Maroteaux-Lamy Syndrome: a case report

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ABSTRACT

Maroteaux-Lamy syndrome is an autosomal recessive lysosomal storage disease, determined by mutations in the arylsulfatase B (ASB) gene located in chromosome 5 (5q13-5q14). The diagnosis is confirmed by the decrease or absence of ASB activity in isolated leukocytes or cultured skin fibroblasts. We report a 16-year-old girl, who was found to have generalized joint stiffness by her mother when she was 2-year- and 6-month old. She had characteristics including short stature, corneal opacity, dysostosis multiplex, and central nervous system manifestations including cervical cord compression caused by thickening of cervical posterior longitudinal ligament. She is considered as a rapidly progressing form of the Maroteaux-Lamy syndrome due to early onset of clinical manifestations and corresponding imaging characteristics. She received cervical decompression surgery from foramen magnum to third cervical spine (C3) and intravenous Naglazyme® infusion for enzyme replacement therapy.