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Impact of glucocorticoid receptor gene polymorphisms on the metabolic profile of adult patients with congenital adrenal hyperplasia under glucocorticoid replacement

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The introduction of glucocorticoid (GC) replacement therapy in the 1950s has allowed for a normal life span in CAH patients. However, it has been described that adult CAH patients presented increased prevalence of obesity, insulin resistance and hypertension, as well as adverse lipid profiles. These findings suggest that CAH patients are prone to develop an unfavorable cardiovascular risk profile and previous reports have not associated these findings with GC doses and/or duration of therapy; suggesting that other factors may be involved, such as genetic predisposition. In the general population, glucocorticoid receptor (NR3C1) gene polymorphisms are linked with increased cardiovascular risk: the BclI allele has been associated with increased GC sensitivity and consequently to higher BMI, waist circumference and lipid levels, compared to wild-type carriers. Taking into account these data, we recently tested the association analysis between NR3C1 polymorphisms and these traditional cardiovascular risk factors in adult CAH patients and observed that the BclI carriers presented with increased BMI and waist circumference, independent of sex and age. Additionally, the frequency of the BclI allele was higher in the obese CAH compared to the non-obese CAH patients and their carriers also presented with higher systolic blood pressure levels. Our data suggest that NR3C1 polymorphisms could play an important role in the susceptibility for adverse metabolic profile in CAH patients. Hence, screening of NR3C1 alleles during the treatment of CAH patients could help to improve the quality of GC replacement, by identifying subgroup patients at-risk who would benefit the most from personalized treatment.

Biography

Tania Sanchez Bachega has completed her PhD in Endocrinology, School of Medicine- Sao Paulo University. She is working as an Associate Professor of Endocrinology, School of Medicine-Sao Paulo University. She is Consultant of Brazilian Ministry of Health in Neonatal Screening of Congenital Adrenal Hyperplasia. She is also a Medical Coordinator of Congenital Adrenal Hyperplasia Ambulatory, Hospital das Clinicas, School of Medicine, Sao Paulo University, Brazil.

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