Extramedullary Hematopoiesis in β-Thalassemia

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A 37-year-old man is being followed up at the hematology clinic at our institution for β-thalassemia intermedia. He was diagnosed with β-thalassemia during infancy and required multiple blood transfusions, resulting in iron overload. At the age of 23 years, he underwent splenectomy, obviating the need for further transfusions. Several years later, pulmonary arterial hypertension (PAH) was diagnosed by means of right heart catheterization. Physical examination of the patient revealed a short stature and a body mass index (calculated as the weight in kilograms divided by the height in meters squared) of 22 kg/m^2; frontal bossing and a prominent maxilla were consistent with extramedullary hematopoiesis; bronze-colored skin and marked hepatomegaly were evident as signs of iron overload. Recent hemoglobin and ferritin levels were 8 g/dL and 2502 mg/dL, respectively. Chest radiography in the posterior-anterior projection revealed thickened ribs bilaterally, disruption of the anatomic border of the anterior ribs, as well as narrowing of the intercostal spaces. Along the costovertebral junctions, multiple extrapleural masses were noted (Figure A). The lateral projection demonstrated additional masses on the anterior chest wall (Figure B). Furthermore, roentgenographic signs of pulmonary hypertension were noted in the form of an enlarged pulmonary trunk, prominent right atrium (Figure A), and opacification of the retrosternal space, indicating an enlarged right ventricle (Figure B). These radiological findings, consistent with extramedullary islets of hematopoietic tissue and signs of pulmonary hypertension, were later confirmed by computed tomography of the chest, which also showed gross trabeculation of the ribs and vertebra (see supplemental Figure, available online at http://www.mayoclinicproceedings.org).

Extramedullary hematopoiesis is a well-documented manifestation of thalassemia,[1] as well as other severe disorders of hematopoiesis. Hematopoietic cell colonies may disrupt the cortex of the bones involved and even form masses of extramedullary hematopoietic material in the thoracic[2] or pelvic[3] cavities as well as other anatomic regions. These, in turn, can cause a mass effect in their respective anatomical location, thus causing damage to adjacent structures, such as spinal cord compression and vascular occlusions.

Pulmonary arterial hypertension is a common complication of the more severe forms of thalassemia,[4] with a prevalence of more than 50% in some cohorts.[5] It is postulated that the excess of free hemoglobin in the blood causes endothelial injury as well as nitric oxide depletion, thus leading to the development of PAH.[6] The condition is often asymptomatic but may cause functional impairment in some of the patients. Splenectomy and older age have been shown to be risk factors for developing PAH.[7]

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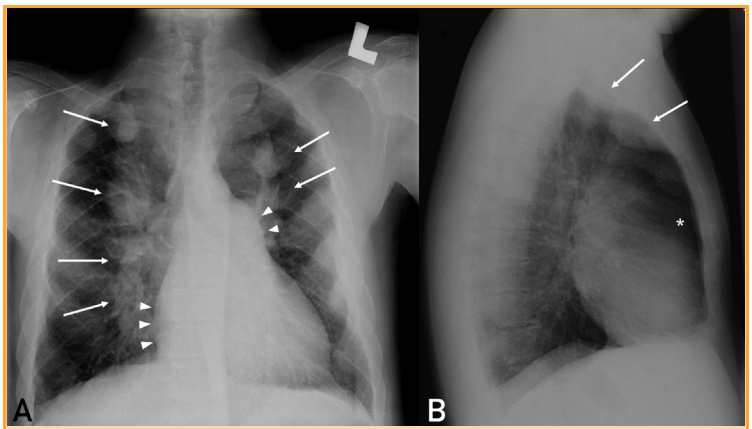

FIGURE. A, Chest radiography in the anterior-posterior projection demonstrating bilateral paravertebral islets of extramedullary hematopoietic tissue (*arrows*) and prominent pulmonary trunk and right atrium (*arrowheads*). B, In chest radiography in the lateral projection, extramedullary islets of the anterior chest (*arrows*) and an enlarged right ventricle (*asterisk*) are evident.

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SUPPLEMENTAL ONLINE MATERIAL

Supplemental material can be found online at http://www.mayoclinicproceedings.org. Supplemental material attached to journal articles has not been edited, and the authors take responsibility for the accuracy of all data.

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