675

SPINAL MUSCULAR ATROPHY: TWO CASES OF RAPIDLY PROGRESSIVE DISEASE

J. Extreia, I. Monteiro, A.R. Ferreira, S. Rocha, C. Didelet, E. Gonçalves

Hospital Nossa Senhora do Rosário, Centro Hospitalar Barreiro- Montijo, Barreiro, Portugal

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.

676

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

F.-M. Chantzi¹, A. Zellos¹, D. Lazopoulou², H. Michelakakis³, L. Lykopoulou²

¹First Department of Pediatrics, University of Athens, ²First Department of Pediatrics, University of Athens, 'Aghia Sofia' Children's Hospital, ³Institute of Child health, 'Aghia Sofia' Children's Hospital, Athens, Greece

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

prognosis. However, the early use of Miglustat, prior to presentation of the characteristic neurologic signs of the disease, may lead to improvement in life quality.

677

HYPERVITAMINOSIS A - IS IT STILL A CAUSE OF IDIOPATHIC INTRACRANIAL HYPERTENSION IN CHILDHOOD?

T. Jacinto¹, C. Sousa¹, I. Sobreira¹, A.T. Raposo¹, L. Lima², C. Pereira Duarte¹

¹*Pediatrics, ²Ophthalmology, Hospital Divino Espírito Santo HDES - Ponta Delgada, Ponta Delgada, Portugal*

Idiopathic intracranial hypertension (IIH) in prepubertal children is rare and different in presentation from the adult, occurring with no apparent preference for sex or weight. Clinically, strabismus with VI nerve palsy and stiff neck are more frequent, and secondary causes more common. Vitamin A toxicity has been described mainly in adolescents treated with topic retinoids for acne, rather than oral intake as dietary supplement.

We present the case of a previously healthy 9-year-old male, with strabismus, ptosis of the left eye and diplopia about a month after a cold with occipital headache and vomiting (medications included supplements containing vitamin A). Fundoscopy revealed bilateral papilledema with enlargement of blind spot and endotropium of the left eye. Brain CT was normal. Lumbar puncture (LP) at admission revealed opening pressure of 500 mmH₂O, with normal CSF results. Admitting IIH, he was started on acetazolamide, with good clinical results. Regular eye exams revealed visual acuity 8/10 bilaterally and stabilization of papilledema. Elevated serum levels of vitamin A were the only positive findings. He was discharged after two weeks, asymptomatic; completed two months of acetazolamide, then repeated LP with opening pressure of 200mm H₂O.

Hypervitaminosis A has been associated with IIH in children, although its causative role is yet to be determined. A limiting factor is that IIH does not correlate directly with increased vitamin A serum levels, but with CSF levels, which are not tested routinely. Nevertheless, vitamin intake in children should be started only in cases with formal indication and monitored carefully.

678

RECURRENT MENINGITIS AS A COMPLICATION OF SKULL BASE MENINGOCELE

I. Sedighi¹, B. Borzouei², S.T. Sadrosadat², M. Ghodsi³

¹*Department of Pediatrics, Hamedan University of Medical Sciences/ Besat Hospital, ²Hamedan University of Medical Sciences, Hamadan,*

³*Department of Neurosurgery, Tehran University of Medical Sciences/ Shariati Hospital, Tehran, Iran*

Background: Malformations of the skull base are rare anomalies, but can cause severe complications such as meningitis. They are often detected not until after a history of recurrent meningitis.

Methods: A 6-year-old boy was admitted in our hospital due to change in mental status, fever, and vomiting. His past medical history revealed two episodes of bacterial meningitis in 12 and 22 months of age. His parents were cousins and had lost their 3 sons before this boy was born. One of them had died due to respiratory distress 15 days after birth, another one had been born dead, and the last one had died due to meningitis in 3 years of age.

Results: He received Vancomycin and Ceftriaxone upon diagnosis of recurrent meningitis. Due to seizure, intravenous Phenytoin was started. Complementary diagnostic evaluations included computed tomography (CT) and magnetic resonance imaging without any abnormal finding. Streptococcus pneumoniae was isolated from CSF culture. Serum immunoglobulins, complement C3, C4, and CH50 levels, NBT slide test, and flow cytometry (CD3, CD4, CD8, CD19, and CD16+56) were normal. After ruling out primary immunodeficiency, we repeated CT and this time a skull base bony defect behind sphenoidal sinus with a meningocele was noted. Endonasal surgery confirmed this bony defect and closure was accomplished. No further meningitis has been observed.

Conclusions: Congenital skull base defects may be difficult to detect. All children with recurrent meningitis should be investigated for these defects. Early recognition of skull base defects is important to avoid delay of definitive surgical management.