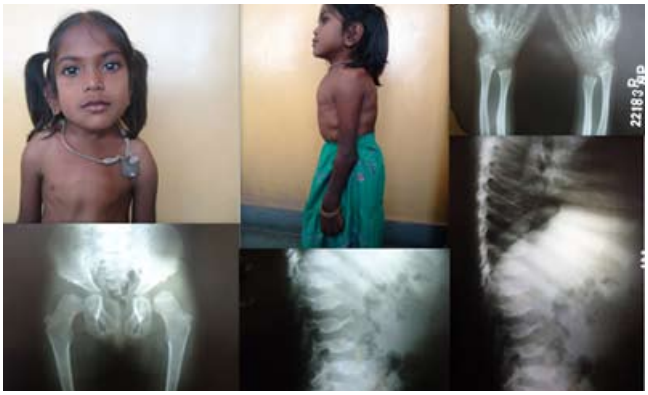
SPOT DIAGNOSIS (IMAGE GALLERY)



SHORT STATURE

K Jagadish Kumar, H C Krishna Kumar, Vinay B S, Karan Raheja, Girish G N, V G Manjunath
Department of Pediatrics, JSS Medical College, JSS University, Mysore, India

Address for Correspondence: Dr K Jagadish Kumar, 85, B,9th Cross , Navilu Road, Kuvempu Nagar, Mysore, Karnataka State, India. E-mail: jagdishmandya@gmail.com in

A 7 years old girl born to a non consanguineously married couple presented to us with chest deformities and short stature (96 cm, expected 119cm, upper segment : lower segment is 0.9:1), short neck, genu valgum and corneal clouding. She had normal development and no organomegaly. Radiological findings revealed biconvex vertebral bodies with reduced height, anterior tongue like projection, widened intervertebral disc spaces with mild degree of generalised osteoporosis of all the bones. X ray hand showed shortened and widened metacarpal bone and phalanges. Metaphyseal ends of radius and ulna were widened with minimal cupping. There was mild shortening of ulna and bowing of radius. Ribs were horizontally placed with widening of distal ends. No flaring of pelvic blades and femoral head is broad. Lungs and cardia were normal.

What is the diagnosis?

Morquio disease. It is also called as Mucopolysaccharidosis IV. There are 2 types of Morquio disease- type A and B. Type A is caused by a deficiency of N-acetylgalactosamine-6-sulfatase and type B by a deficiency of ß-galactosidase. Both result in the defective degradation of keratan sulfate. Both types of Morquio disease are characterized by short-trunk dwarfism, genu valga, kyphosis, waddling gait, fine corneal deposits, small teeth with abnormally thin enamel, frequent caries formation and, occasionally, hepatomegaly and cardiac valvular lesions with preservation of intelligence. MPS IV-A is usually more severe than MPS IV-B, with adult heights of less than 125 cm in the former and more than 150 cm in the latter. Instability of the odontoid process and ligamentous laxity are regularly present and can result in life-threatening atlantoaxial instability and dislocation. Primary prevention through genetic counseling and tertiary prevention to avoid or treat complications remains the mainstay of supportive pediatric care.

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