Abstract

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A Fortuitous Detection of Composite Heterozygous S/C Sickle Cell Disease

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Background: Hemoglobin SC diagnosis is delayed and performed in adulthood in 29% of cases. The unique pathology of HbSC disease is complex, characterized by erythrocyte dehydration, intracellular sickling and increased blood viscosity.

Case history: A 21-year-old male immigrant from Mali presented to our hospital with abdominal pain. Laboratory tests revealed CRP and LDH elevation and mild hypocromic mycrocitic anemia. CT abdomen and pelvis reported hyposplenia and avascular osteonecrosis of femur heads and lumbar spine. A peripheral blood smear showed abundant red blood cell polychromasia and anysocitosis; capillary electrophoresis of hemoglobin and HPLC revealed double SC heterozygosity; other exams (echocardiography and ocular evaluation) were normal. We made diagnosis of composite heterozygous S-C sickle cell disease. The patient was referred to a second level specialist center for further investigations.

Discussion: Systemic complications include vaso-occlusive crises (65%), retinopathy (35%), aseptic hip osteonecrosis (23%) and splenic infarctions (19%). For people with HbSC, unlike HbSS sickle cell disease, the use of disease modifying RBC transfusions, phlebotomy and hydroxycarbamide are supported by low-level evidence of safety and efficacy. Clinical and translational research is needed to develop targeted treatments and to validate management recommendations for efficacy, safety and impact on quality of life for people with HbSC.

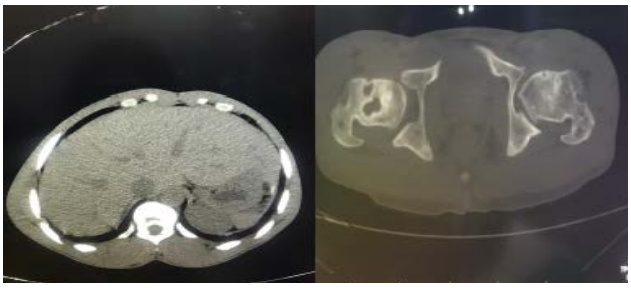

Figure 1 Abdomen and pelvis CT showed a reduction in the size of the spleen (longitudinal diameter 5 cm) and bilateral dysmorphism of the femoral heads (more marked on the right), with lacunar areas alternating with sclerotic areas as from previous necrosis.