Extramedullary hematopoiesis in a child with hereditary spherocytosis: an uncommon cause of an adrenal mass

Abstract We report a case of extramedullary hematopoiesis presenting as an adrenal mass in a young male with hereditary spherocytosis. The unilateral adrenal mass was discovered during an abdominal ultrasound performed for jaundice. CT and MR imaging were subsequently performed, followed by an excisional biopsy at the time of splenectomy and cholecystectomy. Although extramedullary hematopoiesis is a rare cause of an adrenal mass, the diagnosis must be considered in any patient with a history of a congenital hemolytic disorder such as hereditary spherocytosis. In this regard, the morbidity of an unnecessary procedure may be avoided.

Introduction
Extramedullary hematopoiesis is an uncommon soft tissue mass. The diagnosis, however, should be considered in the appropriate clinical setting. Previously described sites have included the posterior mediastinum, epidural space, liver, spleen, lymph nodes and the adrenal gland. Although several cases of adrenal extramedullary hematopoiesis have been described in patients with thalassemia, to our knowledge, this is the first case in a patient with hereditary spherocytosis.

Case report
A 9-year-old male with a history of hereditary spherocytosis was admitted to the hospital for jaundice. Abdominal ultrasound revealed cholelithiasis, splenomegaly and an approximately 5-cm lobulated, solid, hypoechogenic right suprarenal mass (Fig. 1).

Subsequently, the patient underwent a non-contrast CT scan, which also demonstrated a solid, lobulated right suprarenal mass (Fig. 2). An MRI was performed in an attempt to characterize this mass further. The mass appeared to be of adrenal origin and showed low signal intensity on T1, heterogeneously bright signal intensity on T2 and only slight enhancement (Fig. 3a–c). A differential diagnosis was given, including neoplasm, ganglioneuroma, extramedullary hematopoiesis, and adrenal hyperplasia. An endocrine workup including cortisol, VMA, ACTH, HVA and plasma renin levels was negative.

The patient subsequently underwent splenectomy, cholecystectomy, and partial right adrenalectomy. Pathologic evaluation of a 19-g specimen from the right adrenal gland demonstrated a hemorrhagic, soft, bilobed mass measuring 5.5 × 5 × 2 cm with a smooth, shiny external surface. Frozen and microscopic section revealed extramedullary hematopoiesis surrounded by a rim of benign adrenal tissue. Bilirubin gallstones and splenic congestion of the red pulp were also evident. The patient had an uneventful post-operative course and was discharged on post-operative day 4.

Discussion
Hereditary spherocytosis is a familial hemolytic disorder affecting approximately 1 in 5,000. Although the disorder is characterized by anemia, intermittent jaundice and splenomegaly, there is wide variation in clinical features. Morphologically, the disease is hallmarkd by the microspherocyte that results from one or more defects in membrane proteins. Spectrin is the most commonly implicated. The change in membrane proteins results in a rounded erythrocyte rather than the normal biconcave morphology. As a result, splenic hemolysis occurs leading to splenomegaly, jaundice, anemia, and a predilection for pigmented gallstones. Anemia may be
exacerbated by pregnancy, febrile illness, an increase in plasma volume, or an increase in the rate of hemolysis. The resultant compensatory erythropoiesis by bone marrow may result in expansion of the medullary cavities or development of ectopic hematogenous masses [1].

Extramedullary hematopoiesis, as its name implies, is the abnormal production of hematopoietic tissue in sites other than the bone marrow. This phenomenon can accompany a number of congenital hemolytic anemias or chronic myeloproliferative diseases, including thalassemia, sickle cell anemia, polycythemia rubra vera, chronic myelogenous leukemia, agnogenic myeloid metaplasia, and hereditary spherocytosis. Multiple parossseous and extraosseous sites have been reported, including the thorax, abdomen, and pelvis. Parossseous sites predominate with hemoglobinopathies; extraosseous sites predominate with myeloproliferative disorders [2]. Most reported cases of extramedullary hematopoiesis secondary to hereditary spherocytosis have documented intrathoracic sites of involvement [3].

The imaging characteristics in this case are consistent with those previously described in the literature [2, 4]. Since this process presents as a soft tissue mass, it often mimics a more serious diagnosis requiring radically different treatment options. Once diagnosed, surgery and radiotherapy are generally reserved for symptoms such as spinal cord compression [5]. The majority of patients, therefore, require no surgical treatment if the diagnosis of extramedullary hematopoiesis can be made. In some cases, biopsy may still be necessary to confirm the diagnosis.

Our review of the literature yielded reports of extramedullary hematopoiesis of the adrenal gland in patients with beta-thalassemia and agnogenic myeloid metaplasia [3, 5]. To our knowledge, this is the first report of extramedullary hematopoiesis occurring in the adrenal gland in a patient with hereditary spherocytosis. The diagnosis of extramedullary hematopoiesis should be considered in any patient with a history of hereditary spherocytosis and any intrathoracic or intraabdominal mass, including those of adrenal origin. This may potentially spare the patient an unnecessary invasive procedure, such as biopsy or surgical excision.