Case Report

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Thyroid Dysgenesis Causing Congenital Hypothyroidism: A Case Report.

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Abstract

Neonatal hypothyroidism, characterized by insufficient thyroid hormone levels in the neonatal period, is a critical endocrine disorder with potential for severe neurodevelopmental sequelae. Neonatal hypothyroidism is generally diagnosed late. An early diagnosis and prompt hormone replacement therapy is essential in cases of neonatal hypothyroidism to prevent irreversible mental retardation. We present the case of a 1-month-old male infant diagnosed with congenital hypothyroidism who presented with lethargy, cool and mottled skin, and a hoarse cry. A presumptive diagnosis of neonatal hypothyroidism was made. Knee X-Ray was done which showed absent epiphysis suggestive of delayed bone age. Thyroid function test was done which confirmed hypothyroidism. Hormone replacement therapy was promptly initiated and parents were counselled about importance of continued hormone replacement therapy and regular follow up. This case emphasizes the importance of early detection and prompt management to prevent irreversible cognitive impairments. This case underscores the significance of nationwide neonatal screening programs and individualized hormone replacement therapy.

Keywords:- Neonatal Hypothyroidism, Thyroid function test, Hormone replacement therapy, Mental retardation.

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INTRODUCTION

Neonatal hypothyroidism, a vital concern in pediatric endocrinology, results from disrupted thyroid hormone synthesis, profoundly impacting growth, neurodevelopment, and metabolism. Comprehending the disorder's complexities and promptly intervening are imperative for optimal outcomes. The disorder encompasses congenital hypothyroidism (CH) and acquired hypothyroidism. CH is primarily rooted in thyroid gland developmental issues or genetic mutations, affecting hormone synthesis.

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Acquired hypothyroidism arises from extrinsic factors, including iodine deficiency and maternal autoimmune conditions.[1]

Globally, neonatal hypothyroidism affects about 1 in 2,000 to 4,000 live births. National screening initiatives, utilizing thyroid-stimulating hormone (TSH) and thyroxine (T4) measurements, facilitate early detection, enabling timely intervention to avert cognitive deficits. Genetic research has unveiled critical genes influencing thyroid development and hormone synthesis. Mutations in genes related to thyroid transcription factors, enzymes, and TSH receptors provide insights into etiology and potential therapeutic avenues.Diagnostic approaches have evolved, incorporating dried blood spots (DBS) for TSH and T4 assessment. This technique offers cost-effective and practical screening, especially in resource-limited settings.[2]

Effective management hinges on hormone replacement therapy (HRT) using synthetic levothyroxine (L-T4). Initiating therapy promptly and personalizing dosages based on age, severity, and comorbidities are central. Vigilant monitoring ensures optimal adjustments.In conclusion, neonatal hypothyroidism's multifaceted nature demands a holistic approach. Genetic insights, innovative diagnostics like DBS, and tailored HRT collectively advance patient care. This comprehensive review synthesizes the disorder's intricacies, empowering clinicians to navigate its challenges adeptly.[3]

We Present here a case of congenital hypothyroidism who was brought for respiratory tract infection and was later diagnosed to be having neonatal hypothyroidism.

CASE REPORT

A 1-month-old male infant presented to the pediatric Out patient department (OPD) with concerns of poor feeding, prolonged jaundice, and lethargy. The infant was born full-term via uncomplicated vaginal delivery, with no perinatal complications noted. Family history was unremarkable for thyroid disorders. On physical examination, the infant exhibited lethargy, cool and mottled skin, and a hoarse cry. The anterior fontanelle was enlarged, and the umbilical hernia was noted. Further evaluation revealed delayed reflexes and poor muscle tone.

On the basis of clinical examination, a possible diagnosis of congenital or neonatal hypothyroidism was made. X-Ray of Knee was done which showed absent epiphysis. Laboratory investigations demonstrated elevated levels of TSH (148 mIU/L; reference range:

0.5-5.0 mIU/L) and decreased T4 (2.8 µg/dL; reference range: 6.5-14.0 µg/dL), consistent with hypothyroidism. Thyroid ultrasound revealed an absent thyroid gland, confirming the diagnosis of congenital hypothyroidism, specifically thyroid dysgenesis. Brain imaging showed no evident abnormalities. Levothyroxine (L-T4) therapy was promptly initiated at 10 µg/kg/day. The infant's clinical status improved gradually, with increased alertness and improved muscle tone observed over the subsequent weeks. Laboratory follow-up demonstrated a progressive decrease in TSH levels and a rise in T4 levels, indicating an adequate hormonal response to L-T4 replacement therapy. The infant's parents were educated about the lifelong need for thyroid hormone supplementation and the importance of regular follow-up.

