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: Neonatal Screening Of Congenital Adrenal Hyperplasia (Cah): Results After 24 Months Of Its Implementation In The Public Health System Of Rio Grande Do Sul (Rs), Brazil

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Resumo: CAH is caused by the deficiency of the enzyme 21-hydroxylase in 90-95% of cases and leads to the accumulation of 17-hydroxyprogesterone (17OHP). Early diagnosis in neonatal screening (NS) allows adequate treatment and reduces mortality. Objective: To describe CAH NS results in the public health program of the state RS. Methods: The database of the local Reference Service was analyzed and the newborns (NB) with suspected CAH by changing the 17OHP, measured by fluoroimmunoassay technique and adjusted for birth weight (BW) were selected. The 17OHP cutoff levels used were those recommended by the CAH NS National Program for 4 birth weight (BW) ranges. Classic CAH (salt wasting and simple virilizing) was diagnosed by an increase in 17OHP confirmed in the retest, by clinical evaluation and genotype done by PCR, Snap-Shot and MLPA (Multiplex ligation-dependent probe amplification). Results: in the 1st year the positive predictive value (PPV) of the initial screening was 1.6% and an overall rate of false positives (FP) was 0.47%, higher among infants under 2,000g BW. After 24 months, 15 cases were diagnosed, from a total of 217,965 NB, with an estimated incidence of 1:14,531, with a good genotype-phenotype correlation. The most frequent mutation was IVS2-13A/C>G. Conclusion: The results underscore the need for CAH neonatal screening in the public health system and show that the strategy adopted was appropriate. Incidence, PPV and FP results were similar to those reported by other states of Brazil.