P-076 - DIAGNOSTIC CHALLENGE OF DEPOSIT DISEASE IN CASE REPORT

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A 12-year-old female patient, a history of recurrent pain in lower limbs since 3 years of age. At 8 age, she presented pain in the left lower limb, afebrile, and absence of history of trauma or vaccination. Magnetic resonance imaging at the time evidenced acute osteomyelitis in the left distal femur and antibiotic treatment was initiated. Local bone biopsy report presented a discrete inflammatory fibrosing process in cortical bone tissue, with no malignancy or specific process. At 12, she presented recurrence of pain, at this time in the lower right limb, again without fever, traumas or vaccination. Laboratory tests revealed normocytic and normochromic anemia, not responsive to iron replacement, mild thrombocytopenia, without evidence of hemolysis. Magnetic resonance imaging of the right lower limb observed findings compatible with hematological disorders and did not show lytic expansive lesions, blastic, sinus infarct or osteonecrosis. Forwarded to the tertiary service, performed myelogram showed numerous histiocytes with cytoplasm in “crumpled cellophane paper”. The spleen and liver were discretely increased in abdominal resonance examination, there was alteration in second resonance in the right and left hip and thigh, with signs suggestive of avascular necrosis / right infarction. The bone densitometry of the total body column within the limits of normality. Before the myelogram, serum beta-glycoside dosage was below the normal range (0.53 with a reference value of 10-45), and quitotriosity was high (8.457 with reference value). Initiated enzyme replacement therapy 60 IU / kg biweekly! Recurrent limb pain is a common childhood symptom and is found in a variety of common pediatric diseases. Knowledge about rare diseases allows for clinical suspicion, and consequently, earlier diagnosis and treatment, with lower complications for the patient.