Male Gender Identity in an XX Individual with Congenital Adrenal Hyperplasia

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Based on their single case report, Jorge et al. advocated a reexamination of current standards of care for individuals with congenital adrenal hyperplasia (CAH) [1]. However, such reexamination has taken place at the international consensus conference on intersex management in 2005 [2], which reviewed the available data and drew the conclusion that the available evidence supports female sex assignment of 46,XX CAH newborns. Gender dysphoria and/or patient-initiated gender change occur in both female- and male-raised 46,XX individuals with CAH at comparable rates of 5% and 12%, respectively [3], i.e., do not indicate a “common” occurrence. Thus, the burden of proof cannot be shifted by a single case report. In addition, this case report has numerous complicating features that call into question the authors’ central contentions. Specifically, the child was not managed according to best practices even under the optimal-gender policy, leading to later complications.

First, despite genital ambiguity, this child was assigned male without a comprehensive medical evaluation. Second, a definitive diagnosis and associated female reassignment took place at 20 months, a problematic age for such physician-imposed gender reassignment [2]. Third, it is undocumented for this patient, whether and to what extent the parents were part of the decision process (as was already recommended by the preceding “optimal gender policy”). Fourth, Figure 4 in the article shows poor hormonal control subsequently (note that patients’ medication-compliance reports have limited validity). Thus, hyperandrogenemia persisted not only during infancy, but continued after the correct diagnosis was established. Fifth, after prior attempts at feminizing genital surgery, “successful” (authors’ evaluation) genital surgery (i.e., clitorectomy) took place only at age 11 years; until that time, an oversized clitoris or clitorophallus was present. (Note the quote from the patient on p. 128, col. 2: “I’m a boy because I have something down there that girls do not have”). Sixth, the absence of any related psychologic report in the medical chart and the patient’s depressive reaction after surgery make it doubtful that a proper comprehensive gender-identity workup and decision making involving the patient preceded the surgery. Finally, many years later, the patient initiated a social gender change to male, but without legalization (reasons not stated).

If the patient had received proper medical care at birth, later patient-initiated gender change to male might have been less likely. From the (limited) information we have on the determinants of gender change in female-assigned 46,XX CAH, it appears that both noticeable genital masculinization and hyperandrogenemia with the associated virilization in childhood and adolescence contribute [4]. An additional point concerns the severity of the CAH variant. We have documented (group level) dose–response relationships of prenatal androgen excess with the degree of masculinization of gender-role behavior in childhood and adulthood [5] and with the degree of gynephilia in adulthood [6]. The simple-virilizing variant of CAH, which was this patient’s condition, represents the lower end of both prenatal androgenization and behavioral masculinization and, therefore, appears to be an even less likely candidate for assignment to the male gender at birth than the more severe salt-wasting variant. Yet, even in the latter, female gender identity is the common outcome despite markedly masculinized gender-related behavior.

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References
Male Gender Identity in an XX Individual with Congenital Adrenal Hyperplasia: A Response by the Authors

The notion of a “common” vs. a “rare” event has always been problematic when it comes to intersexuality [1]. Even medical textbooks do not seem to agree on CAH frequencies [2]. Therefore, it is difficult to assess whether the case reported in Jorge et al. [3] is a “rare” event with regard to gender change, especially when patients who are not satisfied with treatment are lost during follow-up.

Popular and clinical wisdom assumes an “embryology of gender” as if the biologic underpinnings of gender identity formation have been uncovered. But this significant gap in the field does not allow us to understand the basis for self-initiated gender change in a proportion of intersex cases. A long-held and unjustified notion is that atypical genitalia creates problems with gender formation even though earlier versions of the Diagnostic and Statistical Manual for Mental Disorders noted that, “Physical abnormalities of the sex organs are rarely associated with gender identity disorder of childhood; when they are present, the physical disorder should be noted on Axis III” [4]. The other notion is that high androgen levels in a 46,XX background create male gender identity and lesbianism. But male to female and female to male transsexual individuals clearly illustrate that gender nonconformity can occur even when karyotype and androgen background are consistent with each other. More recent studies by Meyer-Bahlburg and collaborators reexamined the androgen hypothesis by looking at the relationships between CAH subtypes, gender formation, and sexual orientation [5,6]. However, a significant body of work during the past 50 years has consistently shown that gays and lesbians have normal neuroendocrine profiles. Nevertheless, it will be of great interest to gain further insight on this hypothesis by correlating measured androgen levels and gender identity. To date, available data do not sustain the androgen hypothesis.

A tremendous challenge in the clinical management of intersexuality is the translation of the clinical definition of “sex” in legal terms. A rise of conservative views on sex is apparent across the United States with recent discussions of same-sex marriage. The Defense of Marriage Act of the United States contrasts sharply with the European Gender Recognition Act and the Civil Partnership Act [7]. The evident ambiguous management of sex in the court system with regard to the recognition of sex change among transsexual individuals across the United States demonstrates that the fate of individual cases largely depends on jurisdiction at the state level [8]. Given that the legal system lags way behind medicine when it comes to the social management of sex, it is risky to protect a consensus statement that may have devastating legal consequences for some intersex individuals. The question that we have to responsibly ask ourselves is: What exactly can constitute the burden of proof to revise the current management of intersexuality?

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