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A Novel Homozygous Thyroglobulin Gene Variant Presenting with Massive Congenital Goiter and Neonatal Airway Obstruction

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Abstract

Congenital hypothyroidism due to thyroid dyshormonogenesis is a rare inherited disorder that may present with goiter of variable severity. Massive congenital goiter causing life-threatening respiratory distress at birth is exceptionally uncommon. We report a late-preterm male neonate who presented immediately after delivery with severe respiratory failure requiring endotracheal intubation due to critical tracheal compression from a massive congenital goiter. Postnatal imaging revealed marked thyroid hyperplasia with airway narrowing. Thyroid function tests demonstrated isolated low free thyroxine levels with normal thyroid-stimulating hormone (TSH), a pattern attributed to non-thyroidal illness syndrome related to perinatal asphyxia and inotropic support. Therapeutic-dose levothyroxine was initiated, resulting in a progressive reduction in goiter size and successful extubation. Genetic analysis using next-generation sequencing identified a previously unreported homozygous missense variant in the thyroglobulin gene, supporting the diagnosis of thyroglobulin-related dyshormonogenetic congenital hypothyroidism. This case highlights that congenital goiter should be considered in neonates presenting with respiratory distress at birth, even in the absence of elevated TSH levels or prenatal findings. It underscores the importance of early diagnosis, genetic evaluation, and timely levothyroxine therapy to prevent airway compromise and ensure favorable outcomes.

Introduction

Congenital hypothyroidism (CH) is the most common endocrine disorder of the neonatal period, with an estimated incidence of approximately 1 in 1500 live births [1]. The condition shows a clear female predominance, with a female-to-male ratio of nearly 2:1, and remains one of the leading causes of preventable intellectual disability worldwide if not diagnosed and treated promptly [2].

The most frequent etiology of primary CH is thyroid dysgenesis, including agenesis, ectopia, and hemiagenesis. In the remaining cases, the thyroid gland is normally located, and fewer than half are attributed to thyroid dyshormonogenesis (TDH), a group of inherited disorders caused by variants in genes involved in thyroid hormone biosynthesis. TDH is typically inherited in an autosomal recessive manner, with pathogenic variants in the thyroid peroxidase (*TPO*) gene representing the most common genetic cause [3]. Congenital TDH due to thyroglobulin (*TG*) gene defects is considerably rarer, with an estimated incidence of approximately 1 in 100000 live births. To date, more than 100 distinct pathogenic variants in the *TG* gene have been reported [4]. Defects in the *TG* gene are frequently associated with goiter, which may develop during fetal life or the neonatal period; the clinical phenotype is highly variable, ranging from mild hypothyroidism to severe forms complicated by airway compromise [5].

The differential diagnosis of a congenital neck mass is broad and includes hemangioma or lymphangioma, teratoma, hamartoma, cystic hygroma, ectopic thymus, branchial cleft cysts, thyroglossal duct cyst, cervical neuroblastoma, meningoceles, and congenital goiter [6]. Most congenital goiters are acquired, commonly related to intrauterine iodine imbalance, maternal antithyroid drug

exposure, or transplacental autoimmune mechanisms. TDH-associated congenital goiter is an uncommon presentation [4].

Here, we report a newborn with genetically confirmed congenital goiter who presented with severe respiratory failure and perinatal asphyxia secondary to critical tracheal compression. Molecular analysis revealed a previously unreported homozygous *TG* gene variant, thereby expanding the mutational spectrum of dyshormonogenetic congenital hypothyroidism.

Case Report

A male infant was delivered at 36 weeks' gestation to a 25-year-old healthy mother by emergency cesarean section due to polyhydramnios and breech presentation. The birth weight was 2570 g (-1.84 SD), length 48 cm (-0.91 SD), and head circumference 32 cm (-2.08 SD). A large anterior neck mass was noted immediately after birth. The infant developed severe respiratory distress shortly after delivery and required endotracheal intubation in the delivery room for airway stabilization. The pregnancy had been regularly followed, and no cervical mass or thyroid abnormality had been identified on detailed prenatal ultrasonographic examinations. The parents were second-degree cousins, and there was no known family history of thyroid disease or congenital anomalies.

Upon admission to the neonatal intensive care unit, the infant's general condition was poor. He was tachypneic, tachycardic, hypoxic, and hypotensive. Physical examination revealed a large cervical mass without any additional congenital anomalies (Fig. 1a). Based on clinical findings consistent with moderate hypoxic-ischemic encephalopathy, therapeutic hypothermia was initiated at the 5th postnatal hour. Owing to persistent hypotension, dobutamine infusion was commenced. Following rewarming, an extubation attempt was made; however, it was unsuccessful due to significant upper airway compression caused by the cervical mass.

