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A Severe Case of Untreated Hypothyroidism

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Introduction

Since the implementation of newborn screening programs in the mid-1970s, most infants in countries with this program are diagnosed and treated for congenital hypothyroidism, preventing neurodevelopmental impairment [1]. However, there can sometimes be barriers to accessing subspecialty care leading to delays in treatment. This case highlights the unfortunate outcomes that can occur in untreated congenital hypothyroidism.

Case Presentation

A 12-year-old female from Guatemala who moved to Dallas, TX fifteen days prior presented to the emergency department. Despite having been diagnosed with congenital hypothyroidism on newborn screen in Guatemala, she was unable to access subspecialty care and was never started on thyroid hormone replacement.

Physical Exam:
Height: 86 cm (<0.01%ile, Z = -9.30)
Weight: 14.6 kg (<0.01 %ile Z= -9.79)
General: significant short stature, global developmental delay, speech delay
HEENT: swelling of face, protruding tongue, no palpable abnormalities of thyroid bed
MSK: marked swelling of face, neck. Bilateral clubbed feet, spastic left hip
GU: pre pubertal female. Tanner 1 breasts

Laboratory and Imaging

Figure 1: Physical exam findings of patient

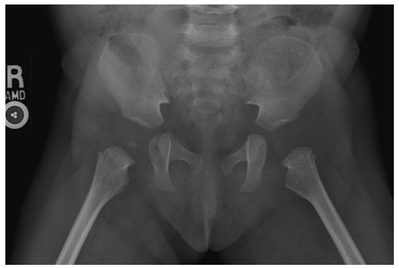

Figure 2: Radiograph of pelvis demonstrates delayed ossification of femoral heads

Figure 3: Ultrasound thyroid and neck shows no thyroid tissue identified in the neck

Laboratory and Imaging

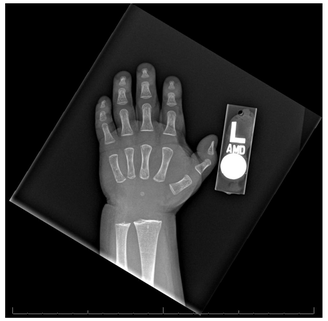

Figure 4: Severely delayed bone age of 3 months (chronological age 12 years 10 months)

Initial Laboratory values at time of presentation

Component	Latest Ref Range & Units	
TSH	0.400 - 5.340 microIU/mL	>150.000 (H)
Free T4	0.89 - 1.76 ng/dL	0.21 (L)
T3, Total	30.00 - 294.00 ng/dL	<30.00 (L)

Laboratory values following treatment

	Oct-22	Nov-22	Jan-23
TSH	> 150	> 150	47.53
Free T4	0.4	0.78	0.89

Started on Synthroid 12.5 mcg daily
Increase to 25 mcg daily
Increase to 37.5 mcg daily

Hospital Course

This patient was admitted to the general pediatrics service for inpatient management. For her significant developmental delays, physical therapy, occupational therapy, and speech therapy evaluated patient and advised follow up outpatient. Given the delayed ossification of pelvis and hips, orthopedics was consulted and advised outpatient follow-up. Treatment with Synthroid 12.5 mcg was started. Repeat labs as outpatient showed mild improvement in free T4, dose was then increased to 25 mcg, and then 37.5 mcg daily.
At recent clinic visit, noted to have height gain of 3.5 cm and improvement in energy level, hair loss, and constipation.

Conclusions

Complications of untreated hypothyroidism can result in various complications such as neurodevelopmental delays, accumulation of glycosaminoglycans resulting in stiffness, muscular pseudohypertrophy, proximal myopathy, and static encephalopathy. This case highlights the importance for health care providers to recognize the phenotype of untreated congenital hypothyroidism.

Bibliography

1.Ford G, LaFranchi SH. Screening for congenital hypothyroidism: a worldwide view of strategies. Best Pract Res Clin Endocrinol Metab. 2014;28(2):175-187. doi:10.1016/j.beem.2013.05.008