Case Report

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Hypothyroidism in a Case of Down Syndrome: A Case Report.

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Abstract

A 28-day-old male neonate with Down Syndrome presented with persistent jaundice and was admitted to the NICU. In this case characteristic physical features of Down syndrome were present. Karyotyping confirmed the diagnosis of Downs syndrome. Further investigations included ultrasonography of the thyroid gland, which showed a reduced gland size, suggestive of thyroid dysgenesis. Abnormal thyroid function tests led to the diagnosis of hypothyroidism. Management with levothyroxine was started in appropriate doses. This report highlights the prevalence and implications of thyroid dysfunction in neonates with Down syndrome and underscores the importance of vigilant screening and comprehensive management to improve outcomes in this population.

Keywords:- Down Syndrome, Congenital Hypothyroidism, Neonatal Jaundice, Levothyroxine Therapy

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INTRODUCTION

Down syndrome, the most common chromosomal disorder, results from a full or partial extra copy of chromosome 21, termed trisomy 21. Characterized by intellectual disability, distinct facial features, and various systemic complications, it affects approximately 1 in every 700 births worldwide. This congenital condition has significant clinical and public health implications, warranting a multidisciplinary approach to management and care.[1]

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The coexistence of Down syndrome with other medical conditions like congenital heart disease, leukaemia, and thyroid disorders is well-documented.[2] Hypothyroidism, in particular, is more prevalent among individuals with Down syndrome than in the general population. This may be attributed to the autoimmune nature of the disorder or the intrinsic dysgenesis of the thyroid gland common in these patients.[3]

Clinically, neonates with Down syndrome often present with features like hypotonia, poor feeding, and developmental delays. Specific challenges include diagnosing hypothyroidism early, as symptoms such as jaundice and lethargy may be mistakenly attributed to other neonatal conditions.[4] Timely diagnosis and management of thyroid dysfunction are crucial, as untreated hypothyroidism can exacerbate cognitive impairments and growth failure.[5]

This case report highlights an important clinical finding of presence of hypothyroidism in a neonate with Down syndrome. The combination of these conditions emphasizes the need for vigilant screening and management strategies in this population to optimize health outcomes.

CASE REPORT

The patient was a 28-day-old male neonate, born at term to a 26-year-old primigravida mother via vaginal delivery. The pregnancy and birth history were unremarkable. The neonate was admitted to the Neonatal Intensive Care Unit (NICU) due to prolonged jaundice.

On examination, the neonate displayed characteristic features of Down syndrome, including brachycephaly, epicanthal folds, and a protruding tongue. Initial blood tests indicated elevated levels of bilirubin, and subsequent thyroid function tests revealed markedly low levels of thyroxine (T4) and elevated thyroid-stimulating hormone (TSH), confirming congenital hypothyroidism (Table 1).

Test	Result	Normal. Range	Interpretation
TSH (Thyroid Stimulating Hormone)	15 mIU/L	0.5 - 10 mIU/L	Elevated TSH indicates primary hypothyroidism, common in congenital cases.
Free T4 (Thyroxine)	0.5 ng/dL	0.9 - 2.3 ng/dL	Decreased T4 levels are indicative of hypothyroidism.
Total T4 (Thyroxine)	3 µg/dL	5 - 12.3 µg/dL	Below normal range, consistent with hypothyroidism.

Table 1 : Thyroid Function test in studied case.

Karyotyping confirmed the diagnosis of Downs syndrome. Further investigations included ultrasonography of the thyroid gland, which showed a reduced gland size, suggestive of thyroid dysgenesis. Management included starting the neonate on levothyroxine, with careful monitoring of thyroid function and developmental status. Over the following weeks, there was significant improvement in jaundice, and thyroid function tests normalized.

DISCUSSION

The interplay between Down syndrome and hypothyroidism presents a unique clinical challenge.[6] In neonates with Down syndrome, the prevalence of hypothyroidism can be as high as 28%, compared to about 1 in 4000 in the general neonatal population. This case parallels several reported instances where congenital hypothyroidism in Down syndrome was initially overlooked due to nonspecific symptoms like jaundice and hypotonia.[7]

Published reports, such as those by Cutler AT et al[8] and Gibson PA et al[9] have emphasized the importance of routine screening for thyroid dysfunctions in patients with Down syndrome.

These studies advocate for early intervention strategies, including the initiation of levothyroxine therapy, to prevent potential neurodevelopmental delay and growth impairment.[10]

This case underscores the need for heightened surveillance and tailored healthcare protocols to address the dual burden of Down syndrome and hypothyroidism. The prompt diagnosis and management in this case likely averted further complications, illustrating the benefits of integrated care approaches in managing complex congenital disorders.

CONCLUSION

The coexistence of Down Syndrome and hypothyroidism in neonates underscores the necessity for vigilant screening and early therapeutic intervention. This case emphasizes the complex interplay between genetic and endocrine factors, highlighting the need for integrated care approaches to optimize outcomes for affected neonates.

Conflict of interest
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