Rheumatologic Manifestations of Lysosomal Storage Diseases

How to Cite this Article: Shiari R, VAdood Javadi P. Rheumatologic Manifestations of Lysosomal Storage Diseases. Iran J Child Neurol Autumn 2012; 6:4 (suppl. 1): 20.

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Lysosomal storage diseases (LSD) result when the lysosome a specific organelle in the body's cell malfunctions. LSDs are caused by lysosomal dysfunction usually as a consequence of deficiency of a single enzyme required for the metabolism of lipids, glycoproteins (sugar containing proteins) or so-called mucopolysaccharides. Since 1955 many LSD have been identified, however, Gaucher, Fabry, MPS I and Pompe disease are the most common of it.

LSDs have a various clinical presentation as same as rheumatologic features. Some of these manifestations are as followings:

Chronic bone pain and/or acute bone crises (with high fever, chills, leukocytosis, increased ESR), Bone marrow infiltration, Osteopenia, Bone remodelling failure (Erlenmeyer flask deformity), Osteonecrosis, Osteolysis, osteosclerosis, Bone fracture (spontaneous), Skeletal growth retardation, hepato and/ or splenomegaly Dysostosis multiplex, Gibbus, deformity, Hip dysplasia, knock knees, Kyphosis, scoliosis, lumbar lordosis, Back pain, Progressive arthropathy with joint pain, stiffness and contractures, Claw hand deformity, Walking capacity decreased, toe walking, Short stature, Thoracic cage abnormalities, and Myopathy.

Early diagnosis in children with LSDs is of particular concern. Common misdiagnoses are Rheumatoid or juvenile idiopathic arthritis, Rheumatic fever, Fibromyalgia / chronic fatigue syndrome, Raynaud's syndrome, Lupus angiokeratomas, Multiple sclerosis, and Growing pains.

Keywords: Lysosomal Storage Diseases, Rheumatology, Bone pain; Arthropathy

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