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Case Report

Comprehensive Imaging Manifestations in Transfusion-Dependent Beta-Thalassemia Major with Secondary Diabetes Mellitus: A Case Report

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A B S T R A C T

Background: Transfusion-dependent beta-thalassemia major (TDT) is characterized by systemic iron overload affecting multiple organ systems. Iron deposition patterns follow distinct distributions, with reticuloendothelial deposition occurring primarily in the liver, spleen, lymph nodes and bone marrow following multiple transfusions. Secondary complications including diabetes mellitus develop as a consequence of pancreatic iron accumulation and beta-cell dysfunction1,2.

Case Presentation: We report a 15-year-old female with TDT presenting with fever and newly diagnosed diabetes mellitus. Sequential imaging studies demonstrated the comprehensive multi-organ manifestations of iron overload and its complications. Computed tomography revealed multiple hyperdense lymph nodes with characteristic coarse peripheral calcifications distributed throughout the porta hepatis, para-aortic and peripancreatic regions, consistent with iron deposition in the reticuloendothelial system1,3. The liver demonstrated significant enlargement with a beaver tail variant of the left lobe, while cholelithiasis was present with calculi measuring up to 7 mm. Post-splenectomy status was noted, which has been associated with accelerated pancreatic iron deposition4. Chest radiography showed rapid progression from normal lung fields to non-homogeneous right-sided opacities over a 4-day interval. Ultrasonography revealed hepatomegaly, pancreatic hyperechoic changes suggesting iron deposition and significantly underdeveloped reproductive organs including an infantile uterus and streak ovaries, indicating hypogonadotropic hypogonadism secondary to iron-related hypothalamic-pituitary dysfunction2-4.

Discussion: This case illustrates the reticuloendothelial pattern of iron deposition characteristic of transfusion-related hemochromatosis, where iron accumulates predominantly in Kupffer cells, splenic macrophages and lymph node reticuloendothelial cells3. The development of secondary diabetes mellitus correlates with pancreatic iron accumulation, which begins in early childhood and progressively affects pancreatic beta-cell function5. The hyperdense lymphadenopathy observed on CT reflects iron-laden reticuloendothelial cells within expanded cortical and medullary sinusoids, a finding first described by Mitnick et al. in 19816. The concurrent endocrine manifestations, including hypogonadism and diabetes, demonstrate the multi-system impact of chronic iron toxicity beyond the classical hepatic and cardiac complications1-3.

Conclusion: This case demonstrates the comprehensive imaging spectrum of transfusion-dependent thalassemia major, highlighting the reticuloendothelial pattern of iron distribution and its associated complications. Early recognition of these imaging findings is crucial for guiding intensive chelation therapy and preventing irreversible organ damage, particularly in the pancreas and endocrine system where iron-induced dysfunction can significantly impact long-term patient outcomes.

Keywords: Beta-thalassemia major; Iron overload; Reticuloendothelial deposition; Diabetes mellitus; Lymphadenopathy; Hypogonadism; Comprehensive imaging

Introduction

Thalassemia and structural hemoglobin variants are the most common monogenic disorders worldwide. In India, the disease burden is significant, with an estimated 100,000 individuals affected by β-thalassemia syndromes and about 150,000 by sickle cell disease. However, most patients do not receive adequate care and curative treatment such as allogeneic stem cell transplantation remains beyond the financial reach of most families[7].

The introduction of regular red cell transfusion therapy six decades ago changed β-thalassemia majorly from a fatal disease of childhood into a manageable chronic condition. Subsequent progress in preventing transfusion-related infections and controlling iron overload has further improved outcomes, enabling patients to achieve survival rates and quality of life that are now close to normal[8].

Survival in transfusion-dependent β-thalassemia (TDT) has improved with advances in transfusion protocols and oral iron chelation therapy. However, mortality continues to remain high in India and other low- to middle-income countries. At 26.9 years of age, actuarial survival is only 50% with under-5 mortality being 7 times higher than in the general population. The most common cause of death in these patients is infection[9].

Radiological evaluation plays a crucial role in monitoring disease progression and detecting complications in thalassemia patients. Cross-sectional imaging modalities, particularly computed tomography (CT) and ultrasonography, can demonstrate characteristic findings of iron deposition organ dysfunction and associated metabolic disturbances. We present a comprehensive case demonstrating the spectrum of imaging manifestations in a teenage patient with transfusion-dependent thalassemia major complicated by secondary diabetes mellitus[10-12].

Case Presentation

Patient demographics and clinical history

A 16-year-old female, Pranali Prakash Hailndgear, presented with persistent fever for 10 days, body ache and recent onset of polyuria and polydipsia. She had a well-documented history of beta-thalassemia major diagnosed in infancy, requiring regular blood transfusions every 3-4 weeks. Splenectomy was performed at age 8 years due to hypersplenism. The patient had been receiving iron chelation therapy with deferasirox.

