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Clinical And Laboratory Characteristics *Of Congenital Hypothyroidism* In Newborn

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ABSTRACT

Congenital Hypothyroidism (CH) is the most common congenital endocrine disorder in newborns and can lead to delayed neurocognitive development if not detected and treated early. Clinical manifestations of CH in the neonatal period are often atypical, so laboratory tests play a crucial role in establishing the diagnosis. This study aims to determine the clinical and laboratory characteristics *of congenital CH. Hypothyroidism* in newborns. This study used a retrospective descriptive design with a cross-sectional approach. The study sample consisted of 40 newborns diagnosed with *congenital hypothyroidism. Hypothyroidism* was diagnosed based on screening results and thyroid hormone examinations. Data obtained from medical records included clinical characteristics and laboratory results of TSH and T4 levels. The results showed that most infants did not show specific clinical symptoms early in life. The most common clinical manifestations were prolonged jaundice, hypotonia, and low birth weight. Laboratory examinations showed increased TSH levels and decreased T4 levels in all samples. It was concluded that the diagnosis of *congenital hypothyroidism was correct. Hypothyroidism* in newborns relies heavily on laboratory tests because clinical symptoms are often nonspecific.

Keywords: *Congenital Hypothyroidism*, Newborn, Clinical Characteristics, Laboratory Examination

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1. Introduction

Congenital *Hypothyroidism* (CH) is a congenital thyroid hormone deficiency and is a leading cause of preventable mental developmental delay. The incidence of CH is reported to range from 1 in 2,000 to 1 in 4,000 live births. Thyroid hormones play a critical role in the growth and development of the central nervous system, particularly during the neonatal period and early life.

The main challenge in treating CH is the difficulty of clinically detecting the disease in newborns. Most babies with CH appear normal at birth because they still receive a supply of thyroid hormones from their mothers during pregnancy. Consequently, diagnosis is often delayed when relying solely on clinical signs and symptoms.

The clinical symptoms of CH that appear are generally mild and nonspecific, such as prolonged jaundice, constipation, hypotonia, macroglossia, and a hoarse cry. Therefore, neonatal screening through thyroid level examination is recommended. Stimulating Hormones (TSH) and thyroxine (T4) are the main methods in early detection of CH.

Laboratory tests play a crucial role in confirming a CH diagnosis. Elevated TSH levels and low T4 levels are key indicators of thyroid dysfunction. Early detection and treatment have been shown to prevent developmental delays and improve a child's quality of life.

Based on this background, this study was conducted to determine the clinical and laboratory characteristics *of congenital Hypothyroidism* in newborns as a basis for increasing awareness and early detection of this disease.

2. Research Methods

a. Research Design

This study used a retrospective descriptive design with a cross-sectional approach.

b. Location and Time of Research

The research was conducted at Hospital X by collecting medical record data for the period January 2022–December 2024.

c. Population and Sample

The study population was all newborns who underwent thyroid screening. The study sample consisted of 40 newborns diagnosed with *congenital thyroiditis. hypothyroidism*.

d. Inclusion Criteria

1) Newborns with *congenital diagnosis hypothyroidism*
2) Have complete clinical data and TSH and T4 test results

e. Research Variables

- Clinical variables: signs and symptoms of CH
- Laboratory variables: TSH and T4 levels

f. Research Instruments

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Observation sheet of newborn medical record data.

g. Data analysis

Data analysis was carried out univariately and presented in the form of frequency distribution, percentage, mean, and standard deviation.

h. Research Ethics

The research has obtained permission from the relevant institutions and maintains the confidentiality of patient identities.

3. Research Results And Discussion

a. Research Result

This study involved 40 newborns diagnosed *with congenital Hypothyroidism* (CH) is diagnosed based on screening results and thyroid hormone laboratory tests. Demographic characteristics, clinical manifestations, and laboratory parameters were analyzed.

