**Parental concerns about genital differences in children with congenital adrenal hyperplasia persist regardless of the selected intervention**

Abstract

Background

Congenital Adrenal Hyperplasia due to 21-hydroxylase deficiency (21HD) can affect the in-utero development of the genital anatomy of people with 46XX karyotype, and health professionals engage parents in decision making regarding managing genital this difference, including genital surgery options and patient communication.

Aim

To investigate parental communication relating to the daughter’s clitoral size-variation.

Methods

Semi-structured in-person interviews of 24 parents of XX children with a neonatal CAH diagnosis comprised three management categories,  (i) clitoral reduction surgery (RS) parents N=7; children N=9; (ii) clitoral concealment surgery (CS); parents N=8; children N= 8 and (iii) children with the same diagnosis who have not undergone surgery on or around the clitoris (NS), parents N=9; children N=7.

Outcomes

Four representative themes, Obvious Choice, Still Different, Parental Burden and Ignorance is Bliss were common across all three treatment groups.

Results

For most parents, genital appearance alteration via clitoral reduction or clitoral concealment surgery, nor avoidance of clitoral surgery, did not ameliorate concerns, with most expressing an aversion to educate their child on the topic of genital difference, past treatment, or future function.

Clinical Implications

The reliance on surgical treatment pathway to manage this psychosocial concern is ineffective and requires psychosocial interventions.

Strengths and Limitations

This qualitative study but is limited to parents of children with a specific genital difference, without directly exploring parental value of the clitoris or the application of adequate psychosocial care.

Conclusion

Services must impact parental ability in essential communication allowing them to more genuine decision-making engagement, adapt to enduring genital reality including possible future sexual challenges for their adult child, without resorting to burdensome strategies focused on attempts to perpetuate a benevolent ignorance.

Introduction

Congenital adrenal hyperplasia (CAH) due to 21 hydroxylase deficiency (21HD) is a family of autosomal recessive conditions that requires life-long medical management of adrenal insufficiency. CAH can be associated with both 46,XX and 46,XY karyotypes. For 46,XX children, the genital morphology is altered by elevated foetal androgens in-utero. Amongst other features, the clitoris is typically enlarged. As such, CAH in 46,XX children is recognised by most medical experts as a difference in/ disorder of sex development (DSD), an intersex condition, or a Variation of Sex Characteristics or Development . These terms are considered pejorative by many of the people they describe. Furthermore, some parties suggest that CAH is removed from the DSD nomenclature 11. This does not negate the bioethical debate on surgical alteration of the clitoris in childhood.

Childhood clitoral surgery has been questioned following concern about negative long-term effects on clitoral sensitivity and the attendant impact on sexual function 2. Furthermore, the expected psychological benefits, such as easing of parental anxiety or impacting the child’s gender identity are not evidenced. The controversy of clitoral surgery was formally acknowledged in the international consensus of intersex management in 2006 3, but translation of a more cautious approach to surgery is inconsistent4. A lack of transparency to date, makes it impossible to ascertain surgery prevalence.

The way in which DSD/intersex is talked about by health professionals can influence the uptake of interventions5. Terming genital difference a disorder, swiftly implies a need for amelioration. Without a de-pathologised perspective and a non-surgical care programme 6, the surgical path becomes a non-dilemmatic choice 7. Thus far, psychosocial research consistently shows that parents have a central role as decision makers, but also that this significant decision is undermined by a strong lead from health professionals 8,9. One rationale for a multi-professional paediatric healthcare team is to offer parents facilitated thinking about treatment options for their child and future adult. However, many DSD services primarily function as biomedical services, with many gaps in psychosocial care both in terms of quantity and quality8,10. Psychosocial professionals can be absent from or quiet within multidisciplinary teams, promoted only when a medical process falters, rather than being strategically positioned and systematically empowered to lead interventions to address psychosocial concerns 11. Unfortunately, parents struggle to communicate with their daughters about CAH, bar aspects relating to medication 12,13. This phenomenon is highlighted in detail by Roen et al 2023, providing detail of historical concealment and communication barriers and challenges for health care providers. This study aims to provide insight into parents’ motivations and concerning about the knowledge shared with their daughters about the impact of CAH on genital anatomy and surgical decision making.

## Methods

In this qualitative study, two UK regional paediatric centres recruited (via clinic) parents of contemporary patients aged 18 months to 16 years with CAH who had been identified with atypical genitalia including clitoromegaly in the neonatal period. Eligible patients were recruited by treating teams as they attended the participating centres, with recruiting ending when the representative sampling was achieved. There was no sampling by perceived or measured clitoral size.

