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Congenital Hypothyroidism Diagnosed at 10 Months of Age in an Infant Born of Consanguineous Parents: A Case Report

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Abstract **Case Report**

Background: Congenital hypothyroidism (CH) is the most common preventable cause of intellectual disability in children worldwide. Although universal neonatal biochemical screening allows detection within the first weeks of life in most high-income settings, the diagnosis can still be substantially delayed where such screening is unavailable. ***Case presentation:*** We report the case of a male infant, the only child of first-degree consanguineous parents, referred at 10 months of age for growth failure. Examination revealed marked hypotonia, pallor, macroglossia, and severe growth retardation (weight −2 SD, length −4 SD, head circumference −2 SD). Biological work-up showed a markedly elevated thyroid-stimulating hormone (TSH 199 µIU/mL) with low circulating thyroid hormones, negative anti-thyroperoxidase antibodies, delayed bone age, and a homogeneous goiter with regular contours on thyroid ultrasound. Levothyroxine replacement, initiated at 50 µg/m²/day and increased to 75 µg/m²/day, produced good clinical evolution. ***Conclusion:*** This case illustrates a typical late presentation of primary CH most consistent with thyroid dyshormonogenesis, favoured by parental consanguinity, and highlights the continued burden of delayed diagnosis in regions without systematic newborn screening.

Keywords: Congenital hypothyroidism; thyroid dyshormonogenesis; consanguinity; goiter; levothyroxine; delayed diagnosis.

1. INTRODUCTION

Congenital hypothyroidism (CH) is defined as a deficiency of thyroid hormone production present at birth and is the most common preventable cause of intellectual disability in children worldwide [1]. It results from dysfunction anywhere along the hypothalamic–pituitary–thyroid axis, leading to insufficient production of triiodothyronine (T3) and thyroxine (T4) [1,2]. The reported incidence ranges from approximately 1 in 2,000 to 1 in 4,000 live births, with higher rates described in populations with elevated rates of parental consanguinity [2].

Two main mechanisms underlie primary CH (Table 1). Thyroid dysgenesis agenesis, ectopy, or hypoplasia of the gland accounts for approximately 85% of cases and is usually sporadic. The remaining 10–15% result from dyshormonogenesis, a group of inherited, typically autosomal-recessive defects affecting one of the sequential steps of thyroid hormone biosynthesis (iodide trapping, organification, coupling, or thyroglobulin synthesis); this mechanism is disproportionately represented among children born to consanguineous parents [3,4].

Table 1: Principal etiological mechanisms of primary congenital hypothyroidism [3,4]

Thyroid dysgenesis (~85%)	Thyroid dyshormonogenesis (~15%)
Absent, ectopic, or hypoplastic gland; usually sporadic	Gland present but non-functional; often hereditary (autosomal recessive); favoured by consanguinity

Because CH is frequently clinically silent at birth, systematic neonatal biochemical screening (heel-prick TSH/T4) is essential for early detection, and timely initiation of levothyroxine remains the single most important determinant of long-term neurodevelopmental outcome [1,6]. In many low- and middle-income

countries, however, universal newborn screening has not yet been implemented, and the diagnosis continues to rely on clinical recognition, which is frequently delayed [6,7]. We report a case of CH diagnosed only at 10 months of age in an infant born to first-degree consanguineous parents, illustrating the clinical, biological, and therapeutic features of this delayed presentation.

2. CASE PRESENTATION

2.1 History

The patient, a male infant and the only child of a first-degree consanguineous union, was referred to our pediatric endocrinology and diabetology unit at 10 months of age for evaluation of growth failure (poor weight and height gain).

2.2 Clinical Examination

On examination, the infant was hypotonic, apathetic, and pale, with marked growth retardation: weight 6 kg (−2 SD), length 60 cm (−4 SD), and head circumference 43 cm (−2 SD). Additional findings included macroglossia and a distended abdomen without palpable mass or organomegaly; the hernial orifices were free.

Figure 1: Clinical photograph at presentation (10 months) showing axial hypotonia, pallor, and a depressed/listless facies. The periocular region has been digitally masked to protect patient identity

Figure 2: Clinical photograph at presentation showing macroglossia and coarse facial features typical of untreated congenital hypothyroidism. The periocular region has been digitally masked to protect patient identity

2.3 Laboratory and Imaging Findings

Table 2: Summary of laboratory and imaging findings at presentation

Parameter	Result
Complete blood count	Normochromic, normocytic anemia
TSH	199 µIU/mL (markedly elevated)
Free T4	0.7 (severely reduced; units as recorded in the original chart)
T3	1.75 (reduced; units as recorded in the original chart)
Anti-thyroperoxidase (anti-TPO) antibodies	Negative
Bone age	Delayed by ~4 months relative to chronological age (10 months)
Thyroid ultrasound	Homogeneous goiter with regular contours, gland in normal (eutopic) position

