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Congenital Hypothyroidism with Atrial Septal Defect Accompanied and Severe Anemia in A 15-Month-Old Infant:A Case Report from A Resource Limitec Tanzanian Setting

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ABSTRACT

Congenital hypothyroidism (CH) is a condition characterized by the thyroid gland's inability to synthesize sufficient quantities of thyroid hormones in neonates. These hormones are indispensable for normal cerebral development, growth, and metabolic functions in infants. Often, it constitutes a permanent condition that necessitates lifelong management. The incidence of congenital hypothyroidism is estimated to be approximately 1 in 2,000 to 4,000 live births. This condition can be classified based on its etiology into "Primary" or "Secondary" hypothyroidism, the latter also referred to as "Central" hypothyroidism, or based on its duration into "Transient," indicating potential improvement over time, or "Permanent" hypothyroidism. The predominant congenital form of hypothyroidism is primary hypothyroidism, primarily resulting from thyroid dysgenesis, which accounts for approximately 80% of cases. Notably, the majority of infants diagnosed with CH are typically asymptomatic at birth.

This paper elucidates a case of congenital hypothyroidism (CH) in a 15-month-old toddler who presented at our pediatric ward exhibiting failure to thrive, chronic constipation, delayed developmental milestones, and multiple congenital anomalies, including congenital heart disease and umbilical hernia. These manifestations arose from the protracted challenges in diagnosis attributable to the absence of policies for newborn screening and maternal thyroid hormone assessment during antenatal care (ANC) in many developing nations, with Tanzania being a notable example

The significance of this case report is to create awareness on presence of rare associated anomalies and to identify gaps in health care accessand also to sdhow the long-term outcomes of delayed vs timely intervention.

Key words: Congenital hypothyroidism (CH), thyroid hormones, toddler, Antenatal Care (ANC)

Abbreviations: HIV - Human Immune Deficiency Virus, VDRL -Venereal Disease Research Laboratory Test, MUAC - Mid upper arm circumference, SD Standard Deviation, TSH- Thyroid stimulating Hormone), T4-Thyroxine

INTRODUCTION

Congenital hypothyroidism (CH) can result in significant repercussions for the brain development and growth of a child, potentially leading to enduring disabilities and cognitive impairments; however, early intervention can yield markedly improved outcomes[1]. The deficiency of thyroid hormone at birth is classified as CH. Through the implementation of Newborn Screening, CH can be effectively mitigated, thereby enhancing the quality-of-life[2]. Thyroid hormone plays an indispensable role in the intricate processes of neurodevelopment specifically in neurotransmission, growth, and energy metabolism[3]. The thyroid gland also plays a pivotal role in hematopoiesis, and blood disorders such as anemia, bicytopenia, or pancytopenia are frequently observed in individuals with thyroid dysfunction[8].

The occurrence of CH is estimated to be approximately 1 in 4,000 infants globally; this prevalence is significantly lower in Japan, where it is estimated at 1 in 5,500 infants, as well as among the African American

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community, where it is around 1 in 32,000 infants. A significant proportion of infants identified with CH are asymptomatic at birth, even in instances of complete agenesis of the thyroid gland[4]. However, in specific cases, during the initial months of existence, symptoms of hypothyroidism—such as difficulties in feeding,infrequent bowel movements, inadequate weight gain, a hoarse cry, and excessive drowsiness—are commonly observed[4].

The prompt identification of congenital hypothyroidism is primarily reliant on universal newborn screening[5]. This screening is typically conducted within the first 72 hours of life, during which the clinical identification of CH can prove challenging, if not entirely unfeasible. International guidelines advocate for the prompt initiation of treatment with levothyroxine (LT4) and emphasize the necessity of meticulous monitoring of hormonal levels to enable appropriate therapeutic adjustments[5].

The primary aim of this paper is to highlight the critical significance of newborn screening for a wide spectrum of congenital and inherited disorders, with CH being one of the essential conditions to be examined.

MATERIAL AND METHOD

Background: This paper presents a case study of a 15-month-old toddler who was admitted to the pediatric department at Tanga Regional Referral Hospital and subsequently diagnosed with Primary Congenital Hypothyroidism (CH). The toddler was hospitalized due to failure to thrive for over eight months, accompanied by delayed developmental milestones, as well as the presence of umbilical and inguinal hernias, along with a history of chronic constipation. Additionally, he exhibited symptoms of fever and cough persisting for approximately three days.Thoriugh physical examination and laboratory investigation was done to cnfirm the diagnosis.

