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Psychological Outcomes and Gender-Related Development in Complete Androgen Insensitivity Syndrome

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We evaluated psychological outcomes and gender development in 22 women with complete androgen insensitivity syndrome (CAIS). Participants were recruited through a medical database ($n = 10$) or through a patient support group ($n = 12$). Controls included 14 males and 33 females, of whom 22 were matched to women with CAIS for age, race, and sex-of-rearing. Outcome measures included quality of life (self-esteem and psychological general well-being), gender-related psychological characteristics (gender identity, sexual orientation, and gender role behavior in childhood and adulthood), marital status, personality traits that show sex differences, and hand preferences. Women recruited through the database versus the support group did not differ systematically, and there were no statistically significant differences between the 22 women with CAIS and the matched controls for any psychological outcome. These findings argue against the need for two X chromosomes or ovaries to determine feminine-typical psychological development in humans and reinforce the important role of the androgen receptor in influencing masculine-typical psychological development. They also suggest that psychological outcomes in women with CAIS are similar to those in other women. However, additional attention to more detailed aspects of psychological well-being in CAIS is needed.

KEY WORDS: intersexuality; gender identity; sexual orientation; androgen; androgen insensitivity syndrome; sex differences; sexual differentiation.

INTRODUCTION

The actions of the two androgens, testosterone and dihydrotestosterone, are mediated by the androgen receptor encoded by the androgen receptor gene (Lubahn et al., 1988). Defects in this gene can result in complete or partial androgen insensitivity, the clinical manifestations of which were first described by Morris (1953). In XY individuals with complete androgen insensitivity syndrome (CAIS), the external genitalia appear to be completely female at birth. However, testicular hormones that do not act through the androgen receptor have caused regression of the Mullerian ducts, and so internal genitalia are lacking.

Because the external genitalia appear female at birth, XY individuals with CAIS are assigned and reared as girls. In addition, they have excellent feminization at puberty, because testosterone is converted to estrogen that promotes development of feminine secondary sexual characteristics (e.g., breast development). Their complete inability to respond to androgen, however, also results in reduced or absent pubic and axillary hair (Money, Ehrhardt, & Masica, 1968). Usually, the condition is detected when menstruation fails to occur and at this time undescended testes are detected and removed. Following removal of the gonads, estrogen treatment is initiated.

There is little information on psychological gender development in CAIS, and even less on long-term well-being and quality of life. Small group observations without healthy controls suggest that individuals with CAIS usually assume a core gender identity and sexual orientation in line with their female sex of rearing (Grino, Griffin, Cushard, & Wilson, 1988; Wisniewski et al., 2002). They also appear to be typically feminine in terms of marriage

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