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Association Between Serum Steroid Profile and Metabolic Risks in Adults with Classic Congenital Adrenal Hyperplasia Due to 21-Hydroxylase Deficiency

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Objective: In adult patients with congenital adrenal hyperplasia (CAH), long-term health outcomes, including metabolic and cardiovascular health, are a priority for the management of CAH. The enzyme 21-hydroxylase deficiency, which accounts for a majority of CAH, leads to a large perturbation in serum steroid profile under glucocorticoid replacement therapy. Serum levels of several adrenal steroids, including 17-hydroxyprogesterone and androstenedione, are associated with disease severity and sexual dysfunction of CAH patients. However, the association between the serum steroid profiles with metabolic risk in CAH patients is not elucidated yet. We aimed to investigate the serum steroid profile of adult CAH patients using liquid chromatography-mass spectrometry (LC-MS) and their association with metabolic risk. **Methods:** Adult patients with classic CAH due to 21-hydroxylase deficiency were included in this study (36 females and 27 males). A multi-steroid panel composed of 25 steroids and their metabolites was applied to morning serum samples of study subjects using LC-MS. An unsupervised clustering algorithm was applied to the serum steroid panel to discover any unique pattern which divides CAH subjects into clusters. The association between serum steroid profiles and clinical characteristics, including age, sex, body mass index (BMI), subtypes (simple virilizing and salt wasting), glucocorticoid dose, and the presence of hypertension, diabetes mellitus (DM), and metabolic syndrome (MetS) was analyzed. The discriminatory power of each steroid or a combination of steroids was estimated using the area under the curve of receiver operating characteristics (AUROC) for metabolic diseases. **Results:** Fifteen steroids including pregnenolone sulfate, 17α-hydroxyprogesterone, cortisol, cortisone, 21-deoxycortisol, 20α-dihydrocortisol, 20α-dihydrocortisone, α-cortolone, β-cortolone, tetrahydrocortisone, dehydroepiandrosterone sulfate, testosterone, androstenedione, 11β-hydroxytestosterone, and 11β-hydroxyandrostenedione were successfully quantified in all subjects. The median age of subjects was 28 years (interquartile range: 25-35). The prevalence of DM, hypertension, and MetS was 3.2% (2/63), 15.8% (10/63), and 28.6% (18/63), respectively. The subjects were divided into two clusters based on the pattern of serum steroid profile using unsupervised hierarchical clustering. The prevalence of MetS was significantly different between the two clusters (cluster 1, 37.8% [17/45] vs. cluster 2, 5.6% [1/18], P = 0. 011). The prevalence of HTN was numerically higher in cluster 1 than cluster 2 (20. 0% [9/45] vs. 5.6% [1/18], P = 0.257). Other clinical characteristics, including age, sex, BMI, subtypes, and glucocorticoid dose, were not different between the two clusters. Among fifteen steroids, the level of tetrahydrocortisone showed the highest discriminatory power for MetS (AUROC 0.795, 95% confidence interval: 0.675-0.914). The multivariate logistic regression model of all 15 steroids showed AUROC of 0.832 (95% CI: 0.732-0.933) for MetS. **Conclusion:** The serum steroid profiles of CAH patients were significantly associated with the presence of MetS. This suggests that serum steroid signatures can guide the optimal management of adult CAH patients to minimize the risk of MetS.

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