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Update on Congenital Adrenal Hyperplasia

Actualización sobre hiperplasia suprarrenal congénita

Nils P. Krone

University of Sheffield. Sheffield (United Kingdom)

Congenital adrenal hyperplasia (CAH) represents a group of autosomal recessive disorders in steroid hormone biosynthesis causing deficient cortisol biosynthesis. The incidence of congenital adrenal hyperplasia in the general population of Western countries is approximately 1 in 10,000 to 1 in 15,000 life births with about 95% of cases caused by 21-hydroxylase deficiency. After the introduction of replacement therapy with glucocorticoids and mineralocorticoids in the 1950s, CAH has become a chronic life-long condition with associated comorbidities and long-term health implications. Increasing evidence suggests that unfavourable outcomes can involve all organ systems. Thus, an important emerging task for paediatricians providing care for children and young people with CAH is the prevention of such long-term health problems. The caring physician should be aware of co-morbidities involving cardiovascular disease, metabolic complications, fertility problems, psychosexual and psychologic health. Patients are ideally looked after by a multi-disciplinary team. A key challenge remains the pharmacotherapy with current approaches that struggle to keep the balance between glucocorticoid overexposure and androgen excess. Over the life-span type and relative dose of glucocorticoid can change. The relative mineralocorticoid dose per body surface area declines with increasing age. Recent evidence suggests that significant differences in treatment regimens persist that are not necessarily explained by interindividual differences. Replacement doses are monitored by clinical and biochemical parameters, with suppression of steroid hormones commonly indicating overtreatment. Several novel experimental pharmacotherapies with the aim to reduce glucocorticoid exposure and to mimic physiological glucocorticoid secretion are under development. The approach to treatment and monitoring remains highly variable and the effects on long-term outcomes remain elusive. This presentation will provide an update on current developments covering care provision, novel adrenal-specific biomarkers with the potential to improve monitoring and experimental approaches. All these ongoing efforts have in theory the potential to improve CAH management; however, the evidence for improved long-term outcomes and large cohorts is still suboptimal.