CASE REPORT: CONGENITAL HEART DEFECT IN THE CHILD OF A MOTHER WITH PHENYLKETONURIA

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INTRODUÇÃO:

Hyperphenylalaninemias (HPA) are inborn errors of metabolism, inherited autosomal recessively, whose primary disorder is located in the conversion of the amino acid phenylalanine into tyrosine due to deficiency of the hepatic enzyme phenylalanine hydroxylase (PAH). As a result, there is an increase in the concentration of phenylalanine and its byproducts in the blood and urine (phenylpyruvate, phenylacetate, phenyllactate and phenylacetylglutamine), with reduced formation of tyrosine.

Maternal phenylketonuria is an aminoacid pathology characterized by elevated plasma levels of phenylalanine in the pregnant woman that may cause abnormalities in fetus development, and which is called maternal phenylketonuria syndrome. This syndrome manifests itself in the fetus through intrauterine growth restriction (IUGR), microcephaly, mental retardation and congenital heart malformations4

Females with CNF or non-CNF hyperphenylalaninemia should receive special guidance after menarche regarding contraceptive methods and pregnancy planning. A phenylalanine-restricted diet, associated with the use of a metabolic formula, in the case of patients responsive to sapropterin dihydrochloride, should be established before and during pregnancy in order to avoid CNF embryopathy or maternal CNF syndrome. Phenylalanine levels during pregnancy should be strictly monitored and the fetus should be monitored, particularly with regard to microcephaly and/or congenital cardiac anomalies.

DESCRIÇÃO DO CASO:

ASA, 21 years old, diagnosed with phenylketonuria by newborn screening. Family from Manaus. Non-consanguineous parents. Poorly adherent to treatment. On 9/21/2023, she attended an appointment at 20 weeks of pregnancy. PHE at that time = 14.8. Referred to the high-risk prenatal clinic. Normal fetal echocardiogram. PHE evolution: - 9/29/2023 = 10.4 mg/dl; 10/17/2023 = 11.5 (appointment with her partner); 11/13/2023 = 6.1; 11/28/2023 = 5.9

There was no opportunity to perform the sapropterin test

Delivery: 12/27/2023. Newborn: Weight = 1.975 ; Length = 42 ; Head circumference = 28.5 (Z-score -2.6). Apgar = 8/9. Gestational Age = 35 w. Echocardiogram: small ventricular septal defect without hemodynamic repercussions. Exclusive breastfeeding for only 1 month. Newborn screening PHE=1.1 mg/dl. Evolution with recurrent respiratory infections and failure to thrive. Currently head circumference Z-score=- 2. Slightly delayed neuropsychomotor development

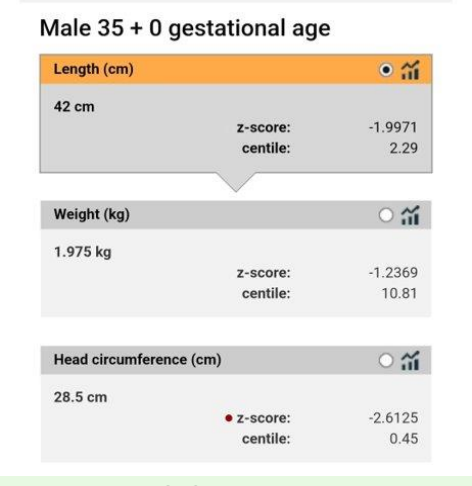

Figure 1 - INTERGROWTH-21st calculator

DISCUSSÃO E COMENTÁRIOS FINAIS:

Women with phenylketonuria must be properly informed so that they can plan their pregnancies and monitor their phenylalanine levels before conception. The care team must make continuous efforts in this regard. The sapropterin test is recommended by the Ministry of Health and should be performed on all adolescents with the condition.

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