On postnatal day 7, neck magnetic resonance imaging (MRI) demonstrated a markedly enlarged thyroid gland consistent with diffuse hyperplasia. The left thyroid lobe measured 26×26×36 mm, the right lobe 27×28×44 mm, and the isthmus thickness was 14 mm (Fig. 2a). The enlarged gland caused mild narrowing of the upper airway at the tracheal level (Fig. 2b).

Thyroid function tests (TFTs) obtained on postnatal day 8 revealed a low free thyroxine (fT4) level of 0.46 ng/dL (0.70–1.48), with free triiodothyronine (fT3) at 3.73 pg/mL (1.58–3.91), thyroid-stimulating hormone (TSH) at 1.93 μIU/mL (0.35–4.94), and TG at 0.45 ng/mL (3.68–64.15). Skeletal radiography demonstrated normal ossification centers in the distal femur and proximal tibia, consistent with appropriate bone maturation for gestational age. Maternal TFTs were within normal limits, and thyroid autoantibodies, including anti-TPO, anti-TG, thyroid-stimulating immunoglobulin, and TSH receptor-blocking antibody, were all negative in both the mother and the infant. In addition, the mother's thyroid function profile during the first trimester of pregnancy had been normal.

Oral levothyroxine at a dose of 15 μg/kg/day was initiated. The initial newborn screening revealed a mildly elevated TSH level of 7.88 μIU/mL, which subsequently normalized on repeat sampling. Given the presence of a massive congenital goiter with markedly low fT4 and TG levels, thyroid dyshormonogenesis was strongly suspected. Accordingly, a clinical exome sequencing panel targeting genes associated with thyroid hormone biosynthesis disorders was performed.

The infant was unable to tolerate extubation on postnatal day 13; however, successful extubation was achieved on day 24, and the patient was breathing room air by day 30. A visible reduction in the size of the thyroid gland was noted during follow-up. Echocardiographic evaluation revealed a mid-muscular ventricular septal defect. Abdominal and cranial imaging studies, electroencephalography, and newborn hearing screening were all normal. Levothyroxine dosage was subsequently adjusted based on biweekly TFT results.

Serial ultrasonography demonstrated a progressive reduction in thyroid gland size, with the final ultrasound showing right lobe dimensions of 28×20×17 mm, left lobe dimensions of 30×16×16 mm, and an isthmus thickness of 5 mm, accompanied by mildly increased vascularity. The infant was exclusively breastfed, exhibited appropriate weight gain, and was discharged on postnatal day 47 with a body weight of 3650 g while continuing levothyroxine therapy.

Genetic analysis performed using next-generation sequencing (NGS) identified a homozygous missense variant in the *TG* gene (NM_003235.5:c.847C > T; p.Arg283Trp), which was classified as a variant of uncertain significance (VUS) according to the American College of Medical Genetics and Genomics (ACMG) criteria [7] (Fig. 3). This variant has not been previously reported in the literature or population databases.

In the context of a massive congenital goiter, markedly reduced fT4 levels, and the exclusion of maternal autoimmune thyroid disease, this homozygous *TG* variant was considered the most likely genetic cause of the observed phenotype. The concordance between the biochemical findings and the genetic result supported the diagnosis of thyroglobulin-related dyshormonogenetic congenital hypothyroidism and suggests that this variant may represent a novel pathogenic alteration contributing to thyroid hormone biosynthesis defects.

At 2 months of age, while receiving oral levothyroxine at a dose of 3.6 μg/kg/day, the infant weighed 5100 g (− 1.48 SD), with a length of 59 cm (− 0.85 SD), head circumference of 38 cm (− 2.29 SD), and body mass index 14.65 kg/m² (− 1.47 SD). TFTs showed a TSH level of 0.48 μIU/mL (0.35–4.94), and an fT4 level of 1.16 ng/dL (0.70–1.48), consistent with adequate hormonal control. Physical examination confirmed complete resolution of the cervical mass (Fig. 1b), and thyroid ultrasonography demonstrated a marked reduction in thyroid volume (right lobe 35×20×14 mm, 4.8 mL; left lobe 30×16×12 mm, 2.9 mL; isthmus 5 mm; total volume 7.7 mL, 24.1 SDS) (Fig. 2c). The patient was clinically euthyroid with age-appropriate growth and neurodevelopment. Written informed consent for publication of this case report and accompanying images was obtained from the patient's legal guardian.

