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Case of Late Presentation of Congenital Hypothyroidism

Introduction

Congenital hypothyroidism is the most common preventable cause of mental retardation in neonates, and is more commonly seen in Indian population with an incidence of 1 in 1000 newborns. The disease has non specific symptoms which are difficult to identify in the neonatal period unless screening is done for thyroid hormones, which is mandatory in all neonates. Symptoms become prominent with age, but neurological symptoms are irreversible at that stage. Causes of congenital hypothyroidism include primary causes, such as thyroid dysgenesis or secondary, which involve central nervous system or peripheral causes which includes resistance to thyroxine. We are presenting a case of late presentation of congenital hypothyroidism.

Case Report

The patient, a 10 year old boy from Davanagere born to parents with second degree consanguinous marriage presented to the Paediatric OPD with history of poor growth since 5 years. All developmental domains were normal [gross and fine motor skills, social skills, language, auditory and visual]. On presentation, height was 110 cm, (<-3D), weight was 14 kg (<-3SD) and head circumference was 48 cm (<-2SD) which suggests chronic malnutrition. In accordance to these parameters, child also has severe wasting and stunting secondary to missed diagnosis of dyshormonogenesis. On examination, few scaly patches, erythema, generalised xerosis along with anal fissure (secondary to chronic constipation) was noted. All other systemic examinations were normal.

On suspicion of thyroid disorder, TSH and T4 levels were checked. Elevated TSH (>100 uIU/mL) and low T4 (3.37 mcg/dl)levels were found suggesting primary hypothyroidism. To confirm the diagnosis Tc99 thyroid scan was done, which showed increased radioactive uptake in both lobes consistent with high TSH confirming eutopic thyroid gland secondary to dyshormonogenesis.
Following diagnosis, child was started on medication for hypothyroidism and conservative management for anal fissure was done.

Discussion

Features of congenital hypothyroidism are nonspecific and difficult to identify in neonates. Absence of neonatal screening causes missed diagnosis and patient presents with disease in later life with permanent neurological deficit, hence screening for hypothyroidism is mandatory in neonates.

Consanguineous marriages increase the chances of the newborn suffering from congenital hypothyroidism, secondary to dyshormonogenesis.

Keywords: eutopic thyroid , dyshormonogenesis, hypothyroidism

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