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General Medicine

CONGENITAL HYPOTHYROIDISM PRESENTING AS SEVERE ANEMIA AND MENORRHAGIA: A CASE REPORT

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ABSTRACT **Background:** Congenital primary hypothyroidism, occurring in approximately 1:2000 to 1:4000 newborns, is one of the most common preventable causes of intellectual disability worldwide.The prevalence of congenital hypothyroidism in India (1:1000) is higher than the other countries (1:3000–4000). A dramatic improvement can be made by early detection, diagnosis, and adequate treatment with levothyroxine in patients. In settings without universal neonatal screening, patients may remain undiagnosed until late childhood, Severe hypothyroidism may also present with menstrual abnormalities and anemia, which can divert attention from the underlying endocrine disorder. **Case Presentation:** A 22-year-old woman presented to the emergency department with excessive per-vaginal bleeding for 15 days, progressive fatigue, exertional breathlessness, cold intolerance, and a history of irregular menstrual cycles. She had delayed milestones, severe short stature, poor scholastic performance, delayed menarche, coarse facies, macroglossia, umbilical protrusion, delayed secondary sexual development, and delayed skeletal maturation. Laboratory evaluation showed severe anemia with hemoglobin of 2.1 g/dL, microcytic hypochromic picture on peripheral smear, very high thyroid-stimulating hormone (>150 microIU/mL), low thyroid hormone levels, and ultrasonographic evidence of a hypoplastic thyroid gland. She was managed with packed red cell transfusion, iron sucrose, vitamin B12 supplementation, and levothyroxine replacement. **Conclusion:** This case highlights a rare adult presentation of untreated congenital hypothyroidism masquerading as a gynecologic and hematologic emergency. In patients with severe menorrhagia and anemia, the coexistence of short stature, developmental delay, coarse facies, macroglossia, delayed puberty, cold intolerance, and umbilical hernia should prompt thyroid function testing. Early neonatal screening and timely levothyroxine therapy remain crucial to prevent irreversible morbidity.

KEYWORDS : Congenital Hypothyroidism; Severe Anemia; Menorrhagia; Thyroid Hypoplasia; Delayed Diagnosis; Newborn Screening

INTRODUCTION

Congenital hypothyroidism (CH) is a common preventable cause of intellectual disability and growth failure. Reported prevalence in India is high, with estimates around 1:1000 live births. Early diagnosis and adequate levothyroxine replacement can substantially improve growth and neurodevelopmental outcomes; therefore, newborn screening and timely confirmation remain important preventive strategies [1-3].

In India, implementation of universal newborn screening for CH remains inconsistent, and delayed diagnosis continues to be reported [2-6]. Delayed or missed diagnosis allows classical features such as short stature, coarse facies, macroglossia, umbilical hernia, delayed skeletal maturation, delayed puberty, and cognitive impairment to progress. Adult presentation is uncommon, but it is clinically important because the patient may first present to emergency, gynecology, hematology, or medicine services rather than endocrinology [2,7,8].

Hypothyroidism is associated with menstrual irregularities, including oligomenorrhea and menorrhagia. Severe menstrual blood loss may lead to iron deficiency anemia and, rarely, life-threatening anemia [6,7]. We report a case of untreated congenital hypothyroidism due to thyroid hypoplasia presenting in adulthood with severe anemia and menorrhagia, emphasizing the need for thyroid evaluation in patients with abnormal uterine bleeding when syndromic clinical clues are present.

Case Presentation

A 22-year-old woman from Bhokardan, Jalna, presented to the emergency department of Government Medical College and Hospital, Chhatrapati Sambhajinagar, on 9 February 2026 with excessive per-vaginal bleeding for 15 days. The bleeding required approximately 3-4 pads per day. Her family reported menstrual irregularities for the preceding 2 years, and the most recent cycle had lasted approximately 6 months. She also had easy fatigability, exertional breathlessness after walking about 100 meters, and intolerance to cold weather.

She was the fifth child of a non-consanguineous marriage. She was delivered at term by normal vaginal delivery, cried immediately after birth, and had no history of neonatal intensive care unit stay. Developmental delay was evident from childhood: she sat independently after approximately 15 months and walked at around 3 years of age. Her family also reported persistent short stature,poor scholastic performance, and inability to read or write. Menarche occurred late, at 18 years of age, followed by irregular cycles lasting between 2 and 5 months.

The patient had two previous hospital admissions for severe anemia. In August 2024, her hemoglobin was reportedly 3.7 g/dL with total leukocyte count of 7200/cumm and platelet count of 2.27 lakh/cumm; she received two packed red cell transfusions but was not evaluated further. In July 2025, she was again admitted for severe anemia. Hemoglobinopathy screening by high-performance liquid chromatography was within normal limits, while the peripheral smear showed microcytes and pencil cells. She was transfused with one packed red cell unit and discharged.

