

# Society for Endocrinology – Media Release



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## Patients missing out on specialist care

A new study suggests that less than 10% of the estimated 3,591 adults in the UK with the genetic condition congenital adrenal hyperplasia (CAH) receive specialist endocrine care, and that there is no standard treatment for these patients. The results are published in the *Journal of Clinical Endocrinology and Metabolism*. The study, which represents a new paradigm for co-ordination of research by a specialist society, was funded exclusively by the Clinical Endocrinology Trust and led by Professor Richard Ross from the Society for Endocrinology and University of Sheffield.

CAH is a genetic disorder where the body cannot produce the stress hormone cortisol and as a result produces excess male hormone (testosterone), which causes virilisation of girls and precocious puberty in both boys and girls. Children with CAH receive standardised treatment from a multi-disciplinary team of specialists, which includes a paediatric endocrinologist. However, for adult patients no standard therapy has been established.

The Society for Endocrinology formed the Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) to address an apparent lack of literature on the clinical management of adults with CAH. 199 adults with CAH (who were under specialist endocrine care) were given a physical examination, had a blood sample taken and completed a quality of life questionnaire. From these data, the researchers analysed the physical, metabolic and subjective health status of this group.

Data on the health status of these patients showed improvements to clinical management are needed. Compared to population-based controls, both men and women with classic CAH were statistically significantly shorter (on average 14 cm in men and 8 cm in women;  $p < 0.001$  for both men and women). Women with classic CAH were significantly more obese (average BMI 32.9 vs 26.7;  $p < 0.0001$ ) and had a greater waist circumference (99.5cm vs 85.9cm;  $p < 0.0001$ ) than age- and sex-matched healthy individuals. Levels of testosterone and other hormones linked to CAH and its medication were outside therapeutic ranges in a high number of cases in all groups. The study showed there is much variation in the day-to-day management of patients with CAH and further research is needed to determine the optimum treatment regimen for these patients. Self-assessed quality of life was poor in all groups.

Patient enrolment data suggested that less than 10% of UK adult CAH patients receive specialist endocrine care. Based on the poor health status and lack of consensus on management, CaHASE recommends that all CAH patients be referred to a specialist clinic for treatment by a multi-disciplinary team to ensure the proper provision of care to these patients.

Further research is urgently needed to understand the effects of long-term glucocorticoid usage on health and to allow clinicians to reach an evidence-based consensus on how the medication should be prescribed. It is hoped that this first study will lead to further investigations into the management of adults with CAH to improve treatment.

**Professor Richard Ross of the Society for Endocrinology and Chairman of the Congenital adrenal Hyperplasia Adult Study Executive (CaHASE) said:**

*“The fact that people with congenital adrenal hyperplasia are now surviving through adulthood is wonderful, but we are now faced with a new problem; how are we best able to support patients with this life-long condition which requires long-term steroid therapy?”*

*“We initially set out to write guidelines to answer this very question, but found that the existing research base was inadequate. The Congenital adrenal Hyperplasia Adult Study Executive was set up by the Society for Endocrinology to examine the current health status of adults with congenital adrenal hyperplasia. The project’s success should awaken other specialist societies to the potential for co-ordination of large cohort studies.”*

*“Our study has discovered that under 10% of adults with congenital adrenal hyperplasia receive treatment from a specialist endocrine clinic. We must remedy this if we are to treat the condition in adulthood as well as we do in childhood, where specialist care has long been the norm. In particular, we have shown that the current use of synthetic glucocorticoids to treat adults with CAH is inconsistent across centres and not supported by sufficient evidence, making this an important item on the research agenda.”*

### **Information on congenital adrenal hyperplasia**

CAH is a congenital disorder resulting from a defect in the adrenal enzyme responsible for synthesis of the stress steroid cortisol. As a result of cortisol deficiency the body stimulates the adrenal glands, trying to drive them to produce cortisol. This excessive drive results in excess of precursor steroids that are predominantly androgens. The over-production of androgens (male hormones; predominantly testosterone) in CAH means that girls with CAH develop partial male genitalia during foetal development, and unless treated from birth they become virilised, developing a male pattern of body and facial hair (hirsutism). Boys and girls can enter a very early precocious puberty. In both sexes growth and fertility problems are common. Much less is known about the condition in adulthood, but inadequate treatment results in infertility in both sexes, and as the natural daily pattern of cortisol levels is hard to mimic with current medication long-term excess treatment can result in adverse effects such as high blood pressure and obesity

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Notes for editors:

This paper appears in the *Journal of Clinical Endocrinology & Metabolism*. A full copy of this paper is available to download [here](#).

Arlt et al. 2010. Health Status of Adults with Congenital Adrenal Hyperplasia: A Cohort Study of 203 Patients. *Journal of Clinical Endocrinology & Metabolism*, **95**, 5110-5121. doi: 10.1210/jc.2010-0917

The Society for Endocrinology is UK's largest national organisation promoting endocrinology and hormone awareness. For general information, please visit our website: [www.endocrinology.org](http://www.endocrinology.org)

'Living with CAH' is a patient group that works to improve the medical management of CAH and offers information and support on CAH to patients and their friends and family: <http://www.livingwithcah.com/>.

## **Please mention the Society for Endocrinology in any story**

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### **ABSTRACT**

**Context:** No consensus exists for management of adults with CAH due to a paucity of data from cohorts of meaningful size.

**Objective:** To establish the health status of adults with CAH.

**Design & Setting:** A prospective cross-sectional study of adults with CAH attending specialized endocrine centers across the UK.

**Patients:** 203 CAH patients (199 with 21-hydroxylase deficiency): 138 women, 65 men, median age 34 (range 18-69) years.

**Main Outcome Measures:** Anthropometric, metabolic, and subjective health status. Anthropometric measurements were compared to Health Survey for England (HSE) data, and psychometric data to appropriate reference cohorts.

**Results:** Glucocorticoid treatment consisted of hydrocortisone (26%), prednisolone (43%), dexamethasone (19%), or a combination (10%), with reverse circadian administration in 41% of patients. Control of androgens was highly variable with a normal serum androstenedione found in only 36% of patients whereas 38% had suppressed levels suggesting glucocorticoid over-treatment. In comparison to HSE participants, CAH patients were significantly shorter and women with classic CAH had a higher body mass index and increased diastolic blood pressure. Metabolic abnormalities were common, including obesity (41%), hypercholesterolemia (46%), insulin resistance (29%), osteopenia (40%) and osteoporosis (7%). Subjective health status was significantly impaired and fertility compromised.

**Conclusions:** Currently a minority of adult UK CAH patients appear to be under endocrine specialist care. In the patients studied, glucocorticoid replacement was generally non-physiological and androgen levels were poorly controlled. This was associated with an adverse metabolic profile, and impaired fertility and quality of life. Improvements in the clinical management of adults with CAH are required.