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CLINICAL CASE REPORT SERIES

Gender Dysphoria in a 62-Year-Old Genetic Female With Congenital Adrenal Hyperplasia

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Abstract We report a case of gender dysphoria (GD) in a 62-year-old genetic female patient, raising the pros and cons of performing corrective surgery later in life. This 46,XX DSD patient was registered and reared as a girl; CAH was diagnosed late in childhood. Poor adherence to treatment and lack of proper psychological management contributed to evident GD. Living for years as a male, the patient applied for a legitimate male identification document in his late 50s; thereafter, he requested a sex-reassignment surgery "to disguise his female body upon his death." We informed the patient and family about surgery hazards, while analytical therapy allowed the group to evaluate the actual wish for surgery. When the wish was brought up, the role of death urged the group to rethink the course of treatment. During the process, it became clear that the patient's desire for surgery, more than a wish for changing the genitalia, expressed an impulse related to issues of endorsement and acceptance of his male identity. This report raises interesting questions about sexuality in a social context and prompts the idea that sexuality is broader than sex itself, raising new questions on the psychological risks faced when considering a body change after years of living with a disorder of sex development.

Keywords Disorders of sex development · Congenital adrenal hyperplasia · Gender dysphoria · Sex-reassignment surgery

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Introduction

Disorders of sex development (DSD) are "congenital conditions in which development of chromosomal, gonadal, or anatomical sex is atypical" (Lee, Houk, Ahmed, & Hughes, 2006). Several disorders may be embraced by this definition, one of which is congenital adrenal hyperplasia (CAH). CAH is a group of autosomal recessive disorders caused by enzyme defects in adrenocortical steroidogenesis.

Deficiency of 21-hydroxylase (21-OHD) is the most common form of CAH, responsible for more than 90 % of cases (White & Bachega, 2012). It results in defective conversion of cortisol and aldosterone from their precursors, leading to elevated levels of androstenedione, testosterone, and dihydrotestosterone—the main male sex hormones. In its classic form, 21-OHD manifests in utero and is associated with ambiguous external genitalia of the newborn female (White & Bachega, 2012). A distinctive steroid profile confirms the clinical impression. Conventional medical treatment consists of early glucocorticoid and mineralocorticoid replacement therapy and corrective surgery, as indicated. Bilateral adrenalectomy for CAH is still a debatable procedure that may be indicated for patients with difficult management.

Along with medical treatment, the importance of psychological care—from diagnosis to treatment—has been regularly emphasized (Hiort et al., 2014; Lee et al., 2006; Meyer-Bahlburg, 2008). Assessment of the patient's doubts, anguishes, and the emotional costs of hormonal and physical changes occurring with treatment helps elaborate and integrate psychosomatic and psychological questions (Cohen-Kettenis, 2010; Telles-Silveira, Tonetto-Fernandes, Schiller, & Kater, 2009). Understanding sexuality and the development of gender identity would assist with appropriate assimilation of the subject both in his/her family and within society. This complex situation requires a multidisciplinary approach to support both the patient and the family (Brauer, 2012; Hiort et al., 2014; Lee et al., 2006).

Dessens, Slijper, and Drop (2005) reported that 13 (5.2 %) of 250 women with CAH and female gender role assigned early in life later exhibited gender dysphoria of such severity that it induced them to seek gender reassignment. This information is crucial for assisting clinicians in counseling parents of affected newborns in gender assignment. The American Psychiatric Association's (2013) *Diagnostic and Statistical Manual of Mental Disorders* defines gender dysphoria as "a marked difference between the individuals expressed/experienced gender and the gender others would assign him or her, and it must continue for at least 6 months."

In this article, we present and discuss a case of gender dysphoria in a 62-year-old genetically female patient with CAH. We raise the pros and cons of performing genital corrective surgery later in life to change a patient's bodily appearance.

Case Report

The patient, in his late 50s, presented to the emergency department because of an adrenal crisis. He was born with ambiguous genitalia and raised as a girl in a rural area in southern Brazil. He had had a 35-year lapse in follow-up at the time of his emergency visit. Once recovered, he decided to resume follow-up. By that time, he had been able to change his ID (off the record) in the city where he was living as a male. His medical chart was then reopened using his elected masculine Catholic name; he did not mention his previous chart as a female patient.

After a few years of attending regular appointments, he unexpectedly requested a sex-reassignment surgery, claiming that it would fulfill his ultimate goal of living successfully in his desired gender role. He justified his request as a wish to disguise his female body upon his death, during preparation for burial. In his own words: "I don't want the funeral agent to find out that I was a woman upon preparing my body."

This sudden scenario urged the medical staff to reexamine this patient's full history. The diagnosis of CAH (due to the classic simple-virilizing form of 21-OHD) was only established late in childhood, when the patient was referred to a specialized medical center. At that time, glucocorticoid replacement therapy was initiated. The karyotype was 46,XX.

