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Case Report

Technetium-99 thyroid scan as an aid to diagnosis of congenital hypothyroidism with normal initial thyroid function test

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ABSTRACT

Congenital hypothyroidism (CH) occurs in 1:2,000 to 1:4,000 newborns, is often asymptomatic at birth, due to transplacental passage of maternal thyroid hormones and some thyroid production by the infant. CH is classified into permanent and transient forms, thyroid dysgenesis accounts for 85% while dyshormonogenesis 10-15% cases. CH is diagnosed through newborn screening, confirmation by elevated serum thyroid stimulating hormone (TSH) and low T4 or free T4 levels. Thyroid radionuclide uptake, thyroid sonography, serum thyroglobulin, help identify the underlying cause, although treatment may begin prior to these tests. Early diagnosis and treatment are crucial for preventing developmental delays and ensuring optimal growth and development in affected infants.

Keywords: Congenital hypothyroidism, Thyroid dysgenesis, Thyroid aplasia, Scintigraphy 99mTc

INTRODUCTION

Congenital hypothyroidism is most commonly due to issues with the development of the thyroid gland, collectively known as thyroid dysgenesis. This includes complete absence (agenesis), underdevelopment (hypoplasia), or abnormal positioning (ectopy) of the gland. Thyroid dysgenesis is responsible for approximately 80–85% of permanent congenital hypothyroidism cases.[1,2] Another cause is dyshormonogenesis, involving defects in thyroid hormone production.

Despite the severity of the condition, most newborns with congenital hypothyroidism— even those lacking a thyroid gland entirely—show no symptoms at birth.[3,4] This is largely due to the transplacental transfer of maternal thyroxine (T4), which helps maintain fetal thyroid hormone levels at roughly one-third of normal at birth.[3,5,6] Measurements in umbilical cord blood indicate that maternal T4 contributes between 25–50% of the newborn's circulating T4 levels, offering neuroprotective benefits during the perinatal period.[7,8]

Additionally, many cases of congenital hypothyroidism involve some residual or partially functioning thyroid tissue, which may mask early clinical signs.[9] Early diagnosis and treatment are crucial, as there is a well-established inverse correlation between the age at which treatment begins and future cognitive outcomes—delayed intervention is associated with reduced IQ scores.

The condition is more prevalent in females, with a reported female-to-male ratio of about 2:1.[10] While most cases of thyroid dysgenesis occur sporadically, inherited genetic mutations are identified in less than 5% of patients.

We report a case of congenital hypothyroidism due to complete absence of the thyroid gland (athyreosis), which initially presented with nearly normal thyroid function tests. Diagnosis was confirmed using Technetium-99m (99mTc) thyroid scintigraphy at three months of age.

CASE REPORT

A 2-month-old male baby born from a non-consanguineous marriage presented with poor feeding, regurgitation of feeds, hypotonia, lethargy, intermittent

cyanotic episodes, no social smile, no vocalization. Antenatal and family history were not significant. Birth history: term/3.25 kg/appropriate for gestational age/ LSCS/MSL/not cried immediately/ mild respiratory distress requiring hood box oxygen for 3 days/lethargic at discharge and could be spoon fed. H/O phototherapy for neonatal jaundice (TSB 19.8/mg/dl) on day 16 of life. Magnetic resonance imaging (MRI) brain (day 19) showed bilateral symmetrical T1 hyperintensity involving globus pallidus, subthalamic nuclei, substantia nigra and dentate nuclei likely s/o of neonatal hyperbilirubinemia. Thyroid function test (day 16) - normal. Newborn screening for inborn error of metabolism (extended panel), SMN gene test for SMA mutation detection, Ach receptor antibody for congenital myasthenia, 2D-ECHO, electroencephalography (EEG) were normal. Given the symptoms and normal initial workup, the infant was considered a case of gastroesophageal reflux disease managed conservatively and discharged on post-pyloric oro-gastric feeding. On follow-up (2-6 weeks after discharge), the infant showed weight gain but continued to exhibit hypotonia and delayed developmental milestones (did not follow objects, no head control, no social smile). BERA test was normal at 3.5 months.

