A 3-year-old boy presented with progressive lower extremity weakness and difficulty walking for 2 months. Neurologic examination found the muscle strength of his left lower limb was grade 3 and tendon reflex was weakened. MRI demonstrated an intradural extramedullary mass between T10-11 with multiple and diffuse enhancement of thickened nerve roots and spinal dura (Figure). Juvenile xanthogranuloma was suspected as positive staining for CD68 and negative for S100 and CD1a, while immunoreactivity for ALK did not support it. The genetic test confirmed the diagnosis of ALK-positive histiocytosis with KIF5B-ALK fusion, which is a relatively new subtype of histiocytic disease.1,2

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References


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