After years of difficult medical therapy and resistance to treatment, the patient—now in his late teens—was transferred to the surgery service, where subtotal adrenalectomy was indicated and performed. Subsequently, he received glucocorticoids and mineralocorticoids replacement therapy. However, irregular treatment persisted with a challenging follow-up, little adherence to therapy, and missed medical appointments. The patient confessed to avoidance of treatment to keep a male appearance and to prevent feminization. At that time, no specific psychological approach was available.

The patient was then lost to medical follow-up for almost 35 years. He reported that he had been living as a "man" in various locations, with no settled residence, no proper ID, and no stable job. Although living as a male, he had never had any sexual involvement.

The case of this patient was comprehensively debated within our multidisciplinary group, which led to the conclusion that his medical and psychological treatment must be observed very closely before a final decision concerning surgery could be made.

Management and Outcome

We put together a multidisciplinary team that included endocrinologists (in-training residents, post-graduates, and attending staff), urologists, psychologists, and bioethical experts to create a comprehensive treatment plan to include familial, social, cultural, and religious aspects of the patient's life. We decided on stringent outpatient follow-up, including psychological care as a priority, while maintaining an ongoing group discussion.

The psychological visits lasted 50 min on average. Consistent with the premises of short-term psychoanalytic therapy, the patient agreed to focus his appointments on the central issue in his conflict, which was his request for surgery. With this in mind, the analyst explored the conscious and unconscious motivations of the request (Fiorini, 2004; Gillieron, 1983; Oliveira, 1999). The sessions took place in parallel with his medical appointments every 3 months (more frequent sessions, although desirable, were hindered by his living far from the city). Informed written consent for the treatment plan, including possible future publication(s) of his case, individually or as part of a group, was signed by the patient following a thorough and careful explanation, according to a previously approved protocol by the ethics committee of our institution.

Once again, the patient faced therapy very reluctantly, as open conversations about his past proved extremely difficult. We therefore decided to employ a therapeutic technique that we had previously developed, referred to as "Life Lines," which, in brief, explored meaningful life facts related to his condition that would trigger free associations. Such life events included the day he gained a male identity card from the church and the moment he first voted using a legal ID with his masculine name. Generating free associations would empower him to address feelings linked to those situations and to build new meanings for them. It was not mandatory that the generated events begin at birth, and the technique did not aim at building a chronology. Instead, the process allowed us to identify and understand the patients' unconscious motives, as well as his inner conflicts.

In the process, we were able to consider the paradoxes and conflicts experienced by the patient; this understanding was enabled by therapeutic listening and by the activation of "corrective emotional experiences" (CEE) (Alexander et al., 1946). CEE are traumatic situations experienced by the patient that echo in the relationship with the therapist. The repetition of these experiences in a trust situation fosters a process of change in which the patient recognizes new possibilities to resolve his or her conflicts.

During the therapeutic process, the patient's family—to this point, vague and distant—began to attend the outpatient clinic. It should be mentioned that the patient has a younger sister with CAH (but she has never questioned her female identity) and had brothers who had a heart condition and had died. In every appointment, family members verbalized their concerns and fears that the patient could die during surgery. Each of his relatives attested to the male role occupied by the patient in their lives and that he was already "too old" to go through that procedure. The family tried to convince us that our endorsement of his request would be senseless.

We realized that there was a serious risk of family break-up if we concurred with his desire for surgery. At the same time, because of his family's position, our patient could gradually recognize his role within the family as a "household leader." Each relative conveyed the idea that their ongoing interrelations were serving to sustain and strengthen the patient's male identity, as, for example, after his brothers' deaths, when his nieces saw him as a second father.

During the psychotherapeutic process, we understood that the request to disguise his feminine body was not necessarily connected with a desire to have a penis at that moment in life, but instead with existential questions triggered by his reaching maturity, such as "Who am I in this body I inhabit? Who am I to others? What did I do with my life? What will I take away and leave behind?" In addressing these questions about life and death, inside his life history with DSD, he could redeem, reaffirm, and develop an archetypal masculine identity.

Despite all the emotions and suffering brought up by these memories, they enabled him to construct an appropriate answer to his initial questions. Along the way, he began to realize that although he had been born a "female," he had always felt a belonging to the male gender, and endorsement by his relatives made him more confident about his male identity.

Imagining what others might say about his life after his death was too distressing for the patient, as if someone could later deny who he was during his entire life, exposing his family to scorn and criticism. This feeling had accompanied him throughout his life. However, realizing that his male role was recognized by others, and that their views of him matched his sense of self as male, made him feel less threatened by others' viewpoints. His family's attitude allowed him to overcome his persecutory nightmare, so that he ultimately gave up the idea of corrective surgery.

