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The management of patients with ambiguous genitalia has long been governed by several principles.\(^1-3\) First, to avoid stigmatization of the patient, gender assignment should be made as quickly as possible, preferably before hospital discharge.\(^2,4\) Second, a primary consideration in assigning gender should be the prognosis for future sexual function and fertility.\(^2,5\) Given that the prognosis is known for some causes of intersexuality, the likelihood of fertility can be deduced by diagnosis of the underlying condition. Third, gender assignment has been strongly influenced by John Money and co-workers’ proposal\(^4-8\) that gender identity at birth is highly malleable. Money emphasized the importance of early and unambiguous gender assignment.

Current pediatric endocrinology textbooks continue to include phallus size among the important considerations in assigning gender in the newborn with ambiguous genitalia. Phallus size \(\leq 1.5\) cm at term\(^5\) is considered inadequate for development of a functional penis (ability to have intercourse and to urinate standing up). Given that reconstructive surgery aimed at achieving functional female genitalia is considered the more effective alternative, XY intersexual patients with microphallus and testes are often assigned female gender. According to Money’s approach, unambiguous presentation of gender identity to the patient’s parents and, later, to the patient would ensure the likelihood of a good outcome. However, there have been few reports of long-term follow-up on the stability of gender reassignment for XY children born with functional testes and androgen receptors.

Recently Diamond and Sigmundson\(^10\) reported on the long-term outcome in the case of John/Joan. This case of a normal male infant whose penis was ablated during circumcision, and who was subsequently raised female, provided early support for Money’s ideas. However, the patient ultimately re-assigned his own gender during adolescence, leading Diamond and Sigmundson to propose alternative principles for the management of intersexual children and of boys suffering from penile trauma.\(^11\)

Shortly after Diamond’s case report, Bradley et al\(^12\) reported another case of ablatio penis at the age of 2 months. Gender reassignment was made at 7 months, and the gender identity of the patient remained female as of 26 years of age. An additional case report in the pediatric literature\(^13\) also describes long-term outcome in an XY intersex child. The condition was undetected at birth and the patient was raised as a female, but later in the teen years declared herself a boy. This case supports Diamond’s contention that the apparent sex of rearing does not suffice to determine gender identity later in life.

In the present report we describe the case of an XY intersexual with microphallus who was assigned female gender shortly after birth. The patient was raised as a female, but the patient reassigned himself as male during adolescence.

CASE REPORT

Baby G’s presentation in the newborn period has been reported previously.\(^14\) The patient’s mother took diphenylhydantoin throughout pregnancy. The infant was born at 38 weeks and was immediately recognized as having ambiguous genitalia. The phallus was considered to be consistent with a diagnosis of either micropenis or an enlarged clitoris, with a urethral opening at its base. A measure of phallus length was not reported. Gonads were palpated bilaterally in what appeared to be labia majora. No uterus was palpable on rectal examination. The newborn also exhibited several dysmorphic features, including hypertelorism, a depressed nasal bridge, and hypoplastic nails of both the hands and feet. These features were felt to be consistent with fetal hydantoin syndrome.\(^15\)

Based on the small phallus size, female gender was assigned on day 4 of life. Chromosome analysis revealed a 46XY karyotype. A cystourethrogram showed a normal bladder, no deformities of the urethra, and no mass between the posterior aspect of the bladder and anterior wall of the rectum. Human chorionic gonadotropin stimulation test was interpreted as indicating normal gonadal steroid production and normal 5\(\alpha\)-reductase activity. No change in phallus size in response to human chorionic gonadotropin was noted. A trial of testosterone was not undertaken. The parents recall being told at birth that the patient had ambiguous genitalia. On day 4 of life, female gender was assigned and the parents were informed that the patient (she) had testes that needed to be removed because of the risk of malignancy. They were advised to raise the patient as a girl, and were reassured that their child would identify herself as a girl. Years later, the patient’s mother said that she fully accepted the patient as female. However, the mother did not recall being made aware of the karyotype result. At the age of 3 weeks, bilateral gonadectomy and labial skin biopsy were performed. Pathology revealed normal infantile testes. At the age of 1 year, the patient’s physical examination showed only a slightly enlarged clitoris.

At age 10, the patient was started on estrogen replacement therapy. At age 16, she was evaluated by a gynecologist in preparation for surgery to create a functional vagina. Physical exami-
nation showed an enlarged clitoris (approximately 6 cm in length) and normal adrenarche. It was at this visit that the patient first questioned her doctors about her diagnosis. Because she did not find the information available from her parents to be sufficient, she began to seek information to better understand her condition. She decided to discontinue estrogen therapy and, at this point, was referred to our group. After our initial discussion she declared herself male and changed her name. With our assistance, she obtained insurance approval for mammoplasty to remove the mature breasts that had developed in response to exogenous estrogen. The patient was started on testosterone replacement. He responded to testosterone with an increase in phallus length from 6.0 to 7.5 cm. Throughout this time, his mother supported his decision. She sought professional counseling for herself and the patient. Conversely, the patient’s father did not accept the patient’s decision. The mother’s support of the patient contributed to dissolution of his parents’ marriage.

