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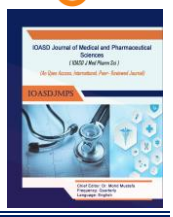

Congenital Hypothyroidism-Role of Clinical Laboratory from Screening to Follow up

Smita Sharma[1*], Rami Abdullah Ali Al Daghreer[2], and Hamad Al Daghreer[2]
[1]Associate Professor Biochemistry, M.M.Institute of Medical Sciences and Research, Mullana, Ambala, India
[2]Ministry of Health, Nejran. KSA

Corresponding Author: Smita Sharma
Associate Professor Biochemistry, M.M.Institute of Medical Sciences and Research, Mullana, Ambala, India | Email: dr.smitasharma@yahoo.com

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ABSTRACT

Background: Congenital hypothyroidism represents a leading preventable cause of cognitive impairment. Indian data shows an incidence of 1:800 to 1: 1700 new-borns. ***Objectives:*** The review outlines the contribution of clinical laboratories in congenital hypothyroidism detection, confirmation and longitudinal monitoring according to current ISPAE and AAP recommendations. ***Conclusion:*** Establishing universal cord blood screening with defined alert limits and fast reporting can ensure therapy starts within the first 48 hours.

Key words: Congenital Hypothyroidism, Neonatal Screening, Thyroid Stimulating Hormone, Free Thyroxine, Clinical Laboratory, ISPAE.

Review Article

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1. INTRODUCTION

Thyroid hormone plays a key role in neuronal growth during early infancy.CH arises when there is insufficient hormone production at birth. The commonest cause is thyroid gland maldevelopment. Without prompt replacement, it results in permanent developmental delay and growth issues.

In the absence of nationwide new-born screening programme in India, individual hospital labs carry the responsibility of early detection. This review helps how biochemical laboratories can bridge this gap.

2. Burden of disease in India

While worldwide prevalence is 1:3000-1:4000, Indian studies report nearly double to triple that rate at 1:800-1:1700. This higher burden makes systemic screening a public health priority. Many cases are still diagnosed late due to absence of routine testing.

3. Screening Strategies from Lab View point

As per ISPAE 2018, every new-born in India should be tested.

Table 1: Screening sample options

Sample	Collection Time
Cord Blood	Immediately after delivery
Dried Blood Spot	48-72 hrs of life
Venous Blood	As required

ISPAE advises cord TSH for all babies. If cord TSH falls between 20-40 µIU/ml, a repeat DBS at 48-72 h is suggested.

Analytical Methods: Most labs use CLIA.Screening is done by TSH alone. Abnormal results are followed by full panel: TSH, FT4, FT3 on serum.

4. Alerts limits and Turnaround Time

Rapid communication saves brain cells.

ISPAE alert thresholds:

I. TSH > 40µIU/L: Urgent verbal intimation plus request for confirmatory testing.
II. TSH>80µIU/L: Advise to begin levothyroxine without waiting for repeat results.
III. FT4 < 5 pmol/L: Indicates severe deficiency, needs immediate action.

Ideal laboratory TAT: < 4 hours for screening TSH AND < 24 hours for diagnostic panel.

5. Biochemical Interpretation by the Lab

Table 2: Pattern of Thyroid Tests

Diagnosis	TSH
Primary CH	Markedly raised
Central CH	Normal/Low
Transient CH	Raised
TBG Deficiency	Normal

The laboratory differentiates these patterns and guides the clinician.

6. Follow-up Testing Protocol

Goal is to keep FT4 in upper normal range and TSH in target range.

Table 3: ISPAE Recommended Follow up

Time Point	Tests Required	Goal
14 days	FT4, TSH	FT4 in upper normal range
1 month	TSH, FT4	TSH<10µIU/mL
2 to 6 months	TSH every 8 weeks	TSH 0.5-5µIU/mL
>6 months	TSH every 12-16 weeks	TSH 0.5-5µIU/mL

Labs must have a system to flag abnormal follow-up results.

7. Road blocks in India

I. Absence of national NBS policy.
II. Difficulty in tracking babies for DBS collection
III. Need for round-the-clock lab services.
IV. Cost of immunoassay platforms.

8. CONCLUSION

Clinical laboratories are central to preventing intellectual disability from CH. By adopting cord blood screening, setting clear critical value policies and maintaining rapid TAT, labs can ensure treatment begins within 48 hrs. A national universal program is need of the hour.

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