Gender Incongruity in Children With and Without Disorders of Sexual Differentiation

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KEYWORDS

• Gender incongruence • Gender dysphoria (GD) • Disorder of sex development
• Sex assignment • Transgender

KEY POINTS

- If gender dysphoria is suspected, early assessment by a multidisciplinary team is encouraged to reduce the many risks of delaying appropriate treatment when indicated.
- Thoughtful and sensitive communication are an important part of developing a therapeutic provider–patient relationship, because appropriate terminology regarding gender-related concerns is rapidly evolving.
- Although current guidelines are helpful, long-term studies on outcomes of current treatment practices will be necessary to determine evidence-based guidelines for the future management of children with gender incongruity.

INTRODUCTION

Providers of child and adolescent health care are increasingly asked to respond to questions and issues relating to gender nonconformity at even younger ages than in years past.[1,2] Knowledge of the terminology and sensitivity to the conditions

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[1] Please note some of the terminology in this article may not be deemed the most widely accepted at the present time, but it was chosen for consistency and smooth readability. For example, natal male and natal female will be used to describe those assigned as male or female at birth, respectively.

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associated with gender questioning are important to understand.[1,3] Pediatric endocrinologists are a natural resource for this population given their long and extensive experience of manipulating puberty in both genders, with blockade for sexual precocity, and for promotion of secondary sexual development in cases of delayed or absent puberty. Consistent with this trend, has been the recent development of the first Special Interest Group within the Pediatric Endocrine Society, for which the focus is specifically around Gender Nonconformity.

We are aware of the deleterious consequences of missing the diagnosis of precocious puberty in children while there is still time to prevent marked loss of adult height. In a similar light, it is critical to identify the transgendered adolescent before they have undergone irreversible secondary sexual development down the undesired pathway. This delay in identification will cause irreversible progression of secondary sexual development and often undesirable adult height.[4]

DISORDERS OF SEXUAL DIFFERENTIATION OVERVIEW

Disorders of sex development describe a group of conditions including one or more of the following elements: congenital development of ambiguous genitalia, congenital disjunction of internal and external sex anatomy, sex chromosome anomalies, and disorders of gonadal development. The case report included below (**Box 1**) reviews a complex DSD case of significant relevance.[5]

The diagnosis of a disorder of sexual differentiation (DSD) is usually made in the newborn period and presents as ambiguous genitalia at birth, which should be considered a clinical emergency. Three broad categories have been identified and can then be further subdivided based on findings of laboratory evaluations and imaging results. The 3 broad categories include 46, XX DSD, 46, XY DSD, and sex chromosome DSD.[6]

Normal sexual differentiation occurs in 2 phases; first, with the development of the bipotential gonad into either a testis or ovary, which is deemed "sex determination" and second, with the process of phenotypic sex development through the action of gonadal and other hormones, deemed "sexual differentiation" (**Fig. 1**).

HISTORY OF SEX ASSIGNMENT IN PATIENTS WITH DISORDERS OF SEXUAL DIFFERENTIATION

In the past several decades, many changes have occurred in the management of sex assignment of infants with DSDs. Before the 1950s, the standard protocol was to assign the infant a gender based on its "true sex," which was determined based on criteria including biological findings such as external genitalia, karyotype, or gonads. The belief at the time was that the child would lead a heterosexual lifestyle based on this "true sex" determination.[7]

Dr John Money and his colleagues suggested a different approach based on their findings. They reported that sex of assignment and rearing was the foremost prognosticator of an individual's gender role.[8] Thus, in the 1950s and 1960s, it became common to consider that infants were born tabula rasa, or as a blank slate, and that gender identity was conditioned through social and environmental influences.[9] Money's group suggested that gender role is a result of an accumulation of life experiences both learned in a casual and unplanned manner and also through explicit, repetitive instruction. These studies paved the way for recommendations to perform early sex assignment and sex-related surgical interventions consistent with the assignment that was made.[10] Another key point expressed by Money was that the psychological well-being of the child or individual should be the utmost priority. The case of John