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Ethical and familial dilemmas of genitoplasty encountered in Congenital Adrenal Hyperplasia

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Introduction and Objectives

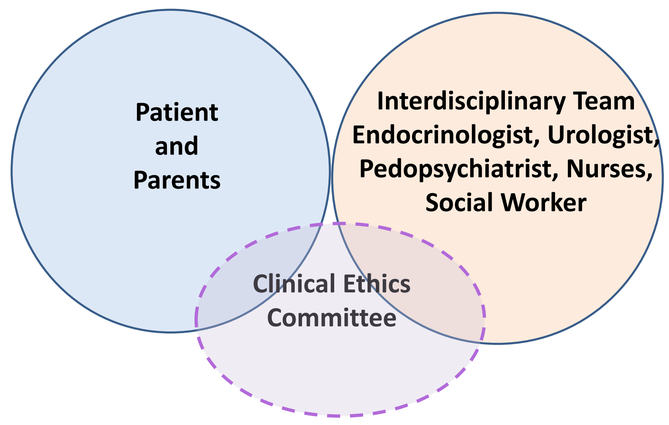
Differences of sex development (DSD) occur in about 1 in 3000 newborns in Switzerland(1). The role of genes and hormones in sex differentiation leading to the phenotypic sex is well known. On the other hand the knowledge of the factors that impact on our psychosexual outcomes (gender role, gender identity, and sexual orientation) remains poor. These outcomes can be inconstant, have endless variations and lead to gender change later in life. Nowadays the societal changes lead to further emphasis on children's rights, autonomy, self-determination and on the acceptance of naturally occurring developmental variations.

In 2012 the Swiss National Advisory Commission on Biomedical Ethics published its position against early irreversible interventions in order to "normalize" the aspect of the external genitalia (2).

Case Report

We report the case of a child with a 46, XX DSD due to fetal androgen excess secondary to a classical form of congenital adrenal hyperplasia (CAH) with Prader stage V virilization. Direct DNA sequencing of *CYP21A2*, showed two maternal (IVS2-13A/C>G; p.Val281Leu) and one paternal mutation (p.Arg354Cys). The interdisciplinary team took care of the patient and the family according to the consensus statement on care of DSD individuals (3). In agreement with the parents, female gender was assigned and no surgery planned until the child could participate in the decision making. When the child was 3 years old, the parents informed our team that they had met a surgeon in their home country and that a genitoplasty was planned in one month. The interdisciplinary team convened the Clinical Ethics Committee of the hospital to reflect on the situation, share the best practice and to give an advisory opinion (Figure 1). But before we could debate with the family to take a shared decision the family had left definitively Switzerland.

Figure 1: Dilemmas in decision making. Supplementary advise by the Clinical Ethics Committee intervention



Discussion and Conclusion

Our case highlights the dilemmas a team may encounter despite applying standard of care with counselling and support.

Determining the best care and interests of this child remain a big challenge and even if the local Clinical Ethics Committee was mandated in order to give an external, advisory opinion and recommended to postpone the intervention, the team couldn't share the decision with the parents.

Human rights organizations defend the children's right to physical integrity and demand to defer any "normalizing" interventions on genitalia until the concerned individual can give his consent.

However, studies showed that a majority of CAH adult women thought that genitoplasty should occur within in the first year of life. The dsd-LIFE study reported that only 0.5% of 46, XX CAH raised women had a Gender Change (GCh) after puberty (4).

Few case reports document a satisfactory outcome of male gender assignment in severely virilized CAH girls with 4% bi-gender identity or gender dysphoria (5) (Table 1).

Table 1: Gender Questionning (Gender Dysphoria GD; Gender Change GCh)

General Population	DSD patient	46, XX CAH raised men	46, XX CAH raised women
0.2-0.7% GD/Incongruence	1 % GCh after puberty (4) 3% if Turner and Klinefelter excluded (4)	4% bi-gender identity or GD (5)	0.5 % GCh after puberty (4)

Gender questioning and changes are marginally more frequent in 46, XX CAH women than in the general population, however it is essential to ensure that no-one is subjected to unnecessary medical or surgical treatment and to guarantee bodily integrity and self-determination.

It was noted that the lack of clear information about the outcomes was one of the most frustrating aspects of parenting a child with a DSD. Assessing the parents' capacity to cope with their child's difference and the perception of society, while ensuring the most open and the least prejudicial decision for the child's psycho-sexual future is primordial and still a delicate balance.

More research will be needed to develop decision support tools, to guide clinician and to base our recommendations on solid results from the DSD community.

References:

1- Sommer G. et al. Enfants et adolescents avec variations du développement sexuel. Swiss Medical Forum 2018; 18(42):858-864

2- Attitude à adopter face aux variations du développement sexuel: Questions éthiques sur «l'intersexualité». Prise de position no. 20/2012 de la Commission nationale d'éthique pour la médecine humaine, novembre 2012

3- Cools M. et al. Caring for individuals with a difference of sex development (DSD): a Consensus Statement. Nature Reviews Endocrinology 2018; Volume 14, July 2018, 415-429

4- Kreukels B. P. C. et al, on the behalf of the dsd-LIFE group. Gender Dysphoria and Gender Change in DSD/Intersex conditions: results of the dsd-LIFE study. J Sex Med 2018; 15: 777-785

5- Lee P.A. et al. Review of Outcome Information in 46, XX Patients with CAH assigned/reared Male: What does it say about gender assignement? Int J Pediatr Endocrinol 2010