AN ADULT MUCOPOLYSACCHARIDOSIS TYPE VI CASE PRESENTING WITH QUADRIPARESIS

Y. Zorlu, F. Güleç, B. Özyılmaz, B.I. Tiftikçioğlu, D. Öncel, Y. Zorlu
Neurology Clinic; Genetics Department and Radiology Department, Tepecik Education and Research Hospital Neurology Clinic, Izmir Turkey

Mucopolysaccaridosis type-VI (MPS-VI) is a rare inherited autosomal disease which results from the deficiency of N- acetylgalactosamine 4-sulfatase (arylsulfatase B) activity and the lysosomal accumulation of glycosaminoglycans (GAG), namely dermatan sulfate. MPS-VI typically manifests in early childhood, although slowly progressive forms can rarely reach adulthood. Progressive accumulation of GAGs in connective tissues leads to multisystemic organ involvement causing substantial morbidity. Here by, we report the clinical and neuroradiological findings of a 24-year-old woman presenting with quadriparesis. Detailed examination revealed corneal clouding, hepatomegaly and cardiac valvular abnormalities. Skeletal abnormalities included short stature, dysostosis multiplex, and hypertrophy of ligamentum flavum causing distinctive myelomalasia in upper cervical cord. Diagnosis was confirmed with the increased urine levels of GAG [94,46 mg/lt (237,94 mg/gr kreatinin)]. Definite diagnosis is crucial both for the decision of enzyme replacement therapy and for the attention in anaesthetic interventions during surgical operations to prevent further neurological complications.