QUIZ 1

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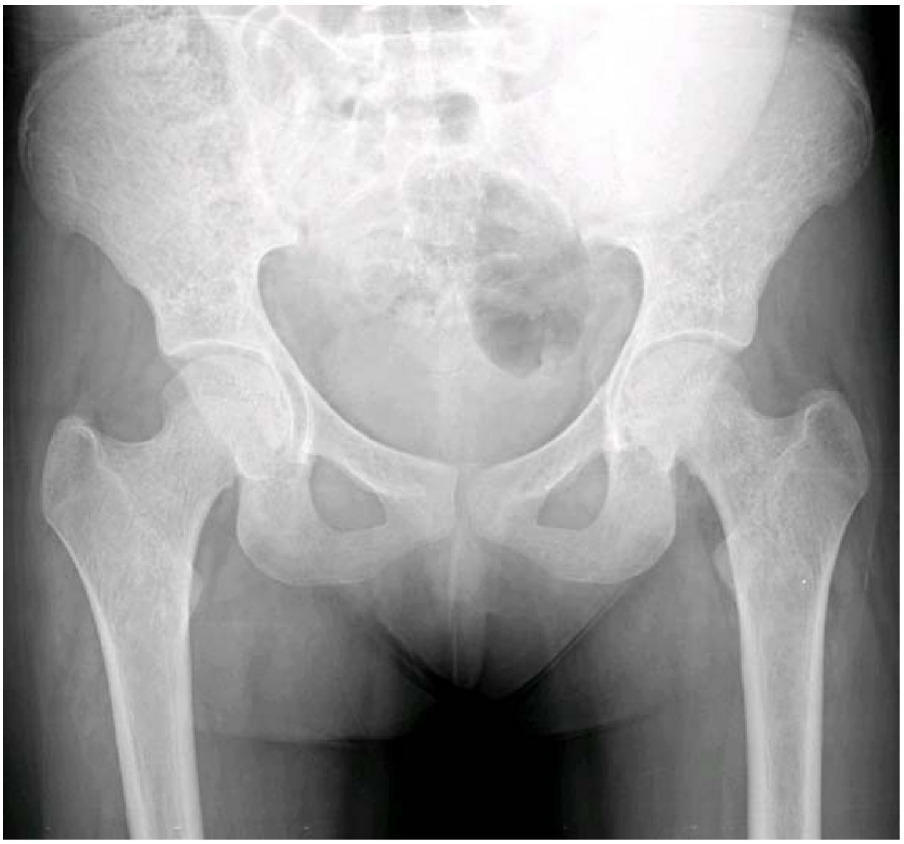
Young female with history of chronic tiredness and lethargy. Now with left hip pain and weakness



Questions

Q1. List the findings.

Q2. What is the likely diagnosis?

Q3. What next exam is indicated?

QUIZ 1

Answers

Answer 1: Splenomegaly
Coarse bone texture.
Osteopenia
Loss of joint space in the left hip.

Answer 2: Haemoglobinopathy, most likely Thalassemia Major with left hip arthritis. Arthritis may be secondary to infection or haemochromatosis.

Answer 3: Specialised imaging of the hip is required. Ideally an MR scan, however in the presence of marrow iron overload may give rise to unacceptable susceptibility effect and therefore a CT may be more useful.

Thalassemia is an autosomal recessively inherited disorder of haemoglobin synthesis. Depending on the gene defect either alpha or beta chains of haemoglobin are deficient. Homozygous Alpha thalassemia is lethal. Homozygous Beta Thalassemia is also called Thalassemia Major and is associated with the most severe abnormalities. These patients are usually transfusion dependent and suffer from consequences of marrow expansion and iron overload. They are susceptible to infection due to leucopaenia from splenomegaly. This predisposes to infections such as septic arthritis. Arthrthropathy is also prevalent especially in patients with long term transfusion therapy due to secondary haemochromatosis and hyperuricaemia.

Patients may present with a variety of symptoms including but not limited to retarded growth, hyperbilirubinemia, hyperpigmentation of skin, hormonal disturbances, bleeding diathesis etc. They usually have severe microcytic, hypochromic anaemia. Iron overload leads to hemosiderosis and myocardial failure.

Typical skeletal abnormalities include diffuse osteopenia with coarse trabeculae, widened medullary spaces, costal osteomas and broad undertubulated ribs. Some patient have prominent facial features with mongoloid or rodent facies.

References

1. Galanello and Origa; Orphanet journal of Rare Diseases 2010, 5:11