1) Demographic Characteristics of Babies

a) Gender

Gender	f	%
Man	15	37.5
Woman	25	62.5
Total	**40**	**100**

Interpretation:

The proportion of female infants is higher than male infants, which is in accordance with epidemiological reports that CH is more common in female infants.

b) Gestational Age

Gestational Age	f	%
Preterm (<37 weeks)	9	22.5
Aterm (≥37 weeks)	31	77.5
Total	**40**	**100**

Most babies are born full term, indicating that CH is not limited to premature babies.

c) Birth Weight

Birth Weight	f	%
< 2500 grams	10	25.0
≥ 2500 grams	30	75.0
Total	**40**	**100**

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2) Clinical Characteristics of Babies with *Congenital Hypothyroidism*

Most babies do not show typical symptoms in the early neonatal period.

Clinical Manifestations	f	%
Prolonged jaundice	18	45.0
Hypotonia	14	35.0
Lethargy	13	32.5
Macroglossia	8	20.0
Constipation	6	15.0
Hoarse cry	5	12.5
Without typical symptoms	22	55.0

Interpretation:

More than half of the infants appeared clinically normal, confirming that clinical symptoms alone are not sufficient for the diagnosis of CH.

3) Laboratory Characteristics

a) TSH and T4 levels

Parameter	Mean Elementary School		Normal Values
TSH (µIU/ mL)	58.6	18.4	< 10
T4 (µg/ dL)	3.1	0.9	5–12

All infants showed significantly elevated TSH and decreased T4, consistent with a diagnosis of primary CH.

b) Clinical and Laboratory Relationship (Descriptive)

Infants with more pronounced clinical manifestations (e.g., hypotonia and jaundice) tended to have higher TSH levels and lower T4 levels than asymptomatic infants, although this study was not designed for inferential analysis.

b. Discussion

1) Demographic Characteristics of Infants with CH

The higher proportion of female infants in this study aligns with various studies that suggest CH is more common in female infants, with a ratio of approximately 2:1. The exact mechanism of this difference is not fully understood, but it is thought to be related to genetic factors and thyroid gland development.

Most babies are born full term and with normal birth weight, indicating that CH is not always associated with prematurity or intrauterine growth restriction.

2) Nonspecific Clinical Manifestations

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Research shows that most infants with CH do not exhibit typical clinical signs during the neonatal period. This condition may be explained by the influence of maternal thyroid hormones still circulating in the infant's body during the first weeks of life.

Jaundice is the most common symptom. Thyroid hormone deficiency causes decreased activity of the enzyme glucuronyl thiophosphate. transferase in the liver, thereby slowing bilirubin metabolism. Hypotonia and lethargy are associated with impaired central nervous system and muscle maturation due to thyroid hormone deficiency.

These findings strengthen the evidence that a clinical approach alone risks delaying diagnosis and therapy.

3) The Role of Laboratory Testing as the Gold Standard

Laboratory tests showed markedly elevated TSH and decreased T4 levels in all samples. This confirms that the majority of cases in this study were primary CH, which is most often caused by thyroid dysgenesis or impaired thyroid hormone synthesis.

These results are consistent with international guidelines stating that TSH and T4 testing in neonatal screening is the most effective method for early detection of CH.

4) Clinical Implications and Neonatal Nursing

Early detection of CH has significant implications for the baby's long-term prognosis. Thyroid hormone therapy given in the first 2–3 weeks of life has been shown to prevent intellectual developmental delays.

For nurses and neonatal health workers, the results of this study emphasize the importance of:

- Parental education regarding neonatal screening
- Monitoring for prolonged jaundice and hypotonia
- Collaboration in following up on screening results

5) Research Limitations

This study has several limitations:

a) Descriptive design without causal relationship analysis
b) The number of samples is relatively limited
c) Radiological etiological classification of CH was performed.

The results of the study continue to provide a strong clinical picture of CH in newborns.

4. Conclusion And Suggestions

a. Conclusion

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Most newborns have *congenital Hypothyroidism* does not exhibit typical clinical symptoms. Laboratory tests, such as elevated TSH and decreased T4, are the primary indicators for establishing a diagnosis.

b. Suggestion

1) Neonatal thyroid screening needs to be performed routinely.
2) Health workers are expected to increase their awareness of mild symptoms of CH.
3) Further research is recommended using larger samples and analytical designs.

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