A patient with Beta thalassaemia major presenting with cauda equine syndrome due to extramedullary haematopoiesis

Abstract:

Beta-thalassaemia major is an autosomal recessively transmitted hereditary haematological disorder characterized by anomalies in the synthesis of the beta chains of hemoglobin resulting in anaemia requiring repeated blood transfusions. Inadequately transfused patients commonly present with growth retardation, hepatosplenomegaly, leg ulcers and skeletal changes resulting from expansion of the bone marrow.

Extramedullary hematopoiesis (EMH) is a recognized complication of ineffective hematopoiesis which results in production of blood elements outside the bone marrow as a physiologic compensatory mechanism. EMH most commonly involves the paravertebral areas followed by the liver, spleen, and lymph nodes.Cauda equina syndrome or spinal cord compression by extramedullary haematopoiesis has also rarely been reported due to development of masses.

This case report reveals a case of 16 year old female with beta-thalassaemia major who presented with bilateral lower limb paralysis secondary to cauda equina syndrome with extra medullary haematopoiesis.

Keywords: Beta- thalassaemia, extra medullary haematopoiesis, cauda equina syndrome

Introduction:

Beta thalassemia major is an autosomal recessive haemoglobinopathy in which there is minimal to no beta globin chain synthesis leading to profound transfusion-dependent anaemia[1]. Inadequate transfusion leads to growth retardation, hepatosplenomegaly, leg ulcers and bone marrow expansion.

Extramedullary hematopoiesis (EMH) with beta thalassemia major is usually secondary to under-treated severe anemia. It can occur in any organ that