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THORACIC EXTRAMEDULLARY HEMATOPOIESIS SECONDARY TO ENZYMATIC DEFICIENCY

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Background: A 63-year-old male was referred to the anesthesiology department for check-up prior to cataract surgery. Clinical and biological findings showed mild jaundice, reticulocytosis, and indirect hyperbilirubinemia which was known for several years and had already taken the patient to cholecystectomy for a presumed diagnosis of cholecystitis se-veral years earlier. Chest X-ray, contrast-enhanced CT and MR of the thorax, and nuclear bone scintigraphy were performed.

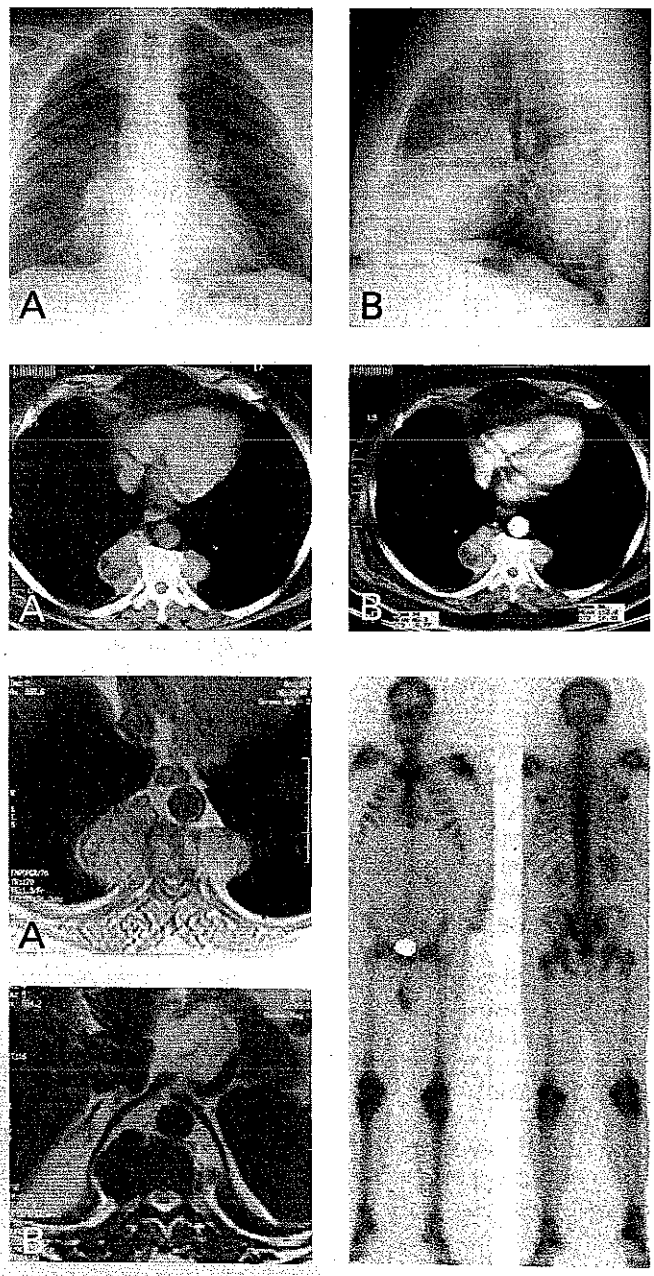

Fig. 1A | 1B / 2A | 2B / 3A | 3B — 4

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Work-up

Chest radiography (Fig. 1) shows bilateral paravertebral opacities with a silhouette sign (descending aorta) for the left one (PA view) and inversion of thoracic spine translucence gradient (lateral view).

Unenhanced and contrast-enhanced CT scan of the chest (Fig. 2) reveals presence of two masses occupying left and right paravertebral mediastinal regions, strongly enhancing after contrast injection, containing areas of fatty tissue. Both masses are well-delineated without peritumoral infiltration.

MR of the chest (Fig. 3) included axial T1- (A) and T2-weighted (B) imaging which confirm the nodular paravertebral masses of intermediate signal intensity containing fatty areas, without perilesional edema or infiltration. All sequences performed show mimicry of signal in the masses with bone marrow.

99-Technecium bone scintigraphy (Fig. 4) shows increased tracer fixation in long bones particularly in the epiphyseal regions. Areas of hyperfixation at the right calcaneum and both tibiae are probably post-traumatic.

Radiological diagnosis

Radiological findings enabled the diagnosis of *thoracic extramedullary hematopoiesis secondary to enzymatic deficiency*.

Discussion

Differential diagnosis of thoracic paravertebral masses includes neurogenic tumors, lymphadenopathies (lymphoma, Castelman disease), tumoral spread from adjacent structures and extramedullary hematopoiesis (EMH). In our patient, MR- and CT-findings were suggestive for marrow pathology, confirmed by nuclear bone scan and by bone marrow biopsy showing an erythroid, predominant hyperplastic marrow suggestive of a peripheral hemolytic pattern. CT-guided biopsy of right paravertebral mass confirmed EMH. Further biological tests revealed enzymatic glucose-6-P isomerase deficiency responsible for inefficient erythropoiesis, resulting in compensatory EMH.

EMH is a response to erythropoetic failure in bone marrow. It can occur in various conditions as thalassemia major or intermedia, polycytemia vera, essential thrombocytosis and other myeloproliferative diseases including myelofibrosis with myeloid metaplasia. Most frequently liver and spleen are affected. EMH may rarely involve other organs such as kidney, thymus or retroperitoneum and rarely give rise to intrathoracic mediastinal, pleural or pulmonary masses. Pathogenesis includes a hemolytic form due to herniation of bone marrow through cortex of ribs and/or vertebrae into paravertebral space, and a myelolytic form due to in situ heterotopic or embolized multipotential cells in visceral sites. In chronic congenital hemolytic anemias, EMH is a response to increased renal erythropoietin excretion depending on hemoglobin level. In our case, hemolysis was induced by a lack of glucose-6-P isomerase, affecting glycolysis and TPA production resulting in hereditary non spherocytary hemolytic anemia.

EMH is mostly asymptomatic. As more than 85% of EMH occurs in patients having long lasting anemia or myeloproliferative disorders, EMH should be searched in these conditions. CT scan and MRI will show characteristic location and density/signal intensity of EMH-lesions.

Spontaneous regression of masses is uncommon, even when the anemia is stabilized. Growth is usually slow and not steady, depending on hemolytic attacks. Complications include compressive syndromes, bone damage (hemolytic EMH) and bleeding which is often massive and fatal, requiring precise and rapid diagnosis. Therefore once discovered, EMH must be followed regularly to look for complications before they manifest clinically.

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