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Ambiguous genitalia: an overview of 17 years' experience

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Abstract

Aim: The newborn with abnormal genital development presents a difficult diagnostic and treatment challenge for the pediatric surgeon providing care. The purpose of this study was to evaluate the results of surgical treatment for children with ambiguous genitalia.

Patients and Methods: The records of 85 children managed surgically for ambiguous genitalia in our unit from 1988 to 2005 were reviewed retrospectively. Age at surgery, operative procedures, sex of rearing, and outcome were recorded.

Results: The intersex committee's decision concerning sex assignment was female for 62 children (75%) and male for 23 children (25%). The etiologies of children reared as female were congenital adrenal hyperplasia (n = 37), male pseudohermaphroditism (n = 12), mixed gonadal dysgenesis (n = 6), true hermaphroditism (n = 4), and Mayer-Rokitansky syndrome (n = 3). Fifteen children with male pseudohermaphroditism, 5 children with congenital adrenal hyperplasia, and 3 children with true hermaphroditism were reared as male. The mean age at surgery was 4.4 years and follow-up period averaged 7 years. Eighteen (29%) patients with feminization procedures and 8 (34%) of 23 patients with masculinization procedures experienced complications and required redo operations. Vaginal stenosis was the most common complication.

Conclusion: The surgical management of ambiguous genitalia has always been difficult, and it must be performed by skilled pediatric surgeon. Genital surgery in infancy needs to be reassessed in the light of literature findings revealing poor outcome. In patients who underwent feminizing genitoplasty, vaginal reconstruction should be delayed until adolescence to achieve better cosmetic and functional results.

Ambiguous genitalia is one of the most distressing problems encountered at birth. The overall management of an infant with an intersex disorder is complex and requires multidisciplinary approach. Genital appearance has been cited as a fundamental factor in childhood sex and psychosexual development, and has led to the current management of infant genital cosmetic surgery for genital ambiguity [1-6]. However, there is an increasing evidence that, in the long term, there is patient dissatisfaction with surgery outcomes, and debate over timing of reconstructive surgery is ongoing [1-6]. Reasons for surgery in infancy include maternal estrogen effect, better compliance with dilations, lessening of the parents' concerns regarding their anomalous child, and the assumption that the child later in life does not remember early interventions [2-5]. On the other side, irreversible infant genital surgery may have life-long impact on social, psychologic, and sexual function.

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The current article presents our experience in children with ambiguous genitale and discusses the management of intersex patients with emphasis on timing of feminizing genitoplasty.

1. Patients and methods

The records of 85 patients managed surgically for ambiguous genitalia in our unit from 1988 to 2005 were reviewed retrospectively with respect to diagnosis, age at first presentation, age at surgery, diagnostic procedures, sex assignment, surgical management, results, and complications. In addition to history and physical examination, the diagnostic studies included chromosomal analysis, urine and blood steroid measurement, pelvic ultrasound, genitogram, cystoscopy, laparoscopy, laparotomy, and occasional gonadal biopsy. All patients were evaluated and treated by a team consisting of a pediatric surgeon, endocrinologist, and pediatric psychiatrist. Sexual identity of child and psychiatric evaluation were regarded as the most important factors for sex assignment. Final decision of the sex was made by both intersex team and family. All the newborns with 46,XX congenital adrenal hyperplasia (CAH) and with 46,XY complete androgen insensitivity syndrome were raised as females. The 46,XX CAH children with delayed admission beyond infancy period were raised as male because of developed male sex identity. Most of the 5-α-reductase–deficient patients were assigned female sex, but after the knowledge revealing virilizing potential of these patients at puberty, assignment of such individuals to the female sex by surgery has been avoided and only undertaken with considerable caution and after full multidisciplinary investigation and counseling of the parents. The remaining undervirilized patients with 46,XY genotype were raised as males. In patients with ovotesticular gonad, the potential for fertility was considered on the basis of gonadal differentiation and genital development, and assuming that the genitalia are, or can be made, consistent with the chosen sex. In the case of mixed gonadal dysgenesis (MGD), prenatal androgen exposure, testicular function at initial diagnosis and after puberty, phallic development, and gonadal location were considered.

Feminizing genitoplasty in our institution is done as a 2-staged or single-staged operation depending on the level of vaginal insertion to the urogenital sinus. When the vaginal insertion is low, clitoroplasty, vaginoplasty, and labioplasty is done as early as 3 to 12 months of age. When there is a high vaginal confluence, clitoroplasty is performed early and vaginoplasty delayed until 2 to 4 years of age.

The reconstructive operations for masculinizing genitoplasty begin at 6 months of age and consists of orchiopexy, correction of chordee, staged hypospadias repair, and removal of müllerian structures and insertion of testicular prosthesis when necessary.

2. Results

The intersex committee's decision concerning sex assignment was female for 62 children (75%) and male for 23 children (25%). The etiologies of children reared as female were CAH (n = 37), male pseudohermaphroditism (n = 12) and MGD (n = 6), true hermaphroditism (n = 4), and Mayer-Rokitansky syndrome (n = 3) (Table 1). The diagnoses of the 12 children with 46,XY genotype and reared as female were complete or partial androgen insensitivity syndrome (n = 8), α-reductase deficiency (n = 4). Fifteen children with male pseudohermaphroditism, 5 children with CAH, and 3 children with true hermaphroditism were reared as male (Table 1). The mean age at surgery was 4.4 ± 2.3 years, and follow-up period averaged 7 years. Five children with CAH were reared as male because of late admission and psychiatric consultation, which demonstrated dominant male behaviors. In addition to correction of chordee and staged hypospadias repair, müllerian ductal structures were resected by laparotomy (n = 2) or laparoscopy (n = 3), and testicular prostheses were placed in all 5 children.

Of the 62 children reared as females, 36 (58%) required clitoral reduction plus perineal vaginoplasty, 14 (22%) had both clitoral reduction and pull-through vaginoplasty, and 12 (20%) had colovaginoplasty.

Twelve children with male pseudohermaphroditism were given a female sex assignment and underwent gonadectomy, clitoral reduction, and vaginoplasty, and the remaining 15 children were given male sex assignment and underwent orchiopexy, correction of chordee, and staged hypospadias repair. All children with gonadal dysgenesis and 4 children with true hermaphroditism (TH) were raised as girls, and 3 children with TH were raised as boys. Ovotestes were removed and gonads and müllerian structures were either remained or removed because of the given sex assignment.

Eighteen (29%) of 62 children with feminization procedures and 8 (34%) of 23 children with masculinization procedures experienced complications and required redo operations. Among 50 patients who underwent clitoroplasty, cosmetic appearance was excellent in 35 (70%) patients, satisfactory in 11 (22%) patients, and poor in 4 (8%) (atrophic clitoris in 2 and clitoromegaly in 2) patients. The

Table 1 The distribution of etiologies in female and male sex assignment groups

Diagnosis	Female sex assignment (n = 62)	Male sex assignment (n = 23)
CAH	n = 37 (60%)	n = 5 (21%)
MPH	n = 12 (20%)	n = 15 (65%)
MGD	n = 6 (10%)	–
TH	n = 4 (6%)	n = 3 (13%)
Mayer-Rokitansky syndrome	n = 3 (4%)	–

MPH indicates male pseudohermaphroditism.