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Should we question early feminizing genitoplasty for patients with congenital adrenal hyperplasia and XX karyotype?

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ABSTRACT

Background: There is a wide difference of opinion between the medical–surgical community and advocacy group regarding Disorders of Sexual Development (DSD) secondary to congenital adrenal hyperplasia (CAH) being ranked in the intersex category. This rupture is even more evident when the issue of genitoplasty is brought up. For physicians it is obvious and unequivocal that a person with CAH and an XX karyotype has a female gender identity, whereas associations tend to rank persons with CAH in the intersex category and advocate holding-off on surgical management.

Material/Methods: A retrospective case study vs. control group, spanning over 40 years, included 21 patients who were treated in 3 different centers. Each patient and their parents were contacted independently and interviewed regarding interpersonal relationships, psychological impact of genitoplasty, gender identity and opinion on optimal care management for this disorder. Three couples controls (parent–child) per CAH patients were used and matched according to age, sex assigned at birth and ethnic origin.

Results: Sex assigned at birth seemed to concord with the gender identity perceived by the patients in 85.7% of cases. In fact, 89.7% of patients and 100% of parents felt that feminizing genitoplasty should be performed within the first year of life. There is however a significant difference compared to controls who felt that surgical management should occur later on in life. No difference was highlighted during childhood regarding parents–child relationships or social integration. However, during adolescence, the parents–child relationship tended to be significantly more painful for the CAH group. Integrating their parenting role was significantly harder for patients in the CAH-DSD group. In the population of CAH-DSD patients who had genitoplasty the level of sexual fulfillment was not lower to the one reported by the control group.

Conclusion: Female sex assignment seems legitimate according to this study and the development of gender identity in these patients matches the sex assigned at birth. Resolving early on the adequacy of the genital anatomy with the sex assigned is promoted by patients as well as their parents. Proper psychomotor development and sexual satisfaction underline the absence of complications related to the surgical technique and the relevance of early surgical management.

There is a wide difference of opinion between the medical–surgical community and advocacy group regarding Disorders of Sexual Development (DSD) secondary to congenital adrenal hyperplasia (CAH) being part of the intersex category. This rupture is even more evident when the issue of feminizing genitoplasty is brought up.

Whereas for physicians it is obvious and unequivocal that a person with CAH-related DSD and XX karyotype has a female gender identity, some patient associations are in fact advocating to qualify CAH-DSD patients in the intersex category. They underline that ambiguous genital organs are not the disease but rather one of its consequences not requiring treatment.

Intersex patients' associations impose postponing surgery to an age when patients can be informed and decide for themselves which gender they would like to be assigned to. However, surgery is still regularly proposed early on in life. The ideal moment for this surgery remains debated [1].

Some seemingly disappointing long-term results of early feminizing genitoplasty [2,3] with more than one-third of intra-vaginal stenosis [3,4] challenge the early surgical management of DSD [5]. However, other authors report highly satisfactory functional, social and psychological results for this timing for reconstruction [6,7].

It is appropriate and essential to re-evaluate the need and relevance of early vs. delayed surgical management and to objectively assess the life experience of these patients.

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1. Methods

This retrospective multicenter (3 centers) case study vs. controls focused on the general impact of genitoplasty in CAH patients and their own life experience.

This study was registered with the French data protection national agency (Commission Nationale de l'Informatique et des Libertés) and was approved by institutional review boards.

All CAH-DSD children who underwent surgical management for this pathology (virilizing type) between 1970 and 1999 and were older than 15 years and 3 months (legal age of consent in France) at the time of the study were invited to participate in the study.

Out of the 28 patients contacted, 21 patients and 20 parents (1 parent per child) accepted to answer the questionnaires. 4 refused (either the patient or the parents); 3 were lost to follow-up.

For patients, mean age was 27 ± 7 years with an age range of 16–40 years. Of the 28 patients included, 4 were diagnosed with salt-wasting congenital adrenal hyperplasia, the other 24 with the simple virilizing form.

Two subgroups were constituted: early feminizing genitoplasty only (EFG) and early feminizing genitoplasty +/− redo at adolescence or late feminizing genitoplasty (LEFG).

An information letter, exposing the objectives and modalities of the research and inviting both patients and their parents to join the study, was mailed to all the patients over 18 and to the parents for underage children. In the absence of express refusal, a phone contact was established a minimum of 15 days after the date the information letter was sent.

Designed with the help of a psychologist, a directed interview with open questions focused on several elements: psychological impact of feminizing surgery, evaluation of the child's integration and well-being during childhood and adolescence within a family and friend environment as well as society integration, evaluation of the medical community and opinion on the optimal medical–surgical management for this pathology. For parents, questions focused on evaluating the potential difficulties of accepting the sex assignment of the child at birth and the perception of their child before and after surgery. For patients, we focused on how they felt about their phenotype, their gender identity, their perceived gender, as well as their sexual satisfaction. The answers of parents and children were transcribed fully, without interfering with the way they were formulated.

