

0131 - GROWTH PATTERNS AND TREATMENT-RELATED FACTORS IN CHILDREN WITH CONGENITAL ADRENAL HYPERPLASIA IDENTIFIED BY NEWBORN SCREENING IN SOUTHERN BRASIL

Mode: Research paper

Thematic Unit: Adrenals

PEÑA LITUMA, Gabriela Michelle¹ | SARTORETTO, Renata¹ | METZDORF HESSEL, Laura² | OSÓRIO, Georgia³ | KOPACEK, Cristiane⁴

UNIVERSIDADE FEDERAL DE CIÊNCIAS DA SAÚDE DE PORTO ALEGRE¹; PONTIFÍCIA UNIVERSIDADE CATÓLICA DO RIO GRANDE DO SUL²; UNIVERSIDADE DO VALE DO RIO DOS SINOS³; UNIVERSIDADE FEDERAL DO RIO GRANDE DO SUL E SERVIÇO DE REFERENCIA EM TRIAGEM NEONATAL DO RIO GRANDE⁴

Background/Objectives: Congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency is the most common cause of congenital hyperandrogenism. Although early diagnosis through newborn screening reduces morbidity and mortality, long-term growth may be affected by both disease severity and chronic glucocorticoid therapy. This study evaluated growth patterns, skeletal maturation, hormonal control, and treatment-related factors in patients with CAH identified through the public newborn screening program in Southern Brazil.

Methods: A cross-sectional retrospective study was conducted including patients diagnosed with classic CAH through newborn screening between May 2014 and April 2025. Diagnosis was confirmed by biochemical and molecular analyses. Auxological parameters, bone age, glucocorticoid and mineralocorticoid doses, and 17-hydroxyprogesterone (17-OHP) levels were evaluated. Statistical analyses were performed with a significance level of 5%.

Results: Fifty-four patients were included, comprising 47 (87%) with salt-wasting CAH and 7 (13%) with simple virilizing CAH. Growth outcomes during the first 48 months of life were generally favorable, supporting the benefits of early diagnosis and prompt treatment. No significant differences in growth parameters were observed between clinical subtypes. Higher glucocorticoid doses were associated with lower growth velocity ($p=0.011$), particularly among females ($p=0.013$). In girls, higher mineralocorticoid doses were also associated with lower height ($p=0.034$) and lower weight ($p=0.048$). Hormonal control remained challenging, with only 46% of patients presenting 17-OHP levels within the target range, while 33% had markedly elevated levels. No association was observed between glucocorticoid type and hormonal control.

Conclusion: Early diagnosis through newborn screening was associated with favorable early growth outcomes in children with CAH. Growth impairment was more strongly influenced by treatment-related factors than by clinical subtype, with females appearing particularly vulnerable to glucocorticoid excess. These findings highlight the importance of individualized therapeutic adjustments and careful longitudinal monitoring to optimize growth and hormonal control.