A retrospective history revealed that the patient had, according to his mother, always “acted like a boy.” His play had always been aggressive, and he always displayed an interest in toys that his mother considered more appropriate for a boy. Friends and family had always described the patient as a “tomboy.” He related feeling like a “boy trapped in a girl’s body” since early childhood.

DISCUSSION

The last several decades have seen marked progress in understanding the developmental biology of sexual differentiation. However, much less progress has been made in understanding what determines gender identity. Gender identification is a complex biological and psychological process that most certainly has prenatal and postnatal components, although the relationship between prenatal biological processes and postnatal psychological influences is not understood. Furthermore, there are insufficient long-term clinical data to allow understanding of the relative importance of prenatal and postnatal influences, especially in humans. Money has long contended that the brain is malleable at birth with regard to gender identity.4–8 He has vigorously counseled against sex reassignment after toddler age if at all possible.16

As noted above, Diamond et al10 recently reported long-term follow-up in a male infant with ablation penis and female gender reassignment. Initial reports indicated that the reassignment had been successful.7 This report was accepted as evidence that individuals are psychosexually neutral at birth, and that healthy psychosexual development is largely dependent on the appearance of the external genitalia combined with unambiguous sex of rearing. Only after 20 years did it become known that this patient had self-reassigned his gender as male.10 Additional case reports by Reiner et al,13 and by Gooren and Cohen-Kettenis15 described XY intersexuales raised unambiguously as females who later sought sex reassignment as males. Taken together with the present report, these cases indicate that early sex assignment as female does not ensure female gender self-identification in XY infants with female external genitalia. To explain these results, Diamond and Sigmundson cite animal studies indicating that the brain is not psychosexually neutral at birth, and that the hormonal milieu in utero may affect brain development. However, the role of in utero hormone exposure in determining gender identity is by no means established, as little information exists on the nature and timing of hormone exposure in the fetal human brain.

Approximately 1 in 1000 to 2000 newborns are considered ambiguous enough to warrant genital surgery.18 The prospective studies required to define the lifelong consequences of gender assignment in these patients have not yet been performed. Thus, individual cases have been highly influential in determining practice. Equally important are data indicating that patients with microphallus assigned male gender may have a better outcome regarding sexual function than would generally be anticipated. Reilly and Woodhouse19 showed that a group of patients diagnosed with micropenis in infancy, when reevaluated after completion of puberty, had a high frequency of normal, heterosexual function. Bin-Abbas et al20 similarly report a favorable outcome in boys with micropenis secondary to fetal testosterone deficiency who were treated with testosterone during infancy or childhood.

In light of the accumulating long-term follow-up data (as described above), our experience with the present case stimulated us to reconsider and modify our approach to gender assignment in male pseudohermaphrodites. Overall, our current practices coincide with Diamond and Sigmundson’s recommendations.11 Most notably, our experience has led us to conclude that the sex of assignment should be based on the underlying diagnosis, even if sex of rearing may not coincide with size and functionality of the phallus. Whenever possible, reconstructive genital surgery should be delayed until the patient’s gender identity can be incorporated into the decision-making process. In addition, we have undertaken to provide complete and realistic information to families regarding the potential uncertainty inherent in assigning gender in cases of newborns with sexual ambiguity. We recognize the critical importance of long-term counseling for the child with genital ambiguity and his or her family. We also recognize that Diamond and Sigmundson’s recommendations and our own decision to adopt a change in our clinical approach to patients born with ambiguous genitalia will remain controversial until long-term follow-up data are available.

REFERENCES

EXPOSING MEDICAL MISTAKES

President Clinton moved aggressively to cut down on medical mistakes at the nation’s hospitals . . . Under his proposal, reports of serious errors would go to state authorities and the public, identifying the hospitals where patients were injured. That would flag the attention of regulators and potential patients. But the reporting programs, while mandatory, would not disclose the names of individual physicians or patients . . . Mr. Clinton would spend about $20 million to create a center on patient safety to oversee the development of standardized forms for hospitals to report medical errors. The center would also organize research on patient safety and distribute information on systems that best prevent mistakes . . . [He] called for focusing on faulty systems rather than careless doctors . . . For example, if systems for administering drugs are designed carefully, errors will be caught before they harm patients . . . The importance of Mr. Clinton’s proposals is that they no longer leave hospitals solely responsible for improving patient safety. That safety will now also be a federal and state responsibility.


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