Central adrenal insufficiency in young adults with Prader-Willi syndrome.

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Objective: A high prevalence (60%) of central adrenal insufficiency (CAI) has been reported in Prader-Willi syndrome (PWS) using the metyrapone test. We have assessed CAI in adults with PWS using the low-dose short synacthen test (LDSST).

Design: Basal cortisol and ACTH, and 30-min cortisol after the administration of 1 μg synacthen, were determined in 53 PWS adults (33 females). A peak cortisol value of ≥ 500 nmol/l was taken as normal. Hormonal profiles were analysed in relation to gender, genotype and phenotype. Deficient patients were retested by high-dose short synachten test (HDSST) or a repeat LDSST.

Results: Mean ± SD basal cortisol and ACTH were 336.6 ± 140.7 nmol/l and 4.4 ± 3.7 pmol/l respectively. Cortisol rose to 615.4 ± 135.0 nmol/l after LDSST. Eight (15.1%) patients had a peak cortisol response <500 nmol/l, with a lower mean ± SD (range) basal cortisol of 184.9 ± 32.0 (138.0–231.7) compared with 364.1 ± 136.6 (149.0–744.5) in normal responders ($P < 0.001$). Seven of the eight patients underwent retesting, with 4 (7.5%) showing persistent suboptimal responses. Basal and peak cortisol correlated in females ($r = 0.781$, $P < 0.001$). Logistic regression revealed that only female gender and baseline cortisol were predictors of cortisol peaks (adjusted R square 0.505).

Conclusions: Although CAI can be part of the adult PWS phenotype, it has a lower prevalence (7.5%) than previously reported. Clinicians are advised to test PWS patient for CAI. Our study also shows that basal cortisol is closely correlated with adrenal response to stimulation, indicating that its measurement may be helpful in selecting patients for LDSST.