Clinical Examination

At the current presentation, her vital parameters were stable. General examination revealed pallor, facial and periorbital puffiness, coarse dry skin, thick hair, depressed nasal bridge, and macroglossia. Anthropometry showed severe short stature with height of 92 cm, weight of 24 kg, and upper-to-lower segment ratio of 42:50. Secondary sexual characteristics were delayed, with breast development corresponding to Tanner stage 2 and pubic hair corresponding to Tanner stage 1. Cognitive assessment using the Binet-Kamat Test of Intelligence showed a mental age of approximately 3 years. Abdominal examination revealed distension with umbilical protrusion. The clinical photographs documented short stature, coarse facial appearance, macroglossia, puffy extremities, and protuberant abdomen with umbilical protrusion (Figures 1 and 2).

Figure 1. Clinical Phenotype. Panel A Shows Severe Short Stature;

Panel B Shows Coarse Facial Appearance With Facial Puffiness; Panel C Shows Macroglossia. The Eyes Were Obscured in the Source Images.

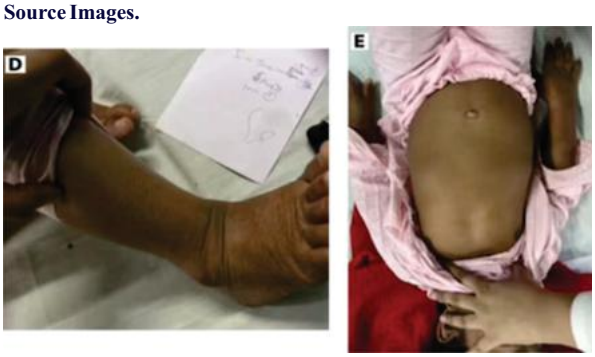

Figure 2. Additional Clinical Signs. Panel D Shows Puffy/coarse Extremity Changes; Panel E Shows Protuberant Abdomen With Umbilical Protrusion.

Investigations

Serial complete blood count and biochemical parameters are summarized in Table 1. The hemoglobin level at presentation was critically low. The peripheral smear showed anisocytosis, microcytes, polychromatic red blood cells, and moderate-to-severe hypochromia, supporting microcytic hypochromic anemia. Sickling test and direct and indirect Coombs tests were negative, reticulocyte count was 4%, and non-contrast computed tomography of the brain was within normal limits.

Table 1. Serial Hematological and Biochemical Parameters During Admission

Parameter	Reference range	09/02/2026	10/02/2026	12/02/2026
Hemoglobin	12-14 g/dL	2.1	5.3	10.7
Total leukocyte count	4000-10,000/cumm	20,000	12,000	7200
Platelet count	1.4-4.5 lakh/cumm	1.18	1.09	1.8
MCV	80-100 fL	75	76	81
Urea	13-43 mg/dL	16	12	12
Creatinine	0.5-1.3 mg/dL	0.42	-	0.5
Total bilirubin	0.3-1.2 mg/dL	2.59	-	1.5
SGOT/AST	8-40 U/L	87	-	46
SGPT/ALT	7-56 U/L	66	-	40

MCV, mean corpuscular volume; SGOT/AST, serum glutamic oxaloacetic transaminase/aspartate aminotransferase; SGPT/ALT, serum glutamic pyruvic transaminase/alanine aminotransferase.

The thyroid profile was diagnostic of severe primary hypothyroidism, with thyroid-stimulating hormone greater than 150 microIU/mL and low thyroid hormone levels (Table 2). Ultrasound of the neck showed a hypoplastic thyroid gland. Contrast-enhanced computed tomography of the abdomen and pelvis showed exaggerated lumbar lordosis, non-fused sacral vertebrae, umbilical herniation ofomental fat, non-fused bilateral femoral epiphyses, mild ascites, and bulky uterus with bilateral ovaries within normal limits.

Table 2. Hormonal Profile

Parameter	Patient value	Reference range
T3	1.18	2.5-3.9
T4	0.11	0.61-1.12
TSH	>150	0.38-5.33
8 AM cortisol	9.54	6.02-18.4
LH	0.3	2.4-12.6 (follicular phase)
FSH	3.94	3.5-12.5 (follicular phase) mIU/mL
PTH	42.78	15-65 pg/mL
Growth hormone	1.04	0.12-9.88

TSH, thyroid-stimulating hormone; LH, luteinizing hormone; FSH, follicle-stimulating hormone; PTH, parathyroid hormone. Units are presented as recorded in the source presentation where available; values should be verified against the original laboratory report before journal submission.

