

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

3 Abstract

4 Background

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

9 Aim

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

11 Methods

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

16 Outcomes

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

19 Results

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

23 Clinical Implications

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

26 Strengths and Limitations

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

29 Conclusion

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

34

35 Introduction

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

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

51 The way in which DSD/intersex is talked about by health professionals can influence the uptake of
52 interventions⁵. Terming genital difference a disorder, swiftly implies a need for amelioration.
53 Without a de-pathologised perspective and a non-surgical care programme ⁶, the surgical path
54 becomes a non-dilemmatic choice ⁷. Thus far, psychosocial research consistently shows that parents
55 have a central role as decision makers, but also that this significant decision is undermined by a
56 strong lead from health professionals ^{8,9}. One rationale for a multi-professional paediatric healthcare

57 team is to offer parents facilitated thinking about treatment options for their child and future adult.
58 However, many DSD services primarily function as biomedical services, with many gaps in
59 psychosocial care both in terms of quantity and quality^{8,10}. Psychosocial professionals can be absent
60 from or quiet within multidisciplinary teams, promoted only when a medical process falters, rather
61 than being strategically positioned and systematically empowered to lead interventions to address
62 psychosocial concerns¹¹. Unfortunately, parents struggle to communicate with their daughters
63 about CAH, bar aspects relating to medication^{12,13}. This phenomenon is highlighted in detail by Roen
64 et al 2023, providing detail of historical concealment and communication barriers and challenges for
65 health care providers. This study aims to provide insight into parents' motivations and concerning
66 about the knowledge shared with their daughters about the impact of CAH on genital anatomy and
67 surgical decision making.

68 Methods

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

74 We used purposive sampling¹⁴ to include parent participants of children who fell within one of three
75 clitoral management categories: clitoral reduction surgery (RS) parents N=7; children N=9, clitoral
76 concealment surgery (CS); parents N=8; children N= 8 and no surgery on or around the clitoris (NS),
77 parents N=9; children N=7. Informed consent was obtained from all participants. Of those
78 approached two parents (CS) declined to take part due to privacy concerns, Two further parents
79 agreed (CS and NS) and were recruited but not interviewed due to interview scheduling problems.

80 Two of the NS children had undergone surgery that did not involve the clitoris or the skin
81 surrounding the clitoris. All the children of participants in the RS and CS groups also underwent
82 vagino-labial surgery. Clitoral reduction removes tissue from the shaft of the clitoris, while
83 concealment involves clitoral shaft skin dissection, leaving a skin collar to cover the glans¹⁵. These
84 surgeries usually occur as part of feminising genitoplasty involving separating the urethra and vagina
85 openings in the perineum¹⁵⁻¹⁸. The study was specifically aimed at parents due to ethical constraints
86 and was approved by Southwest NHS ethics process (Rec Ref 15/SW/0253).

87 Procedure

88 Audio recorded single 1-1 semi-structured interviews using a schedule of questions and prompts,
89 with individual parents of 35-55 minutes duration took place in the parent's home or in a counselling
90 room and were professionally transcribed. The female clinical psychologist interviewer and first
91 author was known to one third of the participants. All participants knew that she is part of one of
92 the two recruiting clinical teams, with an interest in improving psychosocial family-based care. Using
93 Reflexive Thematic Analysis approach^{19,20} we identified, analysed, and reported patterns across the
94 data set. This provides representative themes and looks beyond the surface-level meanings of the
95 participants' account and is useful in a critical psychology research approach. Interpretive analysis is
96 then scrutinised by a second researcher²¹. For quality assurance²², the coding of nine systematically
97 selected transcripts (33%) were checked, choosing a participant from categories including the
98 recruitment site, the treatment pathway, ethnic background of the family and involvement of both
99 mothers and fathers. Any codes that were not agreed upon were discussed and recoded. Two
100 participants read the draft paper including data quotes, representative themes and conclusions and
101 feedback on the findings was presented at a patient group meeting.