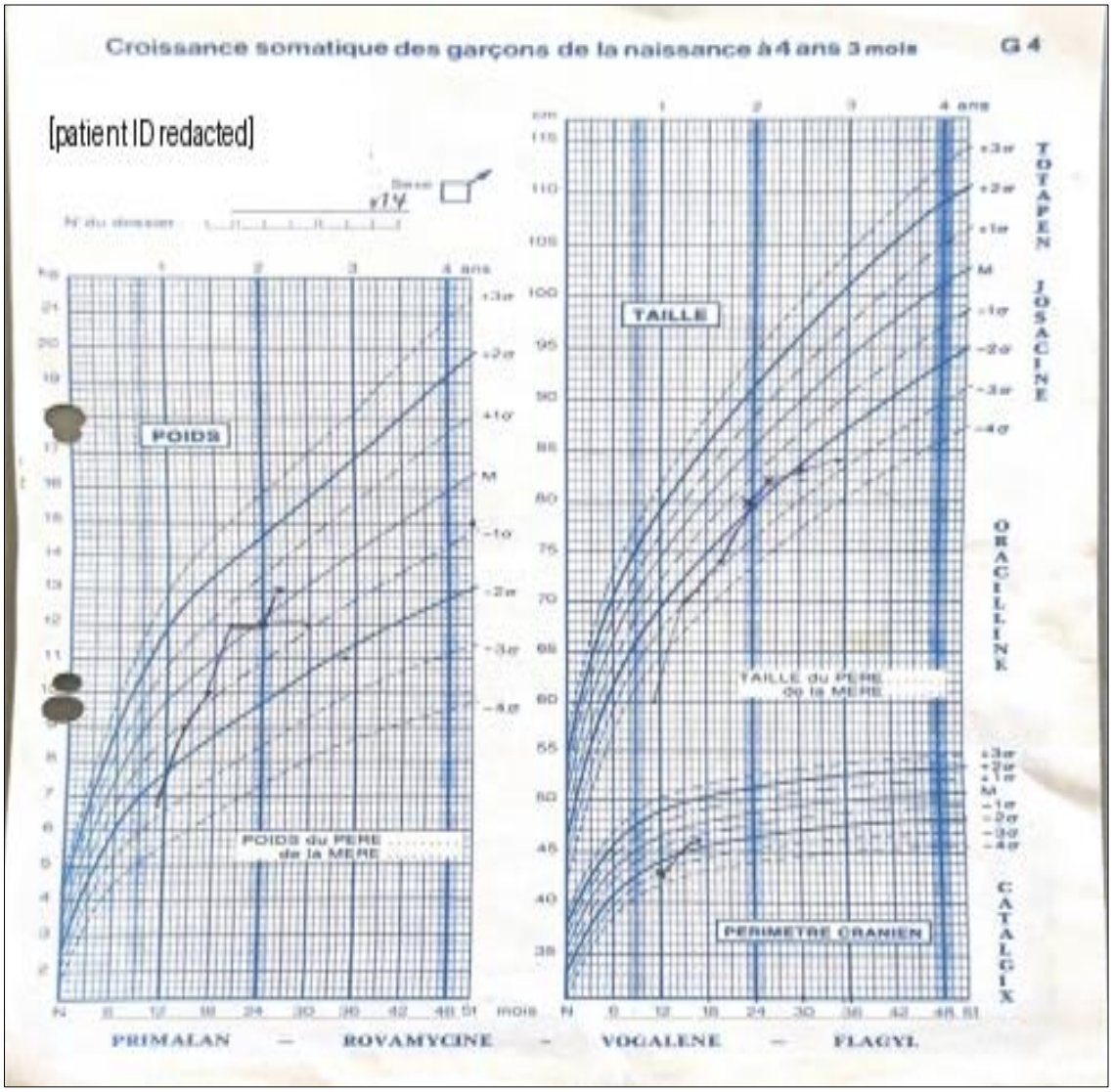

Figure 3: Somatic growth chart (weight, height, head circumference) for boys aged 0–4 years, annotated at the time of presentation, illustrating severe growth retardation (weight −2 SD, length −4 SD, head circumference −2 SD). Patient identifying information has been redacted

2.4 Treatment and Outcome

Levothyroxine (Levothyrox®) replacement was initiated at 50 µg/m²/day and subsequently increased to 75 µg/m²/day. The patient showed good clinical evolution on regular follow-up.

3. DISCUSSION

This child's presentation is a typical illustration of severe, late-presenting primary CH. The triad of hypotonia, major growth retardation, and goiter, together with a strikingly elevated TSH (199 µIU/mL) and profoundly reduced circulating thyroid hormones, is

highly suggestive of the diagnosis [1,2]. The negative anti-TPO titer argues against an autoimmune or transplacentally-transmitted blocking-antibody mechanism and instead points toward non-autoimmune dyshormonogenesis, a possibility reinforced by the first-degree parental consanguinity [3,4]. Population-based studies have confirmed that parental consanguinity significantly increases the risk of both permanent and transient CH, presumably by increasing the likelihood of homozygosity for recessive mutations in genes governing thyroid hormone synthesis, such as TPO, TG, SLC5A5 (NIS), SLC26A4 (PDS), DUOX2, and DUOXA2 [4,5].