Discussion

Massive congenital goiter is an exceptionally rare yet potentially life-threatening cause of neonatal respiratory distress. This case underscores the importance of including congenital goiter associated with thyroid dyshormonogenesis resulting from a novel homozygous variant in the *TG* gene in the differential diagnosis of cervical masses detected at birth, even in the absence of prenatal imaging abnormalities or elevated TSH levels. Massive congenital goiter causing respiratory distress at birth is extremely rare, with only a limited number of cases reported in the literature [6]. In most previously published reports, exhibited milder degrees of airway compromise or were diagnosed antenatally by prenatal ultrasonography, allowing for planned perinatal management [5, 8].

Dyshormonogenesis may present with goiter during the neonatal period or later in life and is only rarely associated with fetal goiter. The mechanical consequences of fetal goiter may include polyhydramnios due to esophageal compression, malpresentation resulting from excessive neck extension at delivery, and respiratory failure resulting from tracheal obstruction [9, 10]. In the present case, the coexistence of polyhydramnios and severe respiratory distress at birth suggests that the goiter had already developed in utero.

When fetal thyroid abnormalities are present, transplacentally transferred maternal thyroid hormones are generally sufficient to preserve normal fetal neurodevelopment [11, 12]. However, if both the mother and the fetus are hypothyroid, neurodevelopmental impairment may occur despite the initiation of levothyroxine immediately after birth [13]. Because maternal thyroid hormones provide partial protection for infants with CH for approximately the first two weeks of life, only 1–4% of affected newborns are clinically recognized at birth [14].

In the present case, although fetal goiter was retrospectively suspected, the mother was euthyroid, suggesting that transplacentally transferred maternal thyroid hormones likely protected fetal neurodevelopment. Despite being born with perinatal asphyxia secondary to mechanical airway compression, brain magnetic resonance imaging was normal, and no clinical or electrographic seizures were observed during follow-up. In addition, the infant did not exhibit overt clinical signs of hypothyroidism in the early postnatal period. Neck MRI performed to evaluate cervical swelling demonstrated thyroid hyperplasia, and TFTs revealed isolated low fT4 levels. These findings enabled an early diagnosis of CH due to dyshormonogenesis, which was established before the availability of routine newborn screening results.

In CH due to *TG* gene defects, typical laboratory findings include elevated TSH, low fT4, low or normal fT3, markedly reduced serum TG, and goiter present at or soon after birth [15]. In our patient, a massive goiter was present at birth, with low fT4, low TG, normal fT3, and TSH within the normal range. Non-thyroidal illness syndrome (NTIS) is characterized by abnormal TFTs in the absence of primary thyroid disease and may be triggered by severe systemic illness or the use of medications such as dopaminergic agents or corticosteroids. In NTIS, TSH levels are typically normal or suppressed, and thyroid function parameters generally normalize within 24 hours to 15 days after resolution of the acute

illness [16, 17]. In this patient, despite the presence of CH, we hypothesize that severe systemic stress related to perinatal asphyxia, together with dobutamine therapy, blunted the expected postnatal TSH surge, resulting in a biochemical pattern consistent with transient central hypothyroidism secondary to NTIS. Given the persistently low fT4 levels during the critical illness period, along with neck MRI and thyroid ultrasonography findings demonstrating thyroid hyperplasia, levothyroxine therapy was initiated without delay. This interpretation is further supported by follow-up TFTs obtained after clinical stabilization, showing fT4 and fT3 levels near the upper reference range without suppressed TSH despite ongoing levothyroxine therapy.

Recently published normative data from healthy term newborns in Aydın, an iodine-sufficient region, report mean thyroid volumes of 0.66 ± 0.25 mL at 0–2 days of life and 1.12 ± 0.33 mL at 15–30 days [18]. Although MRI rather than ultrasonography was used in our patient on postnatal day 7, the thyroid measurements were substantially larger than the expected upper limits for this population. At approximately 2 months of age, the thyroid volume on follow-up ultrasonography remained markedly enlarged for age; even when cautiously compared with neonatal reference values that extend only to 30 days of life, the gland size was still clearly above the reported upper limits, indicating persistent substantial goiter.

The primary objectives of genetic testing in CH are to refine diagnosis, guide treatment strategies, and improve prognostic assessment. Such testing is particularly recommended in patients with suspected thyroid dyshormonogenesis or familial forms of thyroid dysgenesis. Genetic analysis facilitates the identification of potential syndromic associations related to the detected variant, helps determine whether hypothyroidism is likely to be transient or permanent, and enables accurate assessment of recurrence risk in future pregnancies based on inheritance patterns and parental segregation analysis. For these purposes, genetic testing is preferably performed using array comparative genomic hybridization, targeted NGS gene panels, or whole-exome sequencing [3]. In our patient, NGS identified a novel homozygous missense variant in the *TG* gene. The absence of extrathyroidal or syndromic features is consistent with the typically non-syndromic nature of thyroglobulin-related dyshormonogenesis.

