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Research Article

AXIAL HYPOTONIA INDICATIVE OF DILATED CARDIOMYOPATHY FOLLOWING PRIMARY CARNITINE INSUFFICIENCY

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Abstract

Primary systemic carnitine deficiency is a potentially lethal fatty acid oxidation disorder. It is usually characterized by cardiomyopathy occurring in early childhood, often associated with weakness and hypotonia, failure to thrive, and recurrent hypoketotic hypoglycaemic attacks. Three tissues / organs are affected in the primary carnitine deficiency: the heart muscle which causes progressive cardiomyopathy; central nervous system which is affected by encephalopathy due to hypoglycemia.

Keywords: Hypotonia, Dilated Cardiomyopathy, Carnitine.

INTRODUCTION

Primary systemic carnitine deficiency is a potentially lethal fatty acid oxidation disorder. It is usually characterized by cardiomyopathy occurring in early childhood, often associated with weakness and hypotonia, failure to thrive, recurrent hypoketotic hypoglycaemic attacks, and / or coma. Primary carnitine deficiency occurs in 1 to 5 per 10,000 inhabitants and most often manifests between 1 and 7 years (Koizumi A et al). We report a case of axial hypotonia indicative of dilated cardiomyopathy secondary to primary carnitine insufficiency

Clinical case

Infant of 1 month who presented in a progressive way of breathing disorder associated with a fall of the head for which they consulted a hospital center in a health center of the place where the present transfer was signed for appropriate management.

Antecedent

Antenatal: pregnancy followed with four antenatal consultations, VAT, SF AF 5 (+), HBsAg negative.

Prenatal: Vaginal birth at 40-week term with early ultrasound, clear amniotic fluid.

Post natal: shouted after birth, no concept of resuscitation

Birth weight: 3300g

Family: mother aged 39, 6G5P, abortion 1, without particular defects, GsO Rh +, hemoglobin at 12.2 no notion of consanguinity

Vital parameters

Temperature: 36.3 ° C, heart rate at 167 beats per minute, respiratory rate at 76 cycles / minute, oxygen saturation 71%, blood sugar at 1.29? TA: 103/63.

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Measurements: waist at 63cm, MUAC and cranial perimeter.

General physical examination: Good general condition, good hydration, Isochoric and reactive pupil.

Neurological examination

Conscious, do not hold up, axial hypotonia associated with hypertonia in the 4 limbs, no oculomotor damage, sharp tendon osteo reflex in the 4 limbs, preserved nociceptive sensitivity, soft neck.

Cardiovascular examination: audible heart sound, tachycardia without noise on added.

Pleuropulmonary: dyspneic breathing, presence of bilaterally crackling rales. Other devices without special features

Syndromic summary:

1. Fiasco spasmodic hypotonia syndrome
2. Tachycardia
3. Dyspnea

Hypothesis: infectious or metabolic encephalopathy
Normal brain scan, the ideal would be MRI
Electrocardiogram: sinus tachycardia

Cardiac ultrasound: dilated cardiomyopathy, with birth defect and left coronary pathway and parachute mitral valve with interatrial communication ostium, secondum. Normal ionogram blood test, white blood cell count at 17,000. The negative reactive -c protein. Determination of free carnitine at 24umol / L normal value (29-43). Total carnitine dosage: 33umol / L Normal value (40-56).

Conclusion: Reduction of free carnitine and total carnitine. Final diagnosis:metabolic encephalopathy on dilated cardio myopathy on carnitine insufficiency. Therapeutic management: treatment of heart disease and symptomatic. Physiotherapy with clinical improvement.

DISCUSSION

Primary carnitine deficiency is a rare autosomal recessive disease of fatty acid oxidation caused by a deficit in the transport of carnitine in the plasma membrane resulting from an alteration of the carnitine transporter (organic cation carnitine transporter 2) in the body. Plasma membrane. This deficiency limits tissue absorption, leading to decreased accumulation in the heart and skeletal muscles and potentiates an increased renal loss of carnitine (Burwinkel B et al) leading to systemic depletion of carnitine (Reuter SE et al. al,). The most common presentation of primary carnitine deficiency is hypoglycaemic hypoketotic encephalopathy. Cardiomyopathy has also been observed (Erguven et al). The gene responsible for Primary carnitine deficiency is SLC22A5. Three tissues / organs are affected in the primary carnitine deficiency: the heart muscle which causes progressive cardiomyopathy; central nervous system which is affected by encephalopathy due to hypoketotic hypoglycemia and skeletal muscle affected by myopathy (Erguven M et al) Human skeletal and heart muscles contain relatively high concentrations of carnitine from plasma because they are incapable of biosynthesis carnitine (Kendler BS). The heart is one of the organs most affected by carnitine-acylcarnitine deficiency (Palmieri F). Cardiomyopathy, cardiac arrhythmia (probably due to the accumulation. Long chain fatty acids and acylcarnitines which cannot be oxidized), heart failure and respiratory distress result from a deficiency of carnitine-acylcarnitine (Palmieri F). For these patients, L-carnitine supplementation is a lifesaving treatment. Three distinct clinical entities have been described; the adult, infantile and perinatal, all with an autosomal recessive mode of inheritance (Sigauke E et al).The different mutations in SLC22A5 probably give rise to differences in disease intensity / onset. The measurement of free carnitine and total carnitine in plasma is important in the diagnosis of this disease.

Conclusion

Pathology of lethal fatty acid metabolism. Characterized by skeletal and muscular involvement. It manifests itself by a heart disease associated with a neurological disorder which can jeopardize the vital and functional prognosis of the patient. Always asks for the dosage of carnitine in front of any clinical picture associating a heart disease and neurological damage.

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