IMAGING CASES

BIOCHEMICAL CLINICAL CASE

CASO CLÍNICO BIOQUÍMICO

Joana Silva¹, Joana Ferreira², Mariana Silva², Miguel Costa²

A nine-month-old infant girl, previously healthy and with unremarkable family history, was referred to the Pediatric consultation due to elevated aminotransferase identified while investigating failure to thrive.

In the first assessment, although the patient achieved the original birth weight percentile, she kept hypertransaminasemia. Physical examination was normal. Complementary study found a bicuspid pattern in albumin fraction on serum electrophoresis (Figure 1). Total albumin variation was within the normal range.

What is your diagnosis?

Figure 1 - Serum protein electrophoresis showing two peaks in the albumin region

References:
1. Department of Pediatrics, Centro Hospitalar Entre Douro e Vouga. 4520-211 Santa Maria da Feira, Portugal. nessajoana@gmail.com; cliromi@gmail.com
2. Department of Clinical Pathology, Centro Hospitalar Entre Douro e Vouga. 4520-211 Santa Maria da Feira, Portugal. jbeatriz81@hotmail.com; marianaspisiva@gmail.com
DIAGNOSIS

Bisalbuminemia

DISCUSSION

Assessment of renal function, total proteins, immunoglobulins, pancreatic enzymes, and thyroid function and abdominal ultrasonography were requested to exclude secondary causes of bisalbuminemia. Complementary studies showed no changes, and no therapy was instituted.

Given suspicion of a hereditary disorder, other family members were tested through serum protein electrophoresis, with the condition only confirmed in the mother.

During follow-up, the patient remained clinically asymptomatic and liver enzymes returned to the normal range, but no overt cause of hypertransaminasemia was identified.

This case describes bisalbuminemia findings in two family members, confirming its inherited nature. Genetic study has not yet been performed, as it has no implications in clinical management.

Bisalbuminemia may be inherited (or permanent) or acquired (or transient). Inherited bisalbuminemia has a frequency between 1:1000 and 1:10000 and may have an autosomal dominant form, being frequently found in several members of the same family. The causative genetic lesion is a point mutation of human serum albumin gene, and more than 100 variants have been identified. Slow-type variants predominate in Europe. The modified albumin form generally has no pathological significance, but some albumin variants may have altered affinity for some hormones, metal ions, fatty acids, and drugs, with clinical implication in some cases. That is the case of familial dysalbuminemic hyperthyroxinemia and familial dysalbuminemic hypertiroidothyroninemia, which have been linked to inherited bisalbuminemia. Mutations involved form a protein with preferential L-thyroxine or triiodothyronine affinity, resulting in increased total serum levels. Bisalbuminemia diagnosis is established by first eliminating the main acquired etiologies: drug interference (mostly high doses of beta lactam antibiotics), acute pancreatitis, and binding of monoclonal immunoglobulins. The present report described a rare case of hereditary bisalbuminemia in an infant after excluding other causes and investigating family members. Although this condition seems to have no clinical implications, it should be acknowledged to ensure the best management of these patients.

ABSTRACT

Bisalbuminemia is a qualitative albumin variation defined by coexistence of two types of serum albumin with different electrophoretic mobilities in the same individual. It can be of two different types: hereditary (or permanent) and acquired (or transient).

Herein is described a rare case of hereditary bisalbuminemia in a healthy infant, incidentally found during elevated aminotransferase study. Despite not having pathological significance, acknowledgement of this analytical alteration is key for adequate management of these patients.

Keywords: blood protein disorder; electrophoresis; serum albumin

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CORRESPONDENCE TO
Joana Silva
Department of Paediatrics
Centro Hospitalar de Entre Douro e Vouga
Rua Dr. Cândido Pinho 5
4520-211 Santa Maria da Feira
Email: nessajoana@gmail.com

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