Although the identified *TG* c.847C > T variant was classified as a VUS according to ACMG criteria, multiple lines of evidence support its clinical pathogenic relevance in the present case. First, the variant was detected in the homozygous state in an infant born to consanguineous parents, consistent with the well-established autosomal recessive inheritance pattern of thyroglobulin-related dyshormonogenesis [15]. Second, the biochemical phenotype, markedly reduced serum thyroglobulin levels in the presence of a massive congenital goiter, closely resembles key elements of the characteristic laboratory profile described in patients harboring pathogenic *TG* variants [2, 15]. Previous studies have demonstrated that *TG* variants causing defective protein folding, impaired intracellular trafficking, or endoplasmic reticulum retention typically lead to very low or undetectable circulating thyroglobulin levels despite pronounced thyroid hyperplasia [15].

Pathogenic *TG* variants reported to date include a substantial proportion of missense changes affecting highly conserved residues and are frequently associated with severe neonatal or early-onset presentations, including congenital goiter and permanent hypothyroidism [2, 4, 5, 15]. The p.Arg283 residue is located within the N-terminal region of the thyroglobulin protein. Structural and biochemical studies have demonstrated that human thyroglobulin is organized into distinct domains, including a well-defined N-terminal domain, which is critical for correct protein folding and forms part of the hormonogenic framework required for thyroid hormone biosynthesis [19, 20]. Disruption of structurally important arginine residues has previously been implicated in loss-of-function effects due to protein misfolding and intracellular retention of thyroglobulin [15]. Accordingly, disruption of conserved residues within this region may impair intracellular trafficking and hormonogenesis, even in the absence of direct residue-specific functional data.

In the absence of alternative etiologies, the strong genotype-phenotype concordance observed in this patient, together with the rapid and sustained reduction in goiter size following levothyroxine therapy, strongly supports the clinical pathogenicity of this novel homozygous *TG* variant. Nevertheless, future functional studies will be required to definitively confirm the molecular mechanism by which this variant disrupts thyroglobulin processing and thyroid hormone biosynthesis.

Conclusion

This case illustrates an exceptionally severe neonatal presentation of thyroglobulin-related dyshormonogenetic congenital hypothyroidism, in which a massive congenital goiter led to critical tracheal compression, immediate postnatal respiratory failure, and mechanical ventilation. While goiter is a recognized feature of *TG* gene defects, neonatal airway obstruction requiring intubation at birth remains exceedingly rare. The absence of antenatal detection despite regular prenatal follow-up, together with the identification of a previously unreported homozygous *TG* missense variant, underscores both the phenotypic variability of *TG*-related dyshormonogenesis and the potential for life-threatening airway compromise in the immediate neonatal period.

Declarations

Ethics approval

Ethical approval was not required for this case report in accordance with institutional policies and national regulations.

Consent to participate

Not applicable.

Consent for publication

Written informed consent for publication of this case report and accompanying images was obtained from the patient's legal guardian.

Conflict of interest

The authors declare that they have no competing interests.

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Data availability

Data sharing is not applicable to this case report as no datasets were generated or analyzed during the current study.

Code availability

Not applicable.

Authors' contributions

AA and KŞK contributed to patient management and data collection. SÖ and ZMY performed and interpreted the genetic analyses. AMK interpreted the radiological findings. AA supervised the study and critically revised the manuscript. All authors contributed to the drafting of the manuscript and approved the final version.

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Figures



Figure 1

Serial clinical images of the neck mass

(a) Clinical appearance at birth showing prominent bilateral cervical swelling causing airway compromise, consistent with massive congenital goiter (b) Follow-up clinical image at 2 months demonstrating complete regression of the neck mass after levothyroxine therapy



Figure 2

Evolution of thyroid imaging from birth to 2 months

(a) Coronal T2-weighted neonatal MRI showing markedly enlarged and diffusely hyperintense thyroid lobes consistent with thyroid hyperplasia, causing anterior neck bulging (b) Sagittal T1-weighted neonatal MRI demonstrating a massive goiter causing tracheal compression and mild airway narrowing

at the cervical level (c) Follow-up ultrasonography at 2 months showing a substantial reduction in thyroid gland size compared with birth, although thyroid volume remained enlarged for age, with normalization of the tracheal contour and resolution of airway compression



Figure 3

Integrative Genomic Viewer image demonstrating a novel homozygous *TG* gene variant

Next-generation sequencing identified a previously unreported homozygous missense variant in the *TG* gene (c.847C>T; p.Arg283Trp), classified as a variant of uncertain significance