P-202 - CONGENITAL ADRENAL HYPERPLASIA; DIFFICULTIES IN THE NEONATAL DIAGNOSIS.

Bastida MG, Garcia V, Alvarez RG, Avila S

“Dr. E Castro Rendón” Hospital. Neuquén Province. Argentina. mgbastida@yahoo.com

INTRODUCTION: Two cases about girl’s brothers with Congenital Adrenal Hyperplasia (CAH), whose mothers were prescribed with corticoids in the first months of their pregnancies which could have altered the 17-hydroxyprogesterone (17OHP) values in the newborns. OBJECTIVES: Report the difficulties that may occur in the 17OHP results interpretation exemplified in the above mentioned clinical cases. MATERIALS AND METHODS: Two male patients with clinical histories of CAH are presented. The 17OHP dosage on dried blood spots (DBS) was made through a competitive enzyme-immunoassay. The serum dosage was made by radioimmunoassay, using a direct measurement protocol (NE-17OHP) and a post-organic phase extraction protocol (E-17OHP). The molecular study of the 21-hydroxylase (CYP21A2) gene was made at Garrahan Hospital. RESULTS: Case 1: Female patient´s brother with CAH salt-wasting (SW). His mother was administrated with Dexamethasone during the first months of pregnancy. Full-term newborn, with normal birth weight, normal ionogram and E-17OHP 4.5ng/ml (cut-off: 5.3ng/ml). Clinical control and follow up allow detecting a weight loss. Blood analysis were repeated after 20 days of life: Na+ 134 mEq/l, K+ 7 mEq/l, NE-17OHP >25 ng/ml (cut-off: 21.3 ng/ml), Testosterone 0.9 ng/ml, Δ4-Androstenedione 7.0 ng/ml, and Cortisol 5.9 ug/dl. Molecular studies were not authorized by the family. Case 2: Brother of two ill sisters, one with CAH simple virilizing and the other with a SW type. His mother was administrated with Dexamethasone until the fifth month of pregnancy. Full-term newborn, normal birth weight, normal ionogram and 17OHP on DBS, NE-17OHP: 12.2 ng/ml, E-17OHP 0.3 ng/ml. At age of 7 months, NE-17OHP > 25ng/ml, E-17OHP 13.9ng/ml. I172N and R356P mutations were found (same mutation in his sisters). CONCLUSIONS: In spite of corticoid therapy has been described that can only affect neonatal screening in the last month of pregnancy, we found two patients with familiar history of CAH whose mothers were prescribed with high doses of Dexamethasone in their first months of pregnancy. Initially the 17OHP value was normal but lately it increased being difficult to know if it was due to the corticoid therapy or to the own CAH evolution. The utility of early molecular studies would reinforce the diagnosis.