The presence of a homogeneous goiter with regular contours on ultrasound, rather than thyroid agenesis or an ectopic gland, further supports dyshormonogenesis over dysgenesis as the underlying mechanism in this infant: an enzymatically blocked but anatomically present, chronically TSH-stimulated gland typically hypertrophies, whereas dysgenetic glands are absent, hypoplastic, or ectopic [3,4].

This case also underscores the burden of delayed diagnosis in settings without universal neonatal screening. Diagnosis at 10 months of age well beyond the first weeks of life during which thyroid hormone is essential for normal brain development places the child at substantially higher risk of irreversible neurodevelopmental impairment, even after biochemical control is achieved with levothyroxine [6,7]. Comparable experience from Algeria, a country without a nationwide CH screening program, found a median age at clinical diagnosis of 1.6 months, yet with a meaningful proportion of children still diagnosed after one year of age and an inverse correlation between age at diagnosis and later IQ [7]. In countries where universal screening has likewise not been adopted, cases have been described that escape diagnosis even further, into adolescence or adulthood, because the cardinal clinical features of CH short stature, coarse facies, macroglossia, abdominal distension, and developmental delay can be subtle and easily attributed to other causes when clinical suspicion is low [8]. Longitudinal data confirm that both the physical and cognitive trajectories of children with CH are closely tied to the timeliness of diagnosis and the adequacy of subsequent levothyroxine replacement [9].

Taken together, this observation reinforces (i) the importance of maintaining a high index of clinical suspicion for CH in infants presenting with unexplained growth failure, hypotonia, macroglossia, or developmental delay, particularly in regions without systematic neonatal screening, and (ii) the case for wider implementation of universal newborn TSH/T4 screening programs, which remain the most effective intervention for preventing the neurodevelopmental sequelae of this otherwise readily treatable disorder [1,6].

4. CONCLUSION

Congenital hypothyroidism most often results from a developmental anomaly of the thyroid gland (complete or partial dysgenesis) or from a molecular defect impairing hormone synthesis (dyshormonogenesis), the latter being more likely in the setting of parental consanguinity, as in the infant described here. Early recognition and prompt initiation of levothyroxine replacement ideally guided by neonatal screening allow normal physical and psychomotor development; delayed diagnosis, as illustrated in this case, exposes the child to a substantially higher risk of irreversible neurodevelopmental sequelae.

Declarations

Patient consent:

Written informed consent for the use and publication of clinical images and case details should be obtained from the patient's legal guardian(s) in accordance with journal policy prior to submission. Identifying details, including the face and recorded name/date of birth, have been digitally masked or redacted in the accompanying figures to protect patient confidentiality.

Conflict of interest: The authors declare no conflict of interest.

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REFERENCES

1. van Trotsenburg P, Stoupa A, Léger J, Rohrer TR, Peters C, Fugazzola L, *et al.,* Congenital hypothyroidism: a 2020–2021 consensus guidelines update an ENDO-European Reference Network initiative endorsed by the European Society for Pediatric Endocrinology and the European Society for Endocrinology. Thyroid. 2021;31(3):387–419.
2. Peters C, van Trotsenburg ASP, Schoenmakers N. Diagnosis of endocrine disease: congenital hypothyroidism update and perspectives. Eur J Endocrinol. 2018;179(6): R297–R317.
3. Rastogi MV, LaFranchi SH. Congenital hypothyroidism. Orphanet J Rare Dis. 2010; 5:17.
4. Grasberger H, Refetoff S. Genetic causes of congenital hypothyroidism due to dyshormonogenesis. Curr Opin Pediatr. 2011;23(4):421–428.
5. Abbasi F, Janani L, Talebi M, Azizi H, Hagiri L, Rimaz S. Risk factors for transient and permanent congenital hypothyroidism: a population-based case-control study. Thyroid Res. 2021; 14:11.
6. Ford G, LaFranchi SH. Screening for congenital hypothyroidism: a worldwide view of strategies. Best Pract Res Clin Endocrinol Metab. 2014;28(2):175–187.
7. Djermane A, Ouarezki Y, Boulesnane K, Kherra S, Bouferoua F, Bessahraoui M, *et al.,* The burden of congenital hypothyroidism without newborn

screening: clinical and cognitive findings from a multicenter study in Algeria. Int J Neonatal Screen. 2025;11(3):78.

8. Alam A, Ashraf H, Khan K, *et al.,* Uncovering congenital hypothyroidism in adulthood: a case study emphasizing the urgency of screening and clinical awareness. Cureus. 2023;15(9): e45611.
9. Nazari J, Jafari K, Chegini M, Maleki A, MirShafiei P, Alimohammadi A, *et al.,* Physical and mental growth and development in children with congenital hypothyroidism: a case-control study. Orphanet J Rare Dis. 2021; 16:404.