We should stress that there was also a cultural and religious context in which the patient was accepted and also recognized as a man. During his therapy, he could acknowledge that besides his family, other people shared his feeling of belonging to the male sex. One of these people was the church's priest, who listened carefully to his story and told him that "to God, there was no distinction between men and women, and that the agreements made on earth were blessed by Him." The priest's words were based on the Book of Psalms, which the patient brought to the sessions and which helped comfort him concerning his anguish about what would happen after his death.

Discussion

The literature addresses the need to reevaluate whether all cases of CAH born genetically female (46,XX) must be reared as women (Lee & Houk, 2010; Lee, Houk, & Husmann, 2010; Meyer-Bahlburg, 2005). Recent guidelines suggest that we should individualize all cases because some patients with a late diagnosis or those with Prader 4 or 5 virilization may choose to identify as male (Lee & Houk, 2010; Lee et al., 2010). Lee et al. evaluated 12 patients with CAH reared as men. In their study, all patients had a male gender identity, appropriate social relationships, interest in women and frequent sexual intercourse.

Our patient did not have the same opportunities as the ones reported by Lee et al. He was not able to discuss his desire to behave as a male throughout his childhood, adolescence, and adult life. It was only as a senior citizen—a time in life when social responsibility declines and existential questions arise (Djordjevic, Salgado, Bizic, & Kuehhas 2014; Ettner, 2013; Wilson, Sharp, & Carr, 1999; Zucker, Wood, Singh, & Bradley, 2012)—that being accepted by others and recognizing himself as a man became an urgent matter.

This urgency was expressed through an uncommon wish, with an odd justification that was brought up to the doctors: "When I die, at my funeral, they will know who I am." When the psychologist began to see the patient, she noticed that the team did not realize how deep this question of identity was. For a junior doctor in training, the patient's rationale for his request sounded like a joke, because, as the doctor said, "When you die, it is over, no matter the gender." This particular patient and a related case later reported in the lay press (Hensley, 2014) denotes the lack of familiarity and experience that young doctors have in dealing with DSD patients and, as a consequence, the need for continuous and specific training to avoid improper attitudes.

At this moment, the role of death prompted the group to rethink the course of treatment, encouraging us to search for an answer to his request. It seemed expected that the entire medical team experienced anguish and wanted to offer a rapid decision for his request, as if time had run out, and a feeling that something that had not been offered before should be done right away (Ceccarelli, 2003). Evaluating the harms and benefits of corrective surgery together with the Bioethics Committee prompted a rethinking of the decision, helping the young endocrinology physicians to develop more appropriate reasoning.

Although technical advances allow increasingly frequent surgical procedures at any moment in life, it is also extremely necessary to evaluate the personal pathways of disease and treatment (Ceccarelli, 2003). In this particular situation, as the psychotherapy process continued, the patient himself changed

his demands as he gained a fresh perspective for facing life, even as the medical team began to rethink its interventions. In fact, a multitude of different points of view came into focus and served to reinforce the importance of listening to patients.

The multidisciplinary team that treats DSD patients with gender dysphoria needs to acknowledge that the gender category helps foster understanding of sex distinction in its own symbolic sociocultural level, "without erasing its natural-biological dimension, but realizing that the anatomy in itself does not determine gender identity and its sexual distinction between man and woman" (Meroni, 2011).

During the therapeutic process, it became clear that our patient's wish for sex-reassignment surgery reflected his need for endorsement and acceptance of his male identity by society even after his death, rather than just a desire for improvement of cosmetics or sexual function. Thus, his subjective experience of genital anatomy and gender identity incongruence—a defining concept in gender dysphoria—was not restricted to the bodily realm. Therefore, to prevent irreversible damage to the patient, we must emphasize the importance of clarifying his or her conscious and unconscious motivations before recommending sex change surgery.

Although having decided not to undertake surgery, the patient feared that, by remaining in a female body, he would be discovered by non-family members upon his death and possibly expose the entire family to ridicule and long-lasting discrimination. This worry continued as a central topic during his subsequent psychotherapeutic sessions. Therefore, we suggested alternate ways to prepare the patient and his family to face the problem, for example, by leaving a statement for close family members with instructions on handling and caring for his deceased body.

The guidelines offered by the Chicago Consensus (Lee et al., 2006) and the Swiss Bioethical Commission (Brauer, 2012) have been based on advances in human rights issues, offering the opportunity to broaden those everyday challenging questions. Sharing the diagnosis with the patients, encouraging them to express their feelings regarding treatment and the condition itself, individualizing approaches, and developing appropriate clinical methods to evaluate those cases remain continual challenges for those committed to this area of knowledge. Therefore, more long-term studies, as well as case reports, will be useful for a better understanding of the patients' points of view and for offering more appropriate tools and techniques for use in these scenarios.

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Compliance with Ethical Standards

Conflict of Interest All authors report no conflicts of interest concerning the subject of this work.

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