Prolonged Icterus in the Course of Galactosemia
Presentation of a Case

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Abstract

Male patient of 7 months admitted to the Pediatric Service of the Institute of Gastroenterology, for yellowing of the skin and mucous membranes, diarrhea and weight loss, as a result of a third pregnancy of parents without a degree of consanguinity, delivery at term, physiological, weight and size appropriate for gestational age, APGAR 9/9; He presented prolonged icterus without apparent complications. A history of multiple admissions due to complicated diarrheal diseases.

Among the clinical signs of interest, the yellowish color of the skin and mucous membranes is remarkable, as well as a marked decrease in the panniculus adipose, the globular abdomen and the hepatomegaly of 5 cm of fine edges. In the interrogation, made to the mother, it was known that the symptoms began with the introduction of breastfeeding, characterized by vomiting and diarrhea. Metabolic disease was suspected and it was consulted with the Hospital Pedro Borras Nutrition, indicating a urine test in which galactose appears, later it was indicated, quantification of galactose in blood that showed values of 20 mg/dl and enzymatic activity whose values were 4.3 u/gHb, conclusive of galactosemia.

Dietary treatment was indicated, excluding dairy products and their derivatives, appreciating clinical, anthropometric and biochemical improvements.

In the follow-up controls, elevated levels of pyruvic transaminase (30 IU) were found, which motivates their re-entry, detecting a transgression in the diet. Adjustment of the daily intake is made, with a satisfactory evolution.

Keywords: Galactosemia; Enzymatic Activity; Dietary Therapy Treatment

Introduction

Galactosemia, is a congenital error of carbohydrate metabolism that affects approximately 1/40,000 to 1/60,000 live births in the world, its clinical presentation if it is not diagnosed in the first days of birth, it can be serious and sometimes mortal. It is caused by a disorder of the metabolism of galactose, accumulating in different tissues of the body such as the lens, liver, kidneys and brain [1-5].

The first symptoms begin a few days after birth, with the introduction of breastfeeding, characterized by vomiting, diarrhea, prolonged icterus, weight loss, hypoglycaemia crisis and delayed psychomotor development.

Mortality is high due to its difficult diagnosis and almost always occurs during the first year of life [4,6-9].
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The levels of total bilirubin in blood at birth are found in values of 5 mg/dl; when these values are exceeded after 24 hours of birth, they are considered pathological and can rise to more than 12.9 mg/dl in children born at term and 15 mg/dl in pre-term terms.

When rising bilirubin levels in the blood at birth is called cholestasis in the newborn and are classified as: Early: when it is evidenced before 24 hours and the cause is due to hemolytic anemia due to RH incompatibility. Intermediate, from 24 hours to 10 days due to ABO hemolytic anemia; Jaundice from breast milk; hypothyroidism and infections. Late: after 10 days; hyperbilirubinemas of direct predominance type; (hepatitis A, B, C and biliary atresia. 27, In galactosemia, it is produced by accumulation in the liver tissue of galactitol (toxic metabolite product of galactose metabolism).

Diagnosis
a. Clinical
   • Symptoms and signs
   • Anthropometric study
b. Laboratory data
   • Thin layer chromatography of carbohydrates in urine.
   • Quantification of galactose in urine and blood
   • Enzymatic study [3,6-12].

Case Presentation
A 7-month-old male patient of non-consanguineous parents, the result of a third pregnancy, which had a normal nutritional status with no associated pathology. Physiological delivery at term with weight and height suitable for gestational age; He presented with the baby at birth that was prolonged and did not remit with the use of a fluorescent lamp. Subsequently, the vomiting and diarrhea symptoms were established, coinciding with the introduction of breastfeeding, evolving to metabolic acidosis with hydromineral imbalance; Septic shock in several admissions in their province of origin [2,4,5,9]. By decision of the parents is transferred to the Capital entering the Pediatric Service of the Institute of Gastroenterology for study.

Interview with Nutrition of the Hospital Pedro Borras for suspected metabolic disease [13-21].

Physical exam
Marked reduction of the adipose panniculus, as well as, yellow color of the skin and mucous membranes, globular abdomen, not painful on palpation, hepatomegaly of 5 cm. not painful of fine edges, normal psychomotor development.

Weight: 4.3 Kg Size: 65 cm.

Assessment of the nutritional status when compared with the reference values of the Cuban tables: Protein energy malnutrition by default.

General and specific complements were indicated to determine liver function with TGP and OGT values, which were pathological. Urine sample was taken from the first urination in the morning by sending it to the specialized laboratory of Medical Genetics for the diagnosis of metabolic diseases finding reducing sugars in urine (galactose), it was decided to take a sample of whole blood to which perchloric acid was added for the quantification of galactose in blood giving values of 23 mg/dl and also whole blood with heparin for the determination of the activity of the enzyme galactose-1-phosphate-uridyltransferase with figures of 4.3 u/gHb, confirming the diagnosis of Classical Galactosemia [4,22-35].

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Treatment therapeutic treatment is initiated taking into account the needs for their age and sex, excluding from the diet the dairy foods and their derivatives with improvement of the clinical and anthropometric situation and therefore they are discharged. 6, 8, 9, 12 Weight/Age: 50/75 P Size/Age: 25/50 P Weight/Size 50/75 P TGP: 8 IU.

In ambulatory follow-up, high figures of pyruvic glutamic transaminase (30 IU) were found, which led to re-entry to the hospital, detecting in the interrogation of the mother that there had been a transgression in the diet with an increase in fats over the recommendations and among them the use of saturated fats. 30 Adjustment of the diet is made with a satisfactory evolutionary commentary.

Discussion and Conclusion

The case we describe is that of a patient with prolonged icterus who, when performing specific complements, was concluded as a diagnosis of the end point of classical Galactosemia to G-1-P-UT enzyme deficiency, whose clinical manifestations began with the introduction of breastfeeding characterized by prolonged ictero, subsequently establishing diarrheal disease that evolved into septic shock and affectation of nutritional status, (energy protein malnutrition by default).

Timely therapeutic treatment was imposed by eliminating dairy foods with clinical, anthropometric and biochemical improvement [2-4,8,12,14]. The ambulatory monitoring controls show levels of galactose in blood of: 1.5 mg/dl and TGP: 8 IU, values within normal limits, their nutritional status according to Weight/Size is: 50/75th percentile. Classifying Normopeso. If galactosemia is not diagnosed in the early stages of life, the intensity of the disease increases as the diagnosis is prolonged.

Early therapeutic treatment is the fundamental pillar in the remission of symptoms and signs, as well as the prognosis [4,8,13,14,18,26,28,36-39].

Bibliography


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