Location: Tanga , Tanzania, East Africa, Africa.

Physical Examination: Pale, dyspneic, undersized for chronological age, with mottled, desiccated skin and a discernible murmur upon auscultation.

Test Performed: Full Blood count, Blood group, TSH, T4,Malaia tests, blood culture and sensitivity Echocardiogram, X-ray.

Ethical Consent: This case was documented with informed consent from from patient's guardians, ensuring compliance with ethical guidelines for case reporting.

Case Presentation

History

A male toddler, aged 15 months, was admitted to our health facility with a history of failure to thrive since the age of one month. He also presented with complaints of umbilical and inguinal swellings that had been evident since he was two months old. In addition to these long-standing issues, he exhibited persistent high-grade fever accompanied by a dry cough and rhinorrhea, as well as a whitish ocular discharge.

The infant was delivered at term via spontaneous vaginal delivery, demonstrating immediate respiratory effort and initiating breastfeeding within minutes of birth. Constipation commenced on the third day of life, initially deemed normal for a newborn; however, as it persisted, it became a significant concern. The infant was seen as an outpatient on multiple occasions due to this issue, typically having a solitary bowel movement every one to two days, often producing hard stools with considerable difficulty.

Despite being exclusively breastfed and receiving appropriate complementary feeding from the age of seven months, he exhibited inadequate weight gain since the age of one month. He experienced episodes of regurgitation when fed beyond his appetite, further contributing to poor feeding habits. His birth weight was recorded at 3.6 kg, but by four months of age, he had not doubled his birth weight, having gained only 1.3 kg. At 15 months, his weight was 4.9 kg, with a maximum weight of 5.9 kg attained.

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At the age of 15 months, the toddler is unable to control his neck and cannot sit, stand, or walk, indicating a significant delay in his developmental milestones. His immunizations and vaccinations are current, adhering to the Tanzanian immunization schedule.

During her pregnancy, the mother initiated antenatal care (ANC) at a gestational age of four months, completing a total of five visits. Throughout this period, she received hematinics, antihelminthic treatment, malarial prophylaxis, and two doses of the tetanus toxoid vaccine. Additionally, she was tested for HIV and VDRL, both of which yielded negative results. Her hemoglobin levels were within the normal range, she maintained euglycemia throughout her ANC visits, and her blood pressure remained normotensive. Although she was treated for a urinary tract infection, no investigations were conducted to assess for thyroid disorders during her pregnancy.

Physical Examination

Upon physical examination, the infant presented with severe pallor, without signs of jaundice, and exhibited macroglossia along with a notably large anterior fontanelle. He was dyspneic, with a respiratory rate of 48 breaths per minute and an oxygen saturation of 91% in room air.

He had anthropometric measurements of body weight of 4.9 kg, a length of 61 cm, and a mid-upper arm circumference (MUAC) of 11.2 cm. The length-for-age and weight-for-age assessments indicate -2 standard deviations, signifying that the toddler is experiencing severe acute on chronic malnutrition.

On examination of the abdomen, there was an everted umbilicus characterized by a soft, reducible umbilical swelling, which was non-tender and more pronounced during episodes of crying, accompanied by an abdominal wall defect in the umbilical region measuring approximately 3 cm. Additionally, a reducible left inguinal swelling was noted, which protruded upon coughing.

In the cardiovascular system, a grade 4 murmur was detected.

In the respiratory examination, bronchovesicular breath sounds were observed.

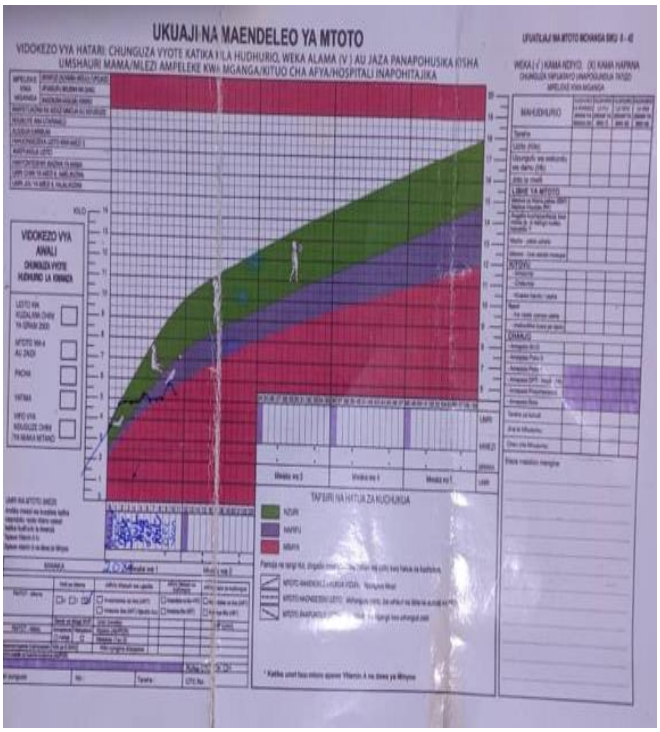

Figure 1: Baby clinic card which shows growth curve at the age of 10 months with weight of 5.2 kg

