Journal of Pediatric Urology (2017) 13, 28.e1–28.e6

Prospective assessment of cosmesis before and after genital surgery

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Keywords
Disorder of sex development; Cosmesis; Genitoplasty

Received 18 May 2016
Accepted 31 August 2016
Available online 8 October 2016

Summary

Introduction
Little data exist about the surgical interventions taking place for children with disorders of sex development (DSD). Most studies that have evaluated cosmetic outcomes after genitoplasty have included retrospective ratings by a physician at a single center.

Objective
The present study aimed to: 1) describe frequency of sex assignment, and types of surgery performed in a cohort of patients with moderate-to-severe genital ambiguity; and 2) prospectively determine cosmesis ratings by parents and surgeons before and after genital surgery.

Study design
This prospective, observational study included children aged <2 years of age, with no prior genitoplasty at the time of enrollment, moderate-to-severe genital atypia, and being treated at one of 11 children's hospitals in the United States of America (USA). Clinical information was collected, including type of surgery performed. Parents and the local pediatric urologist rated the cosmetic appearance of the child's genitalia prior to and 6 months after genitoplasty.

Results
Of the 37 children meeting eligibility criteria, 20 (54%) had a 46,XX karyotype, 15 (40%) had a 46,XY karyotype, and two (5%) had sex chromosome mosaicism. The most common diagnosis overall was congenital adrenal hyperplasia (54%). Thirty-five children had surgery; 21 received feminizing genitoplasty, and 14 had masculinizing genitoplasty. Two families decided against surgery. At baseline, 22 mothers (63%), 14 fathers (48%), and 35 surgeons (100%) stated that they were dissatisfied or very dissatisfied with the appearance of the child's genitalia. Surgeons rated the appearance of the genitalia significantly worse than mothers ($P < 0.001$) and fathers ($P \leq 0.001$) at baseline. At the 6-month postoperative visit, cosmesis ratings improved significantly for all groups ($P < 0.001$ for all groups). Thirty-two mothers (94%), 26 fathers (92%), and 31 surgeons (88%) reported either a good outcome, or they were satisfied (see Summary Figure); there were no significant between-group differences in ratings.

Discussion
This multicenter, observational study showed surgical interventions being performed at DSD centers in the USA. While parent and surgeon ratings were discordant preoperatively, they were generally concordant postoperatively. Satisfaction with postoperative cosmesis does not necessarily equate with satisfaction with the functional outcome later in life.

Conclusion
In this cohort of children with genital atypia, the majority had surgery. Parents and surgeons all rated the appearance of the genitalia unfavorably before surgery, with surgeons giving worse ratings than parents. Cosmesis ratings improved significantly after surgery, with no between-group differences.

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