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Zellweger syndrome: Lethal disease by defect of peroxisomal biogenesis-a case report

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The history of peroxisomal disorders, including the most frequent variant, the brain-liver-kidney syndrome of Zellweger, can be divided in four phases. In which the clinical and pathological manifestations of the Zellweger syndrome were explored and delivered. In 1973 it was discovered that it is due to the absence of peroxisomes in hepatocytes and renal tubular epithelial cells. With this discovery, the second phase of which in the years after begun the discovery of various defective peroxisomal functions. During the third phase, which began in 1980, several other peroxisomal disorders were discovered, including Childhood Refsum Disease, Hyperpipecolic Acidemia, Neonatal Adrenoleukodystrophy and Punctata Rizomelica Condrodisplasia. During 1986, the etiology of the different peroxisomal diseases was identified through complementation studies, which marked the beginning of the fourth phase of the history of peroxisomopathies. It is a recessive autosomous transform that is presented in a proportion of 1 in every 50,000 individuals characterized by a multifunctional progressive deterioration of terminal systems with fatal dissolution in average before 12.5 months of age. In addition to the hypotony and craniofacial dysmorphism, skeletal abnormalities such as equinovarus talipes, rotation of the thumb, calculated spots in the heads and acetabululo mayor, identifiable radiographically, constitute a fundamental distinctive diagnostic trait. Frequently there are cataracts, optical nerve atrophy or pigmentary retinosis. Periportal Fibrosis produces hepatomegalia and cholestatic ictericia. Ultrasonographic recognized renal cortical cysts are formed. Neurological alterations emergen early, with psychomotrized delay and early appearance of convulsive crises.

Description: The case of lower-female infant of 3 months ages is presented, who is born in this institution, presenting to the birth poor respiratory effort, requiring advanced maneuvers of neonatal resuscitation and entry to neonatal intensive care unit, ala physical exploration with facial dismorphism, bridge wide nassal, generalized hypotonia, metabolic acidosis with persistent hyperlacytatemia

Clinical Case: Female infant of 67 days of age, of non-consanguine parents and a brother deceased by meningitis at year of age. Controlled second gestation product, normoevolutive mother of 37 years old, iterative cesarea at term with Apgar 7/9 presents poor respiratory effort, so requiring advanced maneuver of neonatal resuscitation. Weight: 3.3 kg, persistent hypoactivity, scasa suction, oliguria. It is revised at 15 days of life, by neuropediatrie, observing hipoplasia of the bones of the crane, broad fontanelles, weak suction, hypoglycemia and generalized hypotonia. The exploration was identified fontanel anterior amplia, communicated with the rear and lateral, sagittal and metropic diastased sutures, prominent front, prominent supra-orbital edges, wide nasal root and optic papal paper, added to marked global hypotony, hepatomegalia and metacarpofalangicas joints and ró-hyperlax tubes. Clinical evolution becomes unfavorable: progressive neurological depression, epileptic crises, clinical polymorphic classic focal, axial tonics and mioclonies, initiating levetiracetam 30mgkgdi. Added gastro-esophageal reflux and deglutorium disorders with apical pneumonia and hypocromic microcitia anemia 9 g of hb that almost hemo-transfusion alos is performed negative metabolic squeeze negative, profile for negative mucopolisacaridosis, comes case with an innate metabolism error center who requests to take cariotipo, in the metabolic study carried out by the research laboratory of its foundation, at the stauros children's clinic in Barcelona, Spain, the following findings were obtained: the very long chain acids (agcml) in the plasma and in the hematíes were increased significantly. Pristanic acid was slightly high: 3.14 nmol/ml (<1.0) and phytanic acid, within the normality: 0.58 nmol/l (<5.0). plasmalogen levels were very low in hematies (16: 0dma/16: 0 and 18: 0dma/8: 0, 0.016 and 0.040, respectively). Docosahexaenoic acid (DHA, 22: 6n-3) was much reduced in plasma (37, 18) and something less, in erythrocytes (21, 47). What is confirmed diagnosis, is transferred to second level institution continue with handling where the sequential shock presents and subsequently dyes the specific biochemical diagnosis and the expert advisory in the handling.

Conclusion: The purpose of presenting this case is to allow pediatric and neonatological physician to acquire a unified syndromic perspective, common to the observation of new hypothenic births, intendedly looking for distinctive clinical traits common to peroxismal disorders. Still does not have a prenatal practical diagnosis form or in the habitual metabolic screening in the newborn baby.

Biography

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