

CONGENITAL ADRENAL HYPERPLASIA: PARENTS' EXPERIENCES OF TREATING THEIR CHILD'S CONDITION

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Background

Patients with Congenital Adrenal Hyperplasia (CAH) require life-long, daily treatment to avert the risk of growth problems and serious illness. Currently, there is no licensed hydrocortisone hormone replacement therapy for children under 6 in Europe and existing treatment often requires adapting adult doses by crushing tablets. As primary caregivers, parents take on responsibility for their child's routine medication and adapting doses in times of stress or illness. There has been little research to date exploring their experiences, and the challenges associated with treating this rare condition.

Aim

The study aimed to capture parents' experiences of treating their child's adrenal insufficiency, including CAH. It was conducted by Genetic Alliance UK as part of the European Commission funded TAIN (Treatment of Adrenal Insufficiency in Neonates) Project - a clinical trial developing a new formulation of hydrocortisone for neonates and infants².

Method

Taking a mixed methods approach, Genetic Alliance UK captured the views of parents across three European countries. In 2014, 17 semi-structured interviews were conducted in the UK and analysed thematically with the support of Assisted Qualitative Data Analysis Software, NVivo 8. In 2015, an online survey was developed, piloted and disseminated widely to parents of children under the age of 6 in the UK, the Netherlands and Germany. Fifty-four survey responses were received and the data analysis is ongoing. Genetic Alliance UK's study was approved by the University of Sheffield's research ethics committee.

In **conclusion**, the study has provided a unique insight into the wider impact of managing CAH. It has increased understanding of the reality of living with and the challenges of treating adrenal insufficiency from the family perspective. This context is critical to the development of successful interventions, which are lacking for the majority of rare diseases.

Findings

The findings highlighted a number of challenges associated with the treatment regime, and the burden experienced by parents as the primary carers:

- Interviewees reported a '**latent anxiety**' as well as **disruption** to their daily routines and their work life.

It's just the hassle of having to get his medication ready and you can never forget it... So I think that adds stress...you're always thinking right when's the next time, when's the next time. And so there's a, kind of, latent anxiety. [Interview Transcript]

- Interviewees argued that much of the burden was associated with having to get **the right dose of medication to their child, at the right time**. A quarter of survey respondents felt that the medication had to be given at inconvenient times, and 1 in 5 respondents reported that their sleeping pattern was affected.
- Interviewees, and 1 in 5 survey respondents, reported concerns about **delegating medication responsibility** to others.

I think when you're already a bit traumatised and looking after a new baby, having to chop tablets up into little bits... all that sort of faffing around, you could really, really do without. And especially when you're worrying whether you've got the quantities right... [Interview Transcript]

Overall, the survey findings did present a more positive parental experience. Findings suggest that families successfully adjusted to the chronic condition and treatment over time:

- Almost 60% of survey respondents (33 parents) reported that their child's condition and symptoms were well controlled by their medication regime (scoring 8 or above on a scale of 1 to 10).
- Two thirds of survey respondents agreed that they had been equipped with the appropriate skills and knowledge to manage their child's condition.

...he was so young, it probably helped us and probably helped him, I mean, he's grown up with the medicine and it's one of those things where we just sort of got used to it as part of a new routine. [Interview Transcript]



¹Genetic Alliance UK is an alliance of over 180 patient organisations and the national charity working to improve the lives of patients and families affected by genetic conditions www.geneticalliance.org.uk

²This work was funded by the European Commission under a Framework 7 Grant (No: 281654 – TAIN) www.tain-project.org