We used purposive sampling 14 to include parent participants of children who fell within one of three clitoral management categories: clitoral reduction surgery (RS) parents N=7; children N=9, clitoral concealment surgery (CS); parents N=8; children N= 8 and no surgery on or around the clitoris (NS), parents N=9; children N=7. Informed consent was obtained from all participants. Of those approached two parents (CS) declined to take part due to privacy concerns, Two further parents agreed (CS and NS) and were recruited but not interviewed due to interview scheduling problems.

Two of the NS children had undergone surgery that did not involve the clitoris or the skin surrounding the clitoris. All the children of participants in the RS and CS groups also underwent vagino-labial surgery. Clitoral reduction removes tissue from the shaft of the clitoris, while concealment involves clitoral shaft skin dissection, leaving a skin collar to cover the glans 15. These surgeries usually occur as part of feminising genitoplasty involving separating the urethra and vagina openings in the perineum 15–18. The study was specifically aimed at parents due to ethical constraints and was approved by Southwest NHS ethics process (Rec Ref 15/SW/0253).

Procedure

Audio recorded single 1-1 semi-structured interviews using a schedule of questions and prompts, with individual parents of 35-55 minutes duration took place in the parent’s home or in a counselling room and were professionally transcribed. The female clinical psychologist interviewer and first author was known to one third of the participants. All participants knew that she is part of one of the two recruiting clinical teams, with an interest in improving psychosocial family-based care. Using Reflexive Thematic Analysis approach19,20 we identified, analysed, and reported patterns across the data set. This provides representative themes and looks beyond the surface-level meanings of the participants’ account and is useful in a critical psychology research approach. Interpretive analysis is then scrutinised by a second researcher 21. For quality assurance22, the coding of nine systematically selected transcripts (33%) were checked, choosing a participant from categories including the recruitment site, the treatment pathway, ethnic background of the family and involvement of both mothers and fathers. Any codes that were not agreed upon were discussed and recoded. Two participants read the draft paper including data quotes, representative themes and conclusions and feedback on the findings was presented at a patient group meeting.

## Results

Six fathers and 18 mothers (including 3 couples) of children aged 18 months to 15 years old (including five siblings) formed the sample of the current study. Four were of South Asian heritage and others were White British (using standard UK health service categories). The results provide four representative themes to which we gave descriptive labels in Table 1 then detail.

Table 1 Summary of themes and sub-themes

Obvious Choice

Parents reported feeling involved in decision making, yet easily reaching the decision about genital surgery. They were explicit about the influence that health professionals had on their decision making. They felt that professionals often presented one treatment option as an obvious choice, leaving no need for real deliberation. The process was recalled as being straightforward and un-dilemmatic, with the only exception relating to one parent’s request for total clitoridectomy. Some parents recalled being superficially engaged in discussion of a non-surgical care path, *“I very clearly somewhere in my mind do remember that option being said to me that some parents choose not to do it, but certainly - my overwhelming feeling was, Oh, my god, no – we’re gonna do it and I felt that the medical profession as well were more or less saying – ‘well clearly - you’re gonna do it’.*

Table 2 Quotes from Obvious Choice theme

Codes to reference the participants comprise Mo or Fa indicating mother or father, the age of the child and the surgical path: RS (reduction surgery), CS (concealment surgery) and NS (no clitoral surgery).

Still Different

Primary aims of surgery are to normalise the child’s genitals and reduce parental distress. However, parents from all three treatment groups discussed their daughter’s enduring genital difference and parental distress. Some parents did describe their daughter’s genital difference as an unproblematic aspect of life. Those who said genital difference was acceptable and manageable within day-to-day life, still described remaining concerns. The ‘still different’ theme demonstrates multiple possibilities for parental acceptance or concern, with notably polarised positions regarding genital appearance in “It’s Who She Is” and “It’s Not Right Down There”. Whilst interviewees expressed both acceptance and concern, they did not mention having discussed this with health professionals. Acceptance expressed in, “It’s who she is” and fears expressed in, “It’s not right, down there”, the analysis reveals a complex duality of parental feeling. However, parental perception of the child being genitally distinct from others was present across all three surgical and non-surgical groups.

Table 3 Quotes from Still Different

Parental Burden

Participants across the three treatment groups had deliberately delayed talking to their daughter about how CAH had affected her genital development. The parents expressed a lack of confidence in talking to the child about this sensitive and personal subject and maintained a fear of the child finding out for themselves. Ther was an awareness that the distress caused by the child’s genital difference was primarily the parents’ own and that the child could potentially tolerate and accept their bodily difference. Participants of each of the treatment groups, talked about feeling anxious and distressed about feeling compelled to suppress awareness for their daughter of her genital difference. Parents of children who underwent or avoided early childhood clitoral surgery each experienced distress related to their child’s genital difference.