Growth parameters consistently tracked at the 3rd percentile for age. Physical examination revealed characteristic thalassemic facies, pallor and absence of secondary sexual characteristics. Family history was significant for thalassemia trait in both parents.

Laboratory findings

- **Hematological:** Severe microcytic hypochromic anemia (hemoglobin: 7.4 g/dL), requiring transfusion.
- **Metabolic:** Random blood glucose 598 mg/dL, confirming diabetes mellitus.
- **Iron studies:** Markedly elevated serum ferritin (11,896 ng/mL), indicating severe iron overload.
- **Endocrine:** Lower side of normal follicle-stimulating hormone levels (0.35 mIU/ml).
- **Infectious markers:** Hepatitis C virus serology positive, PCR negative.

Imaging Findings

Computed tomography of abdomen and pelvis

Technical parameters: Contrast-enhanced study performed using 120 kVp, with oral and intravenous contrast administration.

Key Findings: Contrast-enhanced CT of the abdomen and pelvis in this patient revealed multiple enlarged lymph nodes with characteristic hyperdense coarse peripheral calcifications distributed throughout the portal, left gastro-hepatic, pre and para-aortic, aorto-caval and peripancreatic stations, with the largest measuring 40.8 x 28.9 mm in the aorto-caval region, all demonstrating uniform post-contrast enhancement (**Figure 1**).

The liver showed enlargement of the left lobe extending to the left hypochondrium consistent with a beaver tail variant (right lobe span 12.9 cm), with smooth contours and no focal lesions. Notably the gallbladder was distended with a 6 mm hyperdense calculus in the fundus.

The spleen was absent consistent with prior splenectomy.

The pancreas, kidneys, ureters and adrenals appeared normal with appropriate contrast enhancement.

The visualized axial skeleton demonstrated coarsened trabeculae characteristic of thalassemic bone changes, with no paravertebral masses identified (**Figure 2**).

No ascites, hydronephrosis or other significant abnormalities were observed in the bowel loops, pelvis or lower lung fields.

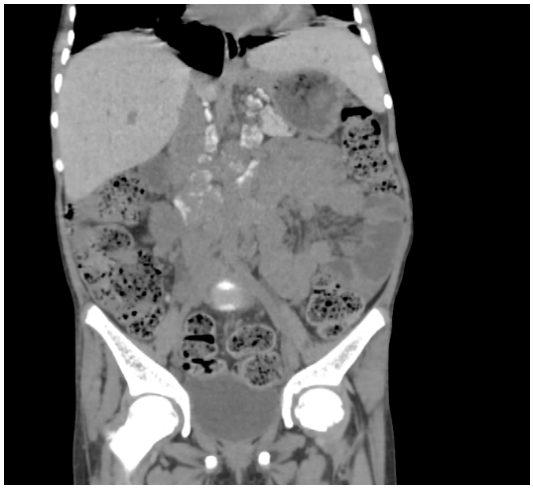

Figure 1: Coronal reformatted non contrast CT images after the administration of neutral oral contrast showing multiple retroperitoneal lymph nodes demonstrating characteristic hyperdense coarse peripheral calcification (as noted by the arrows) in the left gastro-hepatic, pre- and para-aortic and aorto-caval groups

Abdominal ultrasonography

Abdominal and pelvic ultrasonography in this patient demonstrated hepatomegaly with the liver measuring 14.7 cm, with enlarged left lobe and normal echogenicity. Cholelithiasis was consistently present, with gallstones measuring up to 6 mm on follow-up, contained within a normally distended gallbladder with normal wall thickness.

The pancreas showed interval development of hyperechoic

echotexture, potentially reflecting iron deposition or early diabetic changes, correlating with the newly diagnosed diabetes mellitus noted in the clinical history. Notably, the study reproductive system abnormalities including an infantile uterus (corpus length 1.7 cm, AP diameter 0.3 cm; cervix length 1.7 cm, AP diameter 1.3 mm) and bilateral streak ovaries (right: 0.7 x 0.1 cm, left: 1.0 x 0.1 cm), consistent with hypogonadotropic hypogonadism secondary to iron overload affecting the hypothalamic-pituitary-gonadal axis (**Figure 3**).

