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# Congenital hypothyroidism: role of laboratory in its diagnosis

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## Abstract

Congenital hypothyroidism is one of the most common endocrine disorders in newborns. The slow development of clinical symptoms, emphasis on newborn screening and early treatment to prevent complications like delay in milestones and developmental anomalies

**Case Report:** This paper presents a case of congenital hypothyroidism that was born by LSCS with foetal distress. A female newborn , 2 months old was admitted in the NICU with complaints of jaundice which was persistent, even after 1 week of birth,C/O of constipation and abdominal distension.

**Conclusion:** The purpose of this study is to report a case of congenital hypothyroidism in a 2 months old child. Early detection and adequate replacement therapy is very important as this could protect them against irreversible growth and mental retardation.

**Keywords:** Congenital hypothyroidism, newborn screening, mental retardation

## Introduction

Congenital hypothyroidism (CH) is defined as thyroid hormone deficiency present at birth. Problems with thyroid gland development (dysgenesis) and disorders of thyroid hormone biosynthesis (dyshormonogenesis) are the most common causes of congenital hypothyroidism.[1] CH is an important cause of mental retardation which can be preventable. Since the clinical manifestations of CH are often subtle or absent at birth, many newborns remain undiagnosed and may later suffer from mental retardation. [2]

The overall incidence of CH ranges from 1:3000 to 1:4000 globally. Although the exact incidence of CH in India is unknown, a considerably older study conducted in 1998 reported the incidence to be 1:2640 in neonates. [3]

## Case Report

A female newborn, 28days old, was born by LSCS with foetal distress. The new born was admitted in NICU of our institution with C/O constipation and abdominal

distension since birth .The baby was icteric, which was persistent from 2nd day of birth.

The baby had immediately cried after birth and developed difficult to pass stools with abdominal distension after taking feeds since birth.

Baby had jaundice from day 2 of life and also the mother gave the history of baby having drowsiness and poor feeding. There was no H/O fever and bladder disturbance

The new born with the above complaints was first taken to a tertiary care setup, before being admitted in our hospital. There the rectal washes were given after which the baby used to pass stools. Contrast enema showed dilated rectum and distal colon.

Rectal biopsy was done to rule out Hirsch sprung disease (HSD) .Anyway biopsy revealed normal innervation excluding HSD.

There is no H/O hypothyroidism or any thyroid related disorder in the family. The (baby was born by) mother revealed that it was a consanguineous marriage.

Per abdomen examination revealed distension of the abdomen with normal bowel sounds. Respiratory and cardiac examination were within normal limits

The blood investigation were done for TSH which measured 835.5µIU/mL, with low levels of total T4 and T3 (T4 was 5.4nmol/L, T3 was <0.3 nmol/L). Liver function Test showed increased Total bilirubin of 12.25 mg/dL which was mainly due to elevated unconjugated bilirubin.

Accordingly, diagnosis of congenital hypothyroidism was made and replacement therapy with levothyroxine of dose 37.5µg was started to normalize the levels of TSH.Regular rectal wash was recommended to treat constipation.

**Discussion**

The Diagnosis of CH at birth by mere clinical findings is very difficult and screening of new born plays an important role. In India screening of newborn infants for CH is absolutely required and remains challenging due to lack of laboratory facilities, infrastructure and policies of health care especially in the rural areas. [4] The first Indian Newborn Screening (NBS) program was conducted in Bangalore in 1980 for screening of various metabolic disorders responsible for mental retardation. [5] Early detection and treatment of CH with the assistance of NBS programs will improve the outcomes in children and neonates.[6]

The CH diagnosis is based on the thyroid function tests; the results are interpreted based on the values of T4 and TSH. According to American Academy of Pediatrics (AAP) all infants with low T4 concentration and TSH concentration greater than 40 mU/ L are considered to have congenital hypothyroidism. Screening of primary CH is done by measuring thyroid-stimulating hormone (TSH) in cord blood or blood collected after 24 hours of birth and followed by retesting after 48–72 hours of birth after normal term delivery along with thorough follow up. For detecting infants with delayed TSH rise, repeated screening after few weeks is recommended, especially in preterm births. [7]

Hence there is absolute requirement of having well equipped laboratories, stringent health policies and improvisation of health sector for diagnosis of CH. This is because any diagnostic delay, inadequate treatment or even poor compliance to treatment is always associated with irreversible damage. [8]

**Conclusion**

CH is one of the leading causes for mental retardation and can be reversible with proper treatment strategies. This condition warrants the development of protocol for reliable laboratory screening and awareness in parents about proper treatment and follow up for better outcomes.

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