E-Poster

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Infantile Spinal Muscular Atrophy in the Physical Medicine and Rehabilitation Department of the YOPOUGON University Hospital. Diagnostic, Monitoring and Therapeutic Management of Infantile Spinal Muscular Atrophy

Topic: Disability Studies

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BACKGROUND

With an incidence of 1 in 6,000, or approximately 120 new cases per year in France, and under-reported in Africa, infantile spinal muscular atrophy (ASA) is the second most common autosomal recessive disease in children after cystic fibrosis. The diagnosis of ASI is most often strongly suggested at the clinical stage. Definitive diagnosis is provided by molecular genetic evidence of an abnormality in the SMN1 gene on both alleles. The frequency of cis-sequence duplications of the SMN1 gene can vary depending on ethnic origin. It is particularly high in Africa. The prognosis is often life-threatening in early-onset forms. Management is coordinated by a referral or specialist center, in collaboration with the primary care physician, around various areas: respiratory, rehabilitation, orthopedic, nutritional, etc.

CASE DESCRIPTION

First Case: A four-year-old patient with a history of delayed psychomotor development. Clinical examination revealed: A spontaneously reducible equinus posture in the lower limbs, a dextroconcave scoliotic deviation with a slight unmeasured hump and thoracic deformity, axial hypotonia with flaccid lower limbs, and significant amyotrophy in both lower limbs. The elektroensefalografi (EEG) suggested diffuse cerebral distress, likely suggesting a cerebral palsy. The elektronöromiyografi (ENMG) was consistent with polyneuropathy. The clinical and paraclinical presentation suggested infantile spinal muscular atrophy.

Second Case: 14-year-old patient with a family history of dermatomyositis and myopathy, and progressive loss of gait function since the age of 1.5 years (stumbling while walking, limping, loss of gait). Clinical examination noted clinical amyotrophy in all four limbs, more pronounced on the right, and muscle strength that varied between 1/5 and 2/5 from one muscle to another. Study of exon 7 of the SMN1 and SMN2 genes (10/31/2018): Typical profile of proximal spinal muscular atrophy. Homozygous deletion of exon 7 of the SMN1 gene. Genotype: 0 SMN1/3 SMN2 (Parental testing could not be performed) The clinical and paraclinical presentation suggested infantile spinal muscular atrophy.

CONCLUSION

The diagnosis of ASI is most often strongly suggested at the clinical stage. Regular monitoring is necessary to identify early impairments (respiratory, neuro-orthopedic, digestive, and nutritional) and to perform preventive or therapeutic interventions before complications become established or irreversible. Monitoring of device tolerance and comfort is also necessary.

Statement of Disclosures

None

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