Figure 2: Umbilical and scrotal hernia and mottled and dry skin

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Figure 3: Large anterior fontanelle

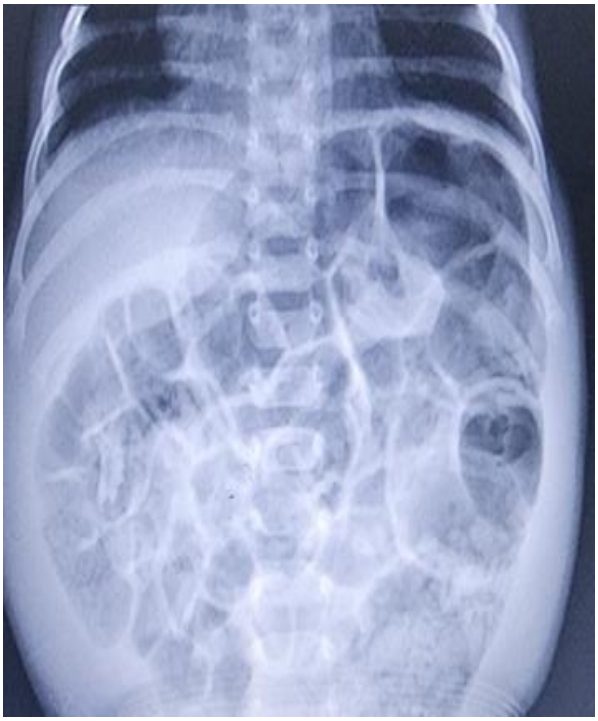

Figure 4: Plain abdominal X-ray

He underwent an investigation for congenital hypothyroidism (CH), during which thyroid-stimulating hormone (TSH) and thyroxine (T4) levels were assessed. The results indicated markedly elevated levels of TSH and diminished levels of T4, thereby substantiating the diagnosis of primary congenital hypothyroidism. A complete blood count revealed significantly low hemoglobin levels, confirming one of the hallmark signs of CH, namely pallor.

Additional diagnostic tests included an echocardiogram, which disclosed the presence of an atrial septal defect, as well as an abdominal-pelvic ultrasound that corroborated the diagnosis of umbilical and inguinal hernias. Renal function tests were found to be within normal limits. Diagnosis of CH with ASD, umbilical and inguinal hernia, severe acute on chronic malnutrition and severe anaemia was confirmed based on clinical findings and investigation done and also conjunctivitis.

Investigation Results

Table 1:

Full Blood Count:
WBC:12.2
ANC:4.91
ALC:5.65
AMC:1.2 elevated which could suggest infection since the machine can picks immature neutrophils as monocytes
Hb 6.47
MCV 79.3
MCH 26.5

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MCHC 32.1 PLT 401
Blood Group B positive
MRDT: Negative
Hormones TSH: >100.0(0.4-4.2 U/mL)- elevated markedly T4:<0.91(5.0-13.0 ug/dl)- low
Echocardiogram Results: Atrial Septal defect with left to right shunt
Abdominal Pelvic Ultrasound results: Umbilical hernia Left Inguinal scrotal hernia
Biochemistry Creatinine 64.1Umol/l (53-120) Urea 3.9 mmol/l(2.5-7.7)
Plain abdominal X-Ray Results (Figure 4) Showed features suggestive of dilated bowels

Management given

The infant was placed on oxygen therapy due to hypoxia and received a course of antibiotics for seven days, consisting of intravenous ampicillin at a dosage of 253 mg administered thrice daily, along with intravenous gentamicin at 14 mg daily. The patient was also evaluated by the surgical team. Additionally, he was provided with multivitamins, including 100,000 IU of vitamin A on days 1, 2, and 14, folic acid, therapeutic milk, and commenced on levothyroxine at a dosage of 50 µg daily. Upon discharge, he continued with levothyroxine at the same dosage and was scheduled for follow-up at the outpatient clinic for both congenital hypothyroidism and hernia.