Diagnosis

Based on the clinical phenotype, biochemical profile, and ultrasonographic evidence of thyroid hypoplasia, a diagnosis of untreated congenital hypothyroidism due to thyroid hypoplasia was considered. Severe microcytic hypochromic anemia was attributed primarily to chronic menstrual blood loss in the setting of hypothyroidism-associated menstrual dysfunction. The patient also had delayed puberty, severe short stature, intellectual impairment, delayed skeletal maturation, and umbilical hernia consistent with long-standing untreated hypothyroidism.

Management

The patient was managed acutely with packed red cell transfusion, iron sucrose, and vitamin B12 supplementation. Levothyroxine was initiated at 25 micrograms once daily and subsequently increased to 50 micrograms once daily. She was advised endocrine follow-up for titration of levothyroxine dose, monitoring of thyroid function tests, assessment of pubertal status, nutritional rehabilitation, and gynecologic follow-up for menstrual regulation and anemia recovery.

DISCUSSION

This case demonstrates an uncommon but clinically instructive adult presentation of congenital hypothyroidism. Menorrhagia and severe anemia were the immediate reasons for emergency presentation, but the associated history and examination revealed a long-standing endocrine disorder. The coexistence of delayed milestones, severe short stature, delayed menarche, coarse facies, macroglossia, umbilical hernia, delayed secondary sexual development, and cold intolerance strongly suggested chronic hypothyroidism rather than isolated gynecologic disease.

The clinical spectrum of congenital hypothyroidism varies with age at presentation. During infancy, children may present with feeding difficulty, constipation, prolonged jaundice, lethargy, hoarse cry, and poor growth. With increasing age, untreated patients develop coarse facies, dry skin, protuberant abdomen, delayed dentition, developmental delay, delayed bone age, growth failure, and cognitive impairment [2,4,8]. Adult cases are rare because most patients are now diagnosed through newborn screening or during childhood; however, reports from India continue to describe delayed diagnosis where screening is unavailable or clinical suspicion is low [2,4,5,7].

The gynecologic manifestation in the present case is important. Hypothyroidism can alter the hypothalamic-pituitary-ovarian axis and may lead to anovulatory cycles, unopposed estrogen effect on the endometrium, altered sex hormone-binding globulin levels, and menstrual irregularities. Profound hypothyroidism has been reported as a cause of severe menorrhagia and life-threatening anemia [6]. Similar adult presentation of congenital hypothyroidism with menorrhagia has also been documented [7]. In the present case, microcytosis, hypochromia, pencil cells, and recurrent transfusion requirement support chronic blood loss with iron deficiency as an important contributor to anemia.

The endocrine profile, with markedly elevated TSH and low T3/T4, confirmed severe primary hypothyroidism. The hypoplastic thyroid gland on ultrasonography supported congenital thyroid dysgenesis as the likely underlying cause. The non-fused epiphyses and delayed secondary sexual development further reflected prolonged thyroid hormone deficiency. Although levothyroxine replacement is simple, inexpensive, and effective, delayed initiation may not reverse established neurocognitive deficits completely; hence prevention through neonatal screening is essential [1,5,8].

This case reinforces two practical messages. First, thyroid dysfunction should be included in the evaluation of unexplained menorrhagia and severe anemia, especially when systemic features of hypothyroidism are present. Second, universal newborn screening for congenital hypothyroidism should be strengthened because early treatment can prevent intellectual disability, severe growth failure, delayed puberty, and avoidable adult morbidity [1,5,8].

CONCLUSION

Untreated congenital hypothyroidism can rarely present in adulthood as severe menorrhagia with life-threatening anemia. The presence of short stature, developmental delay, delayed puberty, coarse facies, macroglossia, cold intolerance, delayed skeletal maturation, and umbilical hernia should prompt immediate thyroid function testing. Early diagnosis through universal neonatal screeningand timely levothyroxine therapy remains the most effective strategy to prevent irreversible growth and neurodevelopmental morbidity.

Learning Points

- Severe hypothyroidism is an important and treatable cause of abnormal uterine bleeding and anemia.
- Menorrhagia with severe anemia should not be treated only as a gynecologic or hematologic problem when systemic features such as short stature, developmental delay, coarse facies, macroglossia, cold intolerance, and delayed puberty are present.
- Delayed diagnosis of congenital hypothyroidism can result in severe short stature, delayed skeletal maturation, intellectual impairment, delayed puberty, and avoidable adult morbidity.
- Universal newborn screening and early levothyroxine therapy are essential preventive measures.

Declarations

Patient Consent: Written informed consent for publication of the clinical details and images should be obtained from the patient and/or legally authorised representative and documented before submission.

Ethical Approval: For a single anonymized case report, formal ethics committee approval may not be mandatory; however, institutional policy should be followed.

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Conflicts of Interest: The authors declare no conflicts of interest.

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