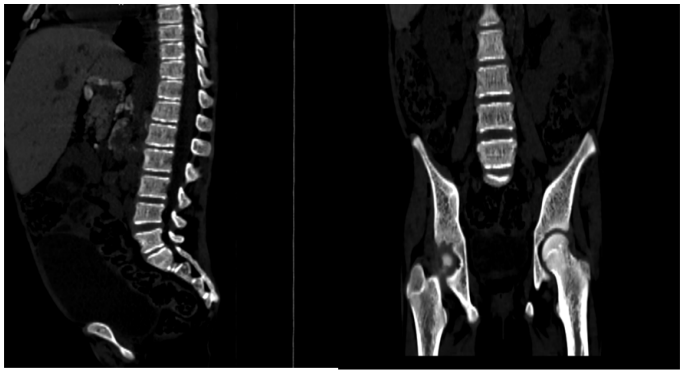

Fig 2(a) Fig 2(b)

Figure 2(a): Non contrast CT abdomen axial image showing hepatomegaly with left lobe enlargement, extending to the left hypochondrium (beaver tail variant), **2(b):** Non contrast axial image of the abdomen showing a distended gall bladder containing a 7 mm hyperdense calculus (arrow).

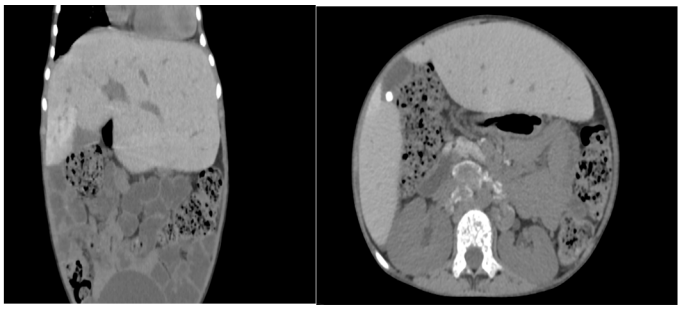

Fig 3(a) Fig 3(b)

Figure 3(a,b): Bone window CT images of the abdomen in the sagittal and coronal plane window showing coarse trabeculations in the axial skeleton.

Both kidneys maintained normal size, echogenicity and corticomedullary differentiation throughout both examinations.

These findings illustrate the multi-organ complications of thalassemia major, including hepatic iron overload, cholelithiasis, pancreatic dysfunction and endocrine failure manifesting as delayed puberty and diabetes mellitus.

Discussion

This case exemplifies the comprehensive imaging spectrum encountered in advanced transfusion-dependent beta-thalassemia major with secondary complications. The radiological findings can be systematically categorized into several key areas of organ involvement.

Iron overload manifestations

The most striking finding was the extensive retroperitoneal lymphadenopathy with characteristic hyperdense rims on non-contrast CT imaging. This appearance is pathognomonic of iron deposition (hemosiderosis) within lymphoid tissue, a direct consequence of chronic transfusion therapy. The hyper density reflects the paramagnetic properties of deposited iron, which appears hyperattenuating on CT. The uniform post-contrast enhancement pattern distinguishes these nodes from neoplastic processes, which typically demonstrate heterogeneous enhancement patterns[1].

Hepatomegaly in this patient represents both extramedullary hematopoiesis and iron deposition within hepatocytes and Kupffer cells. The preservation of smooth hepatic contours without focal lesions suggests early-stage involvement without significant fibrosis, though histological correlation would be definitive[10].

Biliary complications

Cholelithiasis is a well-recognized complication of chronic hemolytic disorders, including thalassemia[13]. The formation of pigment stones results from increased bilirubin production due to chronic hemolysis, compounded by post-splenectomy changes that can alter red blood cell survival. The hyperdense appearance of the calculus on CT suggests a high concentration of calcium bilirubinate[14].

Endocrine dysfunction

The ultrasonographic demonstration of infantile uterus and streak ovaries provides direct imaging evidence of hypogonadotropic hypogonadism, a common endocrine complication of transfusional iron overload. Iron deposition in the anterior pituitary gland disrupts gonadotropin-releasing hormone production, leading to delayed or absent puberty. This finding correlates with the patient's physical examination and low FSH levels[11,12].

Secondary diabetes mellitus

While not directly visualizable on imaging, the development of diabetes mellitus in this patient reflects pancreatic iron deposition affecting beta-cell function. Future imaging with MRI using T2* sequences could quantify pancreatic iron content and monitor the efficacy of chelation therapy[15].

Skeletal manifestations

The coarsened trabecular pattern observed on CT represents the skeletal response to chronic anemia and extramedullary hematopoiesis. Marrow expansion led to cortical thinning and altered trabecular architecture, predisposing patients to pathological fractures[10,16].

Infectious Complications

The development of pulmonary opacities on chest radiography warrants consideration of opportunistic infections, as patients with thalassemia major have increased susceptibility due to iron overload-induced immune dysfunction and post-splenectomy status[17,18].

Clinical implications

This comprehensive imaging assessment demonstrates several key clinical implications:

- **Monitoring protocol:** Regular imaging surveillance is essential for detecting complications before they become clinically apparent.
- **Chelation efficacy:** The extent of iron deposition visualized suggests suboptimal chelation therapy efficacy, warranting dose adjustment or alternative chelating agents.