ADRENAL FUNCTION

Adult CAH—does genotype correlate with phenotype?

In adults with congenital adrenal hyperplasia (CAH), health outcomes are not related to the severity of *CYP21A2* mutations, report Krone *et al.* This finding suggests that an acquired factor, such as inadequate medical care, leads to poor health outcomes.

“No consensus on therapy exists for adult patients with CAH and we previously found that this patient group have poor health outcomes with a reduced quality of life and an increase in cardiovascular disease risk,” says researcher Richard Ross of the University of Sheffield. “A key question coming out of our work was whether the poor health outcomes relate to the particular genetic defect.”

In children with CAH owing to *CYP21A2* deficiency (which accounts for ~95% of cases of CAH), genotype strongly correlates with phenotype, but the correlation between genotype and phenotype in adults with CAH was not known. The researchers studied treatment and health outcomes in 153 adults with CAH with varying degrees

of enzyme deficiency owing to different mutations in *CYP21A2*.

The investigators found a weak correlation between genotype and treatment provision, as higher doses of glucocorticoids were prescribed to patients with more severe *CYP21A2* mutations. However, health status was similarly impaired across groups with different severities of *CYP21A2* mutations, indicating no correlation between health outcomes and genotype.

The team are now looking at whether health outcomes relate to specific treatment regimens, including glucocorticoid therapy.

Carol Wilson

Original article Krone, N. *et al.* Genotype–phenotype correlation in 153 adult patients with congenital adrenal hyperplasia due to 21-hydroxylase deficiency: analysis of the United Kingdom Congenital Adrenal Hyperplasia Adult Study Executive (CaHASE) Cohort. *J. Clin. Endocrinol. Metab.* doi:10.1210/jc.2012-3343