end of the normal range and maintain normal TSH. Over-treatment can lead to craniostenosis. In some cases the TSH levels may remain high for months despite adequate replacement. This is due to acquired hypothalamic feedback insensitivity to circulating thyroxine. Therapy is continued life long except in transient cases.

After infancy the dose is more accurately calculated based on surface area as 100 mcg/m²/day. The dose of thyroxine is usually given on empty stomach early in the morning to set a routine and also to prevent hindrance with absorption

Monitoring

It is important to monitor T_3 , T_4 , TSH levels to assess the adequacy of treatment. After the initiation of therapy T_3 , T_4 , TSH levels are repeated at 1 month. There after it is repeated at 2-3

month interval during infancy, twice or thrice in the second year and twice a year thereafter.

It is also necessary to keep careful record of child's growth. Excessive growth with high growth velocity indicates over-treatment while short stature with poor growth indicates under-replacement. Annual bone age assessment should be a routine and remains a guide to monitoring therapy along with biochemical parameters.

Prognosis

In case where congenital hypothyroidism is detected with neonatal thyroid screening programmes and the treatment is commenced within first month of life prognosis for intellectual development is good. Complete restoration of intellectual performance may not always be possible due to prenatal thyroxine deficiency. If treatment is commenced after 50 days of life there is definite intellectual deterioration which is irreversible. There are few data on the effect of quality of treatment on the outcome. All the evidence to date points to the fact that it is best and of paramount importance to diagnose and treat congenital hypothyroidism as soon as possible and to treat it effectively.

References

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