Mucopolysaccharidosis Type IV (Morquio Syndrome)

A 5-year-old boy was brought to our medical OPD with severe chest deformity and growth retardation. He had six siblings of which two more had similar illness in a 21-year-old sister and 1-year-old brother (Fig. 1). His parents and grand parents had no such illnesses. Clinical examination revealed normal intelligence, truncal dwarfism, pectus carinatum and knock knee. Liver was palpable 3 cm below costal margin in 5-year-old patient. Slit-lamp examination of all the patients did not revealed any corneal opacity. Skeletal survey of all the three patients was done. It revealed pectus carinatum, anterior widening of ribs (spatulate ribs) biconvex vertebral bodies with anterior beaking (Fig. 2), enlarged J-shaped sella turcica, ape-shaped pelvis with gross deformity of femoral heads (Fig. 3). Urine examination of all the three patients tested positive for urinary GAG fragments. Their blood biochemistry was normal. General blood film of 5-year-old boy showed inclusion bodies in lymphocytes (Gasser cells) (Fig. 4). His echocardiography revealed calcified aortic root with thick aortic leaflets.

The mucopolysaccharidoses (MPSs) are a group of diverse inborn errors of proteoglycan catabolism. Catabolites progressively deposit in various tissues and lead to skeletal dysplasia. Short stature is the rule, thick calvaria, an enlarged J-shaped sella turcica, short and wide mandible, biconvex vertebral bodies, odontoid hypoplasia, short thick clavicles, coxa valga, short fingers, wide metacarpals with pointed proximal ends all occur. Many have mental retardation, corneal clouding and organomegaly. The Hurler, Hunter, Morquio and Maroteaux-Lamy syndromes (type IH, II, IV and VI) have short trunk dwarfism. The diagnosis of MPS is made by fractionation of urinary mucopolysaccharides, enzymatic assays and/or molecular genetics. Treatment is palliative and consists mainly of joint replacement and surgical stabilization of cervical instability. Bone marrow transplants have been successful in some forms of MPS.

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