DISCUSSION

Hypothyroidism arises from an insufficiency in the production of thyroid hormones or a dysfunction within the thyroid gland. It may originate from either congenital factors or be acquired later in life[4].The prevalence of CH is approximately 1 in 4,000 live births, with thyroid agenesis or dysgenesis accounting for 80% of cases, while dyshormonogenesis constitutes the remaining 20%. Thyroid gland abnormalities, including aplasia or hypoplasia, represent a significant etiology of congenital hypothyroidism, commonly referred to as thyroidal dysgenesis. Should hypothyroidism develop during the early stages of fetal life, its ramifications may manifest

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in several organ systems, notably the central nervous system and the skeletal structure[1]. However, it is important to note that most infants diagnosed with CH appear normal at the time of birth[4]. Diagnosing CH at birth based solely on clinical signs is infrequently achievable. The challenge of diagnosing CH arises from the non-specificity of its symptoms. Generally, distinct symptoms do not become evident in newborns, which accounts for the reliance on neonatal screening programs for the majority of diagnoses[4]. The specific clinical signs are prolonged jaundice, lethargy, feeding problems, constipation, hypothermia, mottling and dry skin, umbilical hernia, macro-glossia, anterior and posterior open fontanels, hoarse cry, and slow deep tendon reflexes[4,9]. The physical development is usually retarded, such as delayed sitting, standing, and talking ages. The degree of physical and mental retardation increases with the age of diagnosis and treatment. The child demonstrates significant impairment in growth[10].

In our case, the clinical manifestations included severe anemia, feeding difficulties resulting in substantial growth impairment and severe malnutrition, constipation, hypothermia, an open anterior fontanel with prominent scalp veins, a flattened nasal bridge, a short and thick neck, congenital heart disease, umbilical and inguinal hernias, shortened extremities and digits, and myxedema. This patient was unable to sit, walk, or articulate words by the age of 15 months.

Congenital heart conditions are observed in approximately 10 to 18.5% of patients with CH. The incidence of other congenital anomalies apart from CH appears to vary based on genetic and ethnic factors. Notably, three cases of cardiac defects associated with CH have been reported in Africa[6]. Our case contributes to the growing body of documented instances of cardiac anomalies in patients with CH.

The diagnosis of congenital hypothyroidism (CH) necessitates a comprehensive evaluation that includes blood assays, urine analyses for iodine levels, and various imaging modalities. In neonates, dried blood spot tests are conducted to assess thyroid-stimulating hormone (TSH) and thyroxine (T4) levels. In cases of primary CH, there is a notable reduction in T4 levels accompanied by an elevation in TSH. The presence of anti-thyroid antibodies corroborates the diagnosis of transient autoimmune thyroiditis. Iodine concentrations exceeding 200 µg/L indicate excessive exposure to iodine, which may inhibit thyroid receptor function. Imaging techniques, such as thyroid ultrasonography and scintigraphy, play a pivotal role in elucidating the etiology of CH[7].

CONCLUSION

It is imperative to establish a comprehensive policy for neonatal screening of congenital hypothyroidism (CH), despite its rarity, as well as other congenital anomalies, in order to avert disabilities in future generations where such screening is not routinely implemented. Furthermore, prompt intervention and meticulous follow-up care are crucial, particularly in cases of primary CH, as it may necessitate lifelong management. The thyroid gland plays a pivotal role in hematopoiesis, and blood disorders such as anemia, bicytopenia, or pancytopenia are frequently observed in individuals with thyroid dysfunction. Furthermore, appropriate management of thyroid disorders has been shown to ameliorate these cytopenias.

Various instances of congenital hypothyroidism accompanied by additional anomalies have been documented in Africa, particularly in Egypt, although it predominantly manifests in females.Hence, this case underscores the imperative for heightened vigilance among healthcare providers and caregivers to facilitate prompt diagnosis and lifelong support for affected individuals. Furthermore, amplifying screening initiatives and ensuring equitable access to care remain paramount priorities in the effective management of this condition.

Author contribution: E.M.B, K.G.M, M. W and S.S.I involved in obtaining history of the patient, performing physical exam and investigation of the patient. E.M. B was involved in writing original draft of the paper. All four were involved in writing, reviewing and proof reading.

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Consent: Written informed consent was procured from the mother of the infant for the composition and subsequent publication of this case report.

Conflicts of interest: The authors declare no conflict of interest

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