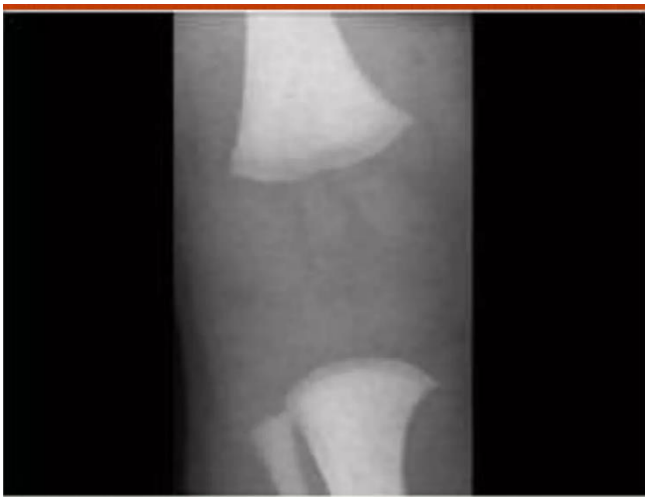

Figure 1: Absent Knee Epiphysis strongly suggestive of delayed bone age.

DISCUSION

Neonatal hypothyroidism, characterized by deficient thyroid hormone levels during the critical neonatal period, remains a significant concern due to its potential impact on neurodevelopment and growth. This discussion delves into the presented case of a 1-month-old male infant diagnosed with congenital hypothyroidism, emphasizing the clinical implications, diagnostic strategies, therapeutic interventions, and the broader significance of early detection.[4]

The case underlines the importance of recognizing the classic clinical features associated with neonatal hypothyroidism, including feeding difficulties, prolonged jaundice, hypotonia, and lethargy. These indicators, although nonspecific, should trigger clinical suspicion, prompting a comprehensive thyroid function evaluation. Elevated thyroid-stimulating hormone (TSH) and decreased thyroxine (T4) levels confirmed the diagnosis, consistent with previous literature highlighting TSH as a sensitive marker for neonatal hypothyroidism.[5]

Thyroid ultrasound, a crucial diagnostic adjunct, confirmed the absence of a palpable thyroid gland,

pointing towards congenital hypothyroidism due to thyroid dysgenesis. This observation underscores the value of imaging modalities in elucidating the etiology and guiding subsequent management decisions. Genetic testing could further illuminate the genetic basis underlying thyroid dysgenesis, offering insights into the molecular pathogenesis.[6]

Timely initiation of levothyroxine (L-T4) replacement therapy, a cornerstone in the management of congenital hypothyroidism, led to a favorable clinical response. Regular monitoring of thyroid function and clinical parameters allowed for personalized dose adjustments, reflective of the patient's evolving needs. The case highlights the importance of monitoring TSH and T4 levels closely, ensuring euthyroid status to prevent not only neurodevelopmental impairment but also facilitating optimal somatic growth.[7]

The presented case aligns with the global practice of neonatal screening, wherein the prompt identification of infants with hypothyroidism through nationwide programs averts adverse cognitive outcomes. Early detection of neonatal hypothyroidism can be attributed to these initiatives, emphasizing the significance of standardized screening protocols utilizing TSH and T4 measurements. The case also highlights the cost-effective utility of dried blood spot (DBS) sampling for TSH and T4 analysis, particularly relevant in resource-limited settings.[8]

Moreover, the case emphasizes the importance of individualized care in neonatal hypothyroidism management. The age of initiation, etiology of hypothyroidism, and comorbidities dictate treatment strategies and dosage titration. Close collaboration between pediatric endocrinologists, neonatologists, and caregivers is paramount in ensuring optimal therapeutic outcomes.[9]

In conclusion, the presented case serves as a poignant reminder of the impact of neonatal hypothyroidism on neurodevelopment and growth. Early detection through neonatal screening programs, accurate diagnosis, and prompt institution of hormone replacement therapy hold the key to averting irreversible cognitive deficits. The case underscores the crucial role of vigilant clinical acumen, advanced diagnostics, and tailored interventions, emphasizing the significance of collaborative efforts in optimizing the long-term prognosis for infants affected by neonatal hypothyroidism.[10]

CONCLUSION

Thyroid dysgenesis is an important cause of congenital hypothyroidism. Its important to make an early diagnosis and promptly start hormone replacement therapy. Any delay in the diagnosis can cause catastrophic consequences such as irreversible mental retardation.

Conflict of interest

None

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