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SPINAL MUSCULAR ATROPHY: TWO CASES OF RAPIDLY PROGRESSIVE DISEASE

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Introduction: Spinal muscular atrophies (SMAs) are a group of autosomal recessive disorders characterized by progressive lower motor neuron degeneration. Several types have been identified, including the acute infantile form - SMA type I or Werdnig-Hoffman's disease, of which we report two cases.

Case 1: 46-day old male caucasian infant, non-consanguineous parents, no relevant family history; admitted for progressive global hypotonia and poor feeding starting at 2 weeks of life.

On physical examination, the following was noted: supine "frog position"; axial hypotonia; rare spontaneous distal motor activity; bell-shaped chest; abdominal breathing with transient polypnea; flexed toes; clawed left hand; tongue fasciculation. Cardiac assessment was normal.

Laboratory tests showed respiratory acidosis. Transfontanellar ultrasound revealed no abnormalities. Electromyography was consistent with SMA; genetic study confirmed the diagnosis.

Three months later, the infant was readmitted for severe respiratory infection and died due to respiratory failure.

Case 2: 2-month old caucasian male term infant, non-consanguineous parents;

referred to the Pediatric Department at 1 month old for progressive generalized hypotonia (starting at 2nd week), monotone motor activity and abnormal breathing pattern.

Physical examination disclosed: supine "frog position"; clawed hands; head lag; global hypotonia in prone position; absent osteotendinous, Moro, palmar and plantar reflexes; bell-shaped chest; paradoxal breathing. Electromyogram was consistent with SMA, confirmed by genetic study.

Commentary: Werdnig-Hoffman's disease has a poor prognosis, death usually occurring in the first years of life. No curative treatment is currently

available. Early diagnosis is key for both management of supportive care and genetic counseling.

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FETAL ASCITES IN NIEMANN-PICK TYPE C: A RARE AND GRIM PRESENTATION OF THE DISEASE

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Niemann-Pick τύπου C (NPC) is an autosomal recessive sphingomyelin storage disorder with wide clinical spectrum, having however as a common denominator the progressive and fatal neurovisceral degeneration.

The presenting case involves a term female infant with history of fetal ascites that was referred for severe dystrophy and malnutrition.

She is the first born infants of healthy, unrelated parents without family history of inherited disease. At birth she weighted 2590g and had pronounced hepatosplenomegaly and mixed jaundice. She was hospitalized in NICU until two months of age for extrauterine growth retardation.

At admission, she was floppy with excessive ascites and hepatosplenomegaly and concomitant non-obstructive portal hypertension. There were no dysmorphic features Other causes of fetal ascites and metabolic disorders associated with liver disease in infancy were excluded by appropriate investigations, but electron microscopy of liver biopsy revealed findings consistent with NPC, which was confirmed after fillipin stain on cultured skin fibroblasts. There was moderate increase of *chitotriosidase enzyme*. Bone marrow aspirate was normal.

The patient apart from the required symptomatic therapies, the last one year she has been receiving Miglustat, and at the age of 2 years is presenting psychomotor, neurological and biochemical improvement, but continuing to have growth deceleration. Eye movements remain normal.

This case is reported because fetal ascites is a relatively rare clinical presentation of NPC with poor