12º COBRAPEM
Congresso Brasileiro Pediátrico de Endocrinologia e Metabologia
31 de maio a 03 de junho de 2017
Rio de Janeiro . RJ

Trabalhos Científicos

Título: Transient Clitoromegaly In Newborns Heterozygous For Cyp21A2 Mutation Detected In The Congenital Adrenal Hyperplasia (Cah) Neonatal Screening

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Resumo: Neonatal screening (NS) for CAH due to 21-hydroxylase deficiency most finality is the diagnosis of severe salt-wast adrenal insufficiency and virilization. Mild cases are often missed by this strategy. CYP21A2 heterozygous patients should not have signs of the disease. Objective: To describe 3 cases of CAH NS Transient Clitoromegaly in Newborns Heterozygous for CYP21A2 Mutation detected in the CAH NS, following the National Newborn Screening Strategy and genotyping by SnapShot multiplex minisequencing. Cases Descriptions: Case 1. 17 OHP of respectively 35.3 and 10.4 ng/mL in sample 1 and 2, birth weight (BW) 2,504g, genotype p.V281L/WT; Case 2. 17 OHP of respectively 22.4 and 16.7 ng/mL in sample 1 and 2, BW 2,560g, genotype p.V281L/DUPCYP21P; Case 3. 17 OHP of respectively 18.2 and 35.0 ng/mL in sample 1 and 2, BW 2,905g, genotype IVS_T/WT. There was no history of consanguinity or salt lost in none of the cases. Comments: CYP21A2 heterozygous patients should not have early signs of the disease. The p.V281L mutation is commonly associated to non-classical CAH (NC-CAH). The description of three cases with transient clitoromegaly is worthy of additional study. It has been reported that some heterozygous patients had higher androgen levels, especially carriers of p.V281L mutation with premature adrenarche. It is expected that NC-CAH and heterozygous patients are asymptomatic at birth, but we hypothesized that possible transient androgen elevations may occur.