Table 4 Quotes from Parental Burden theme

Ignorance is Bliss

Across the three treatment groups, participants described trying to escape detection of their child’s genital difference to minimise potential harm. The research participants postulated that their child’s lack of self-awareness of genital difference would be of benefit. Even parents of adolescents reported not having told their daughter about their genital difference and early surgery. Participants believed that the child remained unaware of CAH having an impact on genital development. Parents of children aged 8-15 years who expressed concern about current genital appearance said that the child did not know about having any physical difference, despite attending a multidisciplinary DSD clinic. A minority of participants claimed an openness within the family and reported that their child was aware of her difference.

Table 5 Quotes from Ignorance is Bliss theme

Discussion

This interview study focuses on clitoral variation, as a means of exploring parents’ communication and barriers, regarding the management of children’s atypical genital appearance. Parents were led by professionals to a ‘Obvious Choice’ regarding accepting or avoiding early childhood clitoral surgery, after which their child’s genitals were perceived by them to be ‘Still Different’ by most. The impact of their child’s genital difference remained a psychological and practical ‘Parental Burden’ and most alluded to a belief that they should prevent their child from knowing about her genital difference because ‘Ignorance is Bliss’.

Parents from all three groups parents in this study worry about their child’s sexual future. However, in line with previous findings 7,9,12,23–25, having reached a management decision, they felt that their child was still genitally different, with some expressing aversion, others expressing a level of acceptance or both reactions. Although there were parents whose child had undergone clitoral reduction or concealment who expressed disappointment in the limited normalising effect, this study notes that overall, parents across all three treatment paths appear content with their treatment-mode decision, irrespective of the specific ‘Obvious Choice’ made. Our findings are that parents recognise that their child’s body is ‘Still Different’ and are burdened by concerns for their child and future adult child, balancing a perception of a need for knowledge being in the future, with their perception of an enduring need for privacy. They expressed considerable hesitancy about educating the child and facilitating a growing awareness of their development at birth and past treatments. This balancing act is felt by parents of children with a range of appearance-affecting conditions 26. Some parents assumed that the child had not noticed anything different about themselves, despite regular hospital appointments that may involve genital examinations. Participants said that they rarely mentioned these “Parent Burden’ with their medical providers, a phenomenon demonstrated by Sanders et al 10. This suggests that certain care contexts are not conducive to exploring the psychological aspects of CAH. Unfortunately, this communication gap could lead to professionals remaining ignorant of the extent of parental distress and the growing child or adolescent’s lack of knowledge. While psychologists are members of the clinical care team their under use 11 precludes helping parents share information with children to promote bodily autonomy.

“Ignorance is bliss” is a common saying in English language. Almost all participants referenced their belief in benevolent ignorance and the burden of responsibility caused by feeling able to maintain the child’s lack of awareness, poor knowledge, and pre sexual life temporary state of ignorance. Limited disclosure to the child is underpinned by a wish to protect, yet first-person accounts suggest that this intention usually backfires. Adults with lived experience have repeatedly interpreted parental inability to talk about their bodily difference as a sign of unspeakable shame 27. It follows that it is indefensible for DSD health services to abdicate their responsibility by delegating the task of educating the child to the parents without adequate training and support to help them manage. This is an area for co-working between DSD clinicians, families and peer-community resources, and should not be left to parents whose health-literacy may be limited (24) .

This a qualitative study, and while number of participants is large for qualitative research, the number is small compared to quantitative research. The study is limited to a group of parents of children with a specific genital difference, i.e., a larger clitoris. It focuses on decisions made concerning whether to surgically alter the appearance of the clitoris. The current analysis does not address the diverse parental views on the importance, role, or value of the clitoris. Nor does it explore the validity of apparent parental expectation of their child’s ignorance about their sexed body. These issues would be interesting areas for further study.

Conclusion

An important dimension of multi-professional care is facilitating patient self-knowledge. This study suggests that DSD services may not be fully aware of the lack of information available to children if the responsibility for patient-education falls to parents. This can result in young people reaching sexual awakening without understanding or even awareness of body difference in appearance and function. Health professionals share responsibility for child wellbeing via partnership and direct support of parents. This study suggests that childhood genital surgery nor its absence does not singularly eradicate parental perception of difference, nor remove parental concerns. This underlines the need for greater psychosocial evaluation and intervention within conventional practice.

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