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Case Report

Maple Syrup Urine Disease

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Abstract

This case report describes a 7-day-old male newborn, product of a normoevolutive pregnancy, born at term without complications with a basic newborn screening reported without alterations. He is brought to the emergency room due to weak suction and generalized weakness. On arrival, he is in an altered state of alertness, with generalized hypotonia and abnormal primitive reflexes. A hypotonic syndrome with central nervous system alteration is integrated, resulting in a diagnosis of neonatal encephalopathy. In the initial approach, hypoglycemia and infectious causes are ruled out. Image studies reveal findings compatible with hypoxic-ischemic encephalopathy, however, there is no history of perinatal asphyxia or cranioencephalic trauma. In addressing metabolic causes, serum electrolytes are normal. Further investigation revealed metabolic acidosis with an elevated anion gap and elevated levels of the amino acids valine, leucine, and isoleucine, consistent with Maple Syrup Urine Disease (MSUD). The patient was treated with dietary restriction of these amino acids and made a recovery. This case highlights the importance of considering metabolic disorders in the differential diagnosis of neonatal encephalopathy, and the potential utility of expanded metabolic newborn screening.

Keywords: Neonatal encephalopathy; Hypotonic syndrome; Inborn errors of metabolism; Newborn screening

Introduction

7-day-old male newborn, product of a second normoevolutive pregnancy with adequate prenatal care, born at 39 weeks of gestation via cesarean section due to lack of labor progression, with Apgar 8/9, Silverman-Anderson 0-0, with an appropriate weight for gestational age, rooming-in with his mother and discharge without any complication. With the result of an auditory, cardiac and basic newborn screening of 5 elements reported without alterations.

He is brought to the emergency room due to presenting weak suction and generalized weakness. On arrival, he is observed to be lethargic, with generalized hypotonia and abnormal primitive reflexes. The diagnosis of neonatal encephalopathy is integrated.

Clinical Findings

Without any background for the current condition, there is a newborn patient on his seventh day of life, with a two-day history of progressive weakness, starting with weak suction reported by the mother as "less breastfeeding time," progressing to generalized weakness with decreased movement of all four limbs and weak crying.

Upon arrival, the patient is well-colored, without respiratory difficulty, with a flaccid position of all four limbs. On physical examination, there are no evident abnormalities. The cranial suture is symmetrical, without evidence of fontanel bulging or asymmetry.

The eyes are symmetrical, without exophthalmos or strabismus. The ears are symmetrically placed, without deformities. The chest is symmetrical, with good expansion and no retractions. The heart sounds are regular, with a rate of 150 bpm, and there are no murmurs or gallops. The abdomen is soft, non-tender, non-distended, and without masses.

On neurological examination, a hypoactive patient is observed, with spontaneous eye opening, however, minimal response to external stimulus is present, not alert to sound or visual stimulation, classifying its neurological state as lethargy.

On motor neurological examination, a generalized decrease in muscle tone is observed, with spontaneous and symmetric movements that do not overcome gravity (force 2/5) of all four limbs. Upon ventral suspension, the patient is observed in an inverted U position, and upon axillary suspension, the patient slides from the hands of the examiner, demonstrating a hypotonic syndrome.

Primitive reflexes are altered: weak sucking reflex, weak plantar and palmar grasp reflex and absent Moro reflex.

A hypotonic syndrome with central nervous system alteration is formed, categorizing it as neonatal encephalopathy, with neurological deterioration and apneas, it is necessary to secure airway with orotracheal intubation [1-3].

Diagnostic Approach

The most common cause of neonatal encephalopathy is hypoxic-ischemic encephalopathy secondary to neonatal asphyxia. However, in the patient's history, there are no perinatal complications. Therefore, the next things to rule out are infectious causes such as neonatal sepsis or neurological infection, structural causes such as ischemia or cerebral hemorrhage, or metabolic causes.

In his approach, laboratory tests are requested, finding a conserved hematic biometry and a negative PCR, ruling out sepsis as a cause of encephalopathy. A biochemical profile is requested, finding preserved renal and liver function, with normal electrolyte levels. However, the blood gas analysis shows metabolic acidosis with an elevated anion gap, leading to suspicion of a metabolic cause.

An MRI is requested, observing changes in density affecting corticospinal tracts, ventrolateral thalamus, internal capsule, mesencephalon, brain steam and cerebellar hemispheres, bilaterally and symmetrically, as well as lack of diffusion, translated as cytotoxic damage of normally myelinated white matter in a newborn, changes commonly observed in hypoxic-ischemic encephalopathy and metabolic diseases due to accumulation of metabolites.

In suspicion of an inborn error of metabolism, an extended metabolic newborn screening is decided upon, observing elevation of certain essential amino acids. Serum amino acids are requested, finding an important elevation of leucine, isoleucine, and valine, confirming the diagnosis of Maple Syrup Urine Disease [4-6].

Therapeutic Approach

The disease is caused by an enzymatic deficiency in the activity of the branched-chain alpha-ketoacid dehydrogenase complex (BCKDC), which is an enzyme in the metabolic pathway of branched-chain amino acids that results in accumulation of these amino acids and cytotoxic damage to myelinated areas in the central nervous system. The nutritional approach is to restrict the intake of these amino acids that are found in breast milk and in almost all lacteal formulas.

Initially, in this patient's case, as there was no availability of an amino acid-free formula, it was decided to restrict protein ingest restricting oral formula intake and supplementing with parenteral fluids with normal requirements of glucose and lipids [7,8].

Follow-Up and Outcome

With the nutritional management provided, the patient showed improvement in the neurological state, and could be extubated and fed orally with the appropriate formula.

Currently, the patient is at home with close monitoring by pediatrics and genetics, fed with an amino acid-free formula.

Discussion

Maple Syrup Urine Disease is a rare entity, with an estimated incidence in the general population of 1 in 185,000 live births, however, it is potentially lethal.

It is a disease that could be detected in the metabolic screening, however, in the Mexican general population that does not have access to private medical service, a basic metabolic screening is taken that includes five diseases.

This means that the disease may not be detected in the general population if they only undergo the basic metabolic screening which only include five diseases: congenital hypothyroidism, galactosemia, phenylketonuria, congenital adrenal hyperplasia, and biotinidase deficiency. It is important to note that the expanded metabolic newborn screening should be considered in cases of atypical presentation of neonatal encephalopathy or any other metabolic disorder that could be suspected by the clinician. Early detection and treatment are crucial for the outcome of patients with Maple Syrup Urine Disease.

Acknowledgement

None.

Conflict of Interest

No conflict of interest.

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