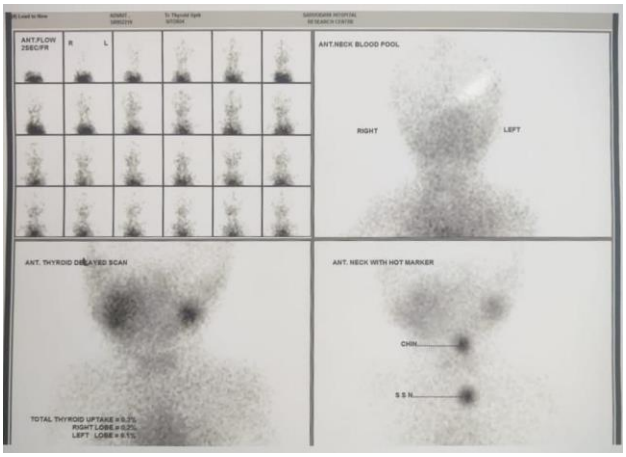

Figure 1: Techtenium 99 scan showing thyroid dysgenesis.

Table 1: Age specific thyroid profile.

Age	FT3 (pg/ml)	FT4 (ng/dl)	TSH (ulU/ml)
16 days	3.04	1.44	5.592
2 months	3.75	0.98	3.7
4 months	3.89	0.90	2.7
After levothyroxine supplementation			
After 4 weeks (5 months)	4.76	1.92	0.072
After 12 weeks (7 months)	5.04	1.34	0.541

A repeat thyroid function test at 4 months revealed FT3- 3.89 pg/ml FT4- 0.9 ng/dl and TSH 2.7 uIU/ml- a low normal FT4 prompted further investigation. A thyroid scan (Scintigraphy 99mTc) was performed (4 months) which revealed no functioning thyroid tissue in the neck or elsewhere, indicating thyroid dysgenesis. Levothyroxine supplementation (37.5 mcg/day) started after thyroid scan. Follow-up at 9.5 months; head control achieved, sits with support, follows objects till 180 degrees, social smile present, and occasional vocalization are vital for optimizing neurodevelopmental outcomes.

DISCUSSION

Thyroid dysgenesis or agenesis is usually sporadic, with no clearly established genetic or environmental risk factors. Clinical presentation can vary widely, from mild developmental delays to more pronounced symptoms such as hypotonia, feeding challenges, and episodes of cyanosis, similar to what was observed in this case. The introduction of newborn screening programs has markedly improved outcomes by enabling early initiation of thyroid hormone therapy in affected infants.

Long-term management of CH typically requires lifelong thyroid hormone replacement, with consistent monitoring of thyroid levels to support normal growth and cognitive development. Among diagnostic tools, radionuclide scanning remains one of the most precise methods for identifying structural abnormalities in the thyroid gland. These scans can detect conditions such as ectopic thyroid tissue, hypoplasia (identified by decreased uptake in a normally positioned gland), or complete gland absence (aplasia).[11]

Although imaging findings can help clarify the etiology of hypothyroidism and may support treatment decisions in cases with borderline biochemical results, they often do not alter the overall management plan and are therefore considered optional in some clinical protocols.

Thyroid dysgenesis is typically classified into three main subtypes: ectopy, athyreosis, and hypoplasia. Ectopic thyroid tissue found along the migration path of the thyroid from the tongue base to the neck is the most common form, accounting for approximately two-thirds of cases and occurring more frequently in females.[12,13] Athyreosis and hypoplasia make up the remaining third. Imaging can also help differentiate between transient and permanent hypothyroidism, providing valuable information for long-term treatment planning, especially in healthcare settings where imaging is a routine part of the diagnostic workup.

CONCLUSION

Despite normal initial assessments including newborn screening and thyroid function tests persistent symptoms prompted further evaluation, ultimately revealing thyroid dysgenesis. This case emphasizes the importance of early identification and treatment in congenital hypothyroidism (CH) to ensure optimal neurodevelopment. Clinicians should remain vigilant for signs of thyroid dysfunction, even when early test results appear within normal limits,

as delayed diagnosis can lead to irreversible developmental consequences. Ongoing follow-up and prompt intervention are essential components of effective CH management.

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