The answers where then sorted and classified into different categories (poor to very good; not-satisfied to very satisfied; female, intersex or male; heterosexual, bisexual or homosexual….) depending on the item related to the question (well-being, social integration, sexual preference, gender perception, external genitalia perception….)

In order to evaluate efficiently sexual satisfaction, this item was scored by sexually active patients on a scale from 0 (unsatisfied) to 10 (very satisfied).

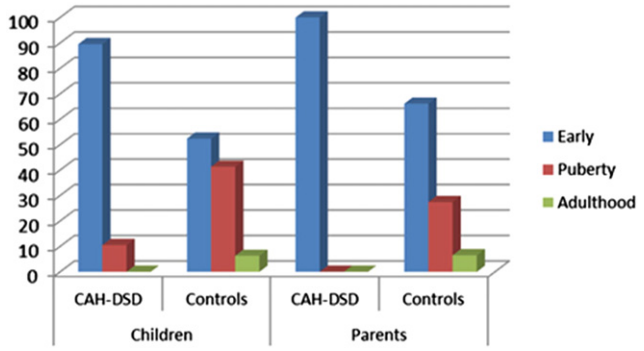

Fig. 1. Choice of the surgical timing, in percentages, according to the groups.

These same elements were respectively evaluated for control subjects, except for their perception of surgery. This point was replaced by their opinion about the best moment to operate virilized girls if needed. We included parents–child couples, with 3 control children for one patient. These control children were matched for age, sex assigned at birth and ethnic origin. They did not present with chronic pathologies or disorders that could have had an impact on social integration.

We performed descriptive statistics (mean and standard deviation for quantitative variables, total and percentage for qualitative variables). The analysis was performed with Fisher's exact test or univariate conditional logistic regression analysis according to the application conditions. The significance threshold (p) for this study was set at 0.05. The statistical analysis was computed with Epi Info® (version 3.5.4) and SAS® (version 9.3) software.

2. Results

The mean age at surgery was 2 ± 1 years in the EFG group and 13 ± 5 years in the LEFG group. The mean Prader stage was 3 (2 to 5) in the EFG group and 4 in the LEFG group ($p = 0.003$).

All patients had both reduction clitoridoplasty and vagina surgery. Vagina surgery consisted of either vagina lowering or surgical plasty for introitus.

For the control group, 63 parents–child couples were matched based on the child's age and ethnicity.

There were 11 patients in the EFG group and 10 in the LEFG group.

All the results are presented in Fig. 1.

Eighteen patients identified themselves as female (86%) and 3 patients as intersex (14 %). No patient identified with the male gender identity. All subjects in the control group were female with a significant difference between cases and controls ($p = 0.031$). No significant difference ($p = 0.612$) was highlighted between the EFG group (83% female gender, 17% intersex) and the LEFG (89% female gender, 11% intersex).

Sixteen CAH-DSD women reported being heterosexual, 2 reported being homosexual and 3 reported being bisexual (EFG group: 7 heterosexuals and 2 homosexuals; LEFG: 9 heterosexuals and 3 bisexuals). No significant difference ($p = 0.647$) was found between the EFG and LEFG groups or between CAH-DSD subjects and controls ($p = 0.155$) where we find 57 heterosexuals, 4 bisexuals and 2 homosexuals.

84% of controls reported being sexually active vs. 86% of CAH-DSD subjects; no significant difference was found between both groups ($p = 0.57$).

There was no difference regarding sexual satisfaction between the EFG and LEFG subgroups ($p = 0.719$) with for both of them 8 ± 1 out of 10. However, this value was significantly higher ($p < 0.05$) for CAH-DSD subjects vs. controls (7 ± 2).

Regarding self-assessed gender morphotype (defined as the visual gender appearance of the subject), there was a significant difference ($p = 0.016$) between controls (97% female and 3 % male) and CAH-DSD subjects (86% female, 9% male and 5% intersex), however there was no significant difference ($p = 0.165$) between the EFG and LEFG subgroups.

No difference was found between the morphotype evaluation of control parents and CAH-DSD parents ($p = 0.992$). The evaluation by the physician was significantly different ($p = 0{,}042$) than the self-evaluation by CAH-DSD patients, since the latter saw themselves as significantly more masculine.

Complete adequacy between female gender identity, patient's self-assessed female morphotype, society's evaluation of the gender identity and heterosexuality was found in 57% of cases.

Overall, 90% of CAH-DSD patients (100% EFG, 80% LEFG; no difference between EFG and LEFG; $p = 0.144$) and only 52% of controls believe that genitoplasty should be performed during the first year of life for CAH-DSD patients born with an XX phenotype ($p = 0.02$).

Furthermore, 50% of CAH-DSD parents brought up difficulties when discussing this type of surgery with their adolescent child, a time when