Quiz

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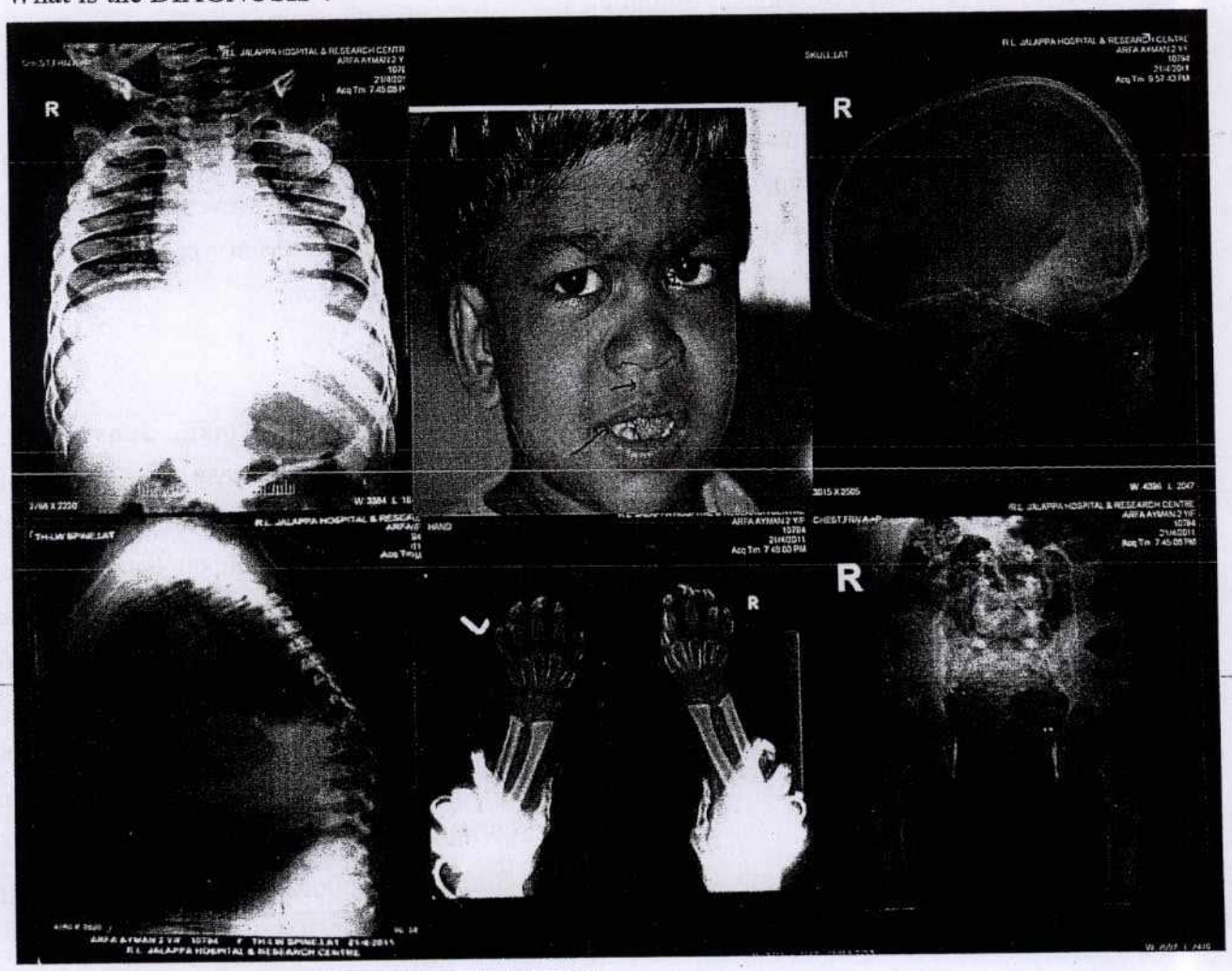
A 4 year old female child presented with history of stuffy nose, repeated upper respiratory infections, deafness and delayed developmental milestones.

On examination she has short stature, coarse facies and other clinical and radiological features as depicted in the photographs.

What is the DIAGNOSIS ?



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HURLERS syndrome - MPS type I

It is a hereditary progressive disease caused by mutation of genes coding for lysosomal enzymes needed to degrade glycosominaoglycons. Failure of this degradation due to absent or grossly reduced activity of mutated lysosomal enzymes results in the intra-lysosomal accumulation. Distended lysosomes accumulate in the cell, interfere with cell function and lead to a characteristic pattern of clinical, radiological and biochemical abnormalities.

Diagnosis is usually made between 6 and 24 months of age with evidence of coarse facial features, corneal clouding, large tongue, prominent forehead, joint stiffness, short stature, skeletal dysplasia, and hepatosplenomegaly. Some patients may present with acute cardiomyopathy. Obstructive airway disease, notably during sleep may necessitate tracheostomy. These patients are prone for repeated respiratory infections.

Most children with Hurler's syndrome acquire limited language skills because of developmental delay, combined conductive and neurosensory hearing loss. Progressive ventricular enlargement with increased intracranial pressure caused by communicating hydrocephalous can also occur.

Radiography shows dysostosis multiplex i.e., thick calvarium, thick and spatulated ribs, ovoid vertebral bodies, enlarged "J" shaped sella, delayed bone age, bullet shaped metacarpals, shallow acetabulum, coarsely trabaculated diaphysis of the long bones with irregular metaphysis and epiphysis along with dentigenous cysts of the teeth. Diagnosis is by characteristic clinical features, biochemical abnormalities, radiological features and molecular diagnosis.

Treatment options are bone marrow transplantation from related or unrelated donors, enzyme replacement using recombinant enzymes. Primary prevention through genetic counselling and tertiary prevention to avoid or treat complications.

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