Exploring the psychosocial impact of Congenital Adrenal Hyperplasia on children and their parents

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Background

Congenital Adrenal Hyperplasia (CAH) is a genetic condition associated with deficiencies in cortisol and aldosterone and an overproduction of androgens. CAH is rare, with an estimated prevalence of 1:10,000[2]. The condition requires life-long, daily medication, although currently there is no licensed therapy for children. Little is known about the psychosocial impact of living with and treating CAH, particularly for the parents of young patients. The study was conducted by Genetic Alliance UK as part of the European Commission funded TAIN (Treatment of Adrenal Insufficiency in Neonates) Project[3], which aims to develop a new formulation of hydrocortisone for neonates and infants.

Method

Taking a qualitative approach, 17 semi-structured interviews were conducted in the UK during 2014, with parents of children affected by CAH. Parents were recruited via the 'Living with CAH' Support Group. Interviews were conducted face-to-face or via the telephone, and each lasted approximately one hour. Interviews focused on parents' experiences during the diagnosis period, the psychosocial impact of the condition on parents and children, and the families' experiences of treating the condition. They were audio-recorded, transcribed verbatim and analysed thematically with the support of Computer Assisted Qualitative Data Analysis Software, NVivo8.

Findings

Parents reported a number of ways in which CAH impacted on them and their children. Many of the impacts can be attributed to two factors associated with the condition including the treatment regime and the rarity of the condition. These are described below in greater detail...

Interviewees described the impact of the current **treatment regime** for children with CAH:

- Disruption to daily routines and sleep patterns (particularly for parents) as a result of the frequency of medication required.
- A latent anxiety that occurred as a result of having to get the right dose of hydrocortisone to their child at the right time. This was exacerbated by a number of things including: the importance of the medication regime for their child's immediate and future health and development; concerns about whether cutting and/or dissolving tablets provided an accurate dose; concerns about whether their child was monitored closely enough by health professionals; and concerns about whether they were altering doses appropriately during times of stress and illness.
- Difficulties delegating responsibility for their child's care and treatment regime (due to some of the reasons outlined above) whether it be to childminders, playgroups, school or the patients themselves. For some parents, this had a direct impact on their employment decisions.

> It's exhausting, absolutely exhausting… 6 o-clock every single morning…

> They were telling me that the accuracy of the dose was the most important thing but then they were giving me tablets which didn't match up to it; there was a huge incongruence between the two

> I've got alarms going off all over the place and I'm very aware of the time all the time …

> I do have a lot of concern when he's out of our immediate control when he grows older....going out, getting drunk [and] forgetting all about their medication...

Parents reported a number of challenges associated with the **rarity** of CAH:

- A lack of awareness about CAH amongst general health professionals and the public. As a result, parents particularly valued the opportunity to connect and share experiences with other families via support groups.
- Delays in getting an accurate diagnosis, with some families having to wait until their child fell seriously unwell (an adrenal crisis) before receiving a diagnosis or treatment.
- Disparities in care and support across the UK according to region or specialist.

> I think we've been very lucky....I know it's incredibly rare...if she was born in a remote hospital they won't see any cases probably and they won't know what on earth's going on

> I'm literally educating the doctors... [they] can't be an expert on every little condition...it means we're continually having to....keep on top of it

In **conclusion**, the study offers a rare insight in to the daily experiences of families affected by CAH and has important implications for the ongoing TAIN project and the care and treatment of the condition in the future. The findings have informed the development of an online survey which will be disseminated to European parents in English, Dutch and German in spring/summer 2015.

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[1]Genetic 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

[2]Orphanet website www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=GB&Expert=418.0 [accessed on 7.4.15]

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