102 Results

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

107 Table 1 Summary of themes and sub-themes

108 Obvious Choice

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

118 Table 2 Quotes from Obvious Choice theme

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

122 Still Different

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

134 Table 3 Quotes from Still Different

135 Parental Burden

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

145 Table 4 Quotes from Parental Burden theme

146 Ignorance is Bliss

147 Across the three treatment groups, participants described trying to escape detection of their child's
148 genital difference to minimise potential harm. The research participants postulated that their child's

149 lack of self-awareness of genital difference would be of benefit. Even parents of adolescents
150 reported not having told their daughter about their genital difference and early surgery. Participants
151 believed that the child remained unaware of CAH having an impact on genital development. Parents
152 of children aged 8-15 years who expressed concern about current genital appearance said that the
153 child did not know about having any physical difference, despite attending a multidisciplinary DSD
154 clinic. A minority of participants claimed an openness within the family and reported that their child
155 was aware of her difference.

156 Table 5 Quotes from Ignorance is Bliss theme

157

158 Discussion

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

166 Parents from all three groups parents in this study worry about their child's sexual future. However,
167 in line with previous findings^{7,9,12,23-25}, having reached a management decision, they felt that their
168 child was still genitally different, with some expressing aversion, others expressing a level of
169 acceptance or both reactions. Although there were parents whose child had undergone clitoral
170 reduction or concealment who expressed disappointment in the limited normalising effect, this
171 study notes that overall, parents across all three treatment paths appear content with their
172 treatment-mode decision, irrespective of the specific 'Obvious Choice' made. Our findings are that
173 parents recognise that their child's body is 'Still Different' and are burdened by concerns for their
174 child and future adult child, balancing a perception of a need for knowledge being in the future, with
175 their perception of an enduring need for privacy. They expressed considerable hesitancy about
176 educating the child and facilitating a growing awareness of their development at birth and past
177 treatments. This balancing act is felt by parents of children with a range of appearance-affecting
178 conditions²⁶. Some parents assumed that the child had not noticed anything different about
179 themselves, despite regular hospital appointments that may involve genital examinations.

180 Participants said that they rarely mentioned these "Parent Burden" with their medical providers, a
[Type text]

181 phenomenon demonstrated by Sanders et al¹⁰. This suggests that certain care contexts are not
182 conducive to exploring the psychological aspects of CAH. Unfortunately, this communication gap
183 could lead to professionals remaining ignorant of the extent of parental distress and the growing
184 child or adolescent's lack of knowledge. While psychologists are members of the clinical care team
185 their under use¹¹ precludes helping parents share information with children to promote bodily
186 autonomy.

187 "Ignorance is bliss" is a common saying in English language. Almost all participants referenced their
188 belief in benevolent ignorance and the burden of responsibility caused by feeling able to maintain
189 the child's lack of awareness, poor knowledge, and pre sexual life temporary state of ignorance.
190 Limited disclosure to the child is underpinned by a wish to protect, yet first-person accounts suggest
191 that this intention usually backfires. Adults with lived experience have repeatedly interpreted
192 parental inability to talk about their bodily difference as a sign of unspeakable shame²⁷. It follows
193 that it is indefensible for DSD health services to abdicate their responsibility by delegating the task of
194 educating the child to the parents without adequate training and support to help them manage. This
195 is an area for co-working between DSD clinicians, families and peer-community resources, and
196 should not be left to parents whose health-literacy may be limited⁽²⁴⁾.

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

204

205 Conclusion

206 An important dimension of multi-professional care is facilitating patient self-knowledge. This study
207 suggests that DSD services may not be fully aware of the lack of information available to children if
208 the responsibility for patient-education falls to parents. This can result in young people reaching
209 sexual awakening without understanding or even awareness of body difference in appearance and
210 function. Health professionals share responsibility for child wellbeing via partnership and direct
211 support of parents. This study suggests that childhood genital surgery nor its absence does not

212 singularly eradicate parental perception of difference, nor remove parental concerns. This
213 underlines the need for greater psychosocial evaluation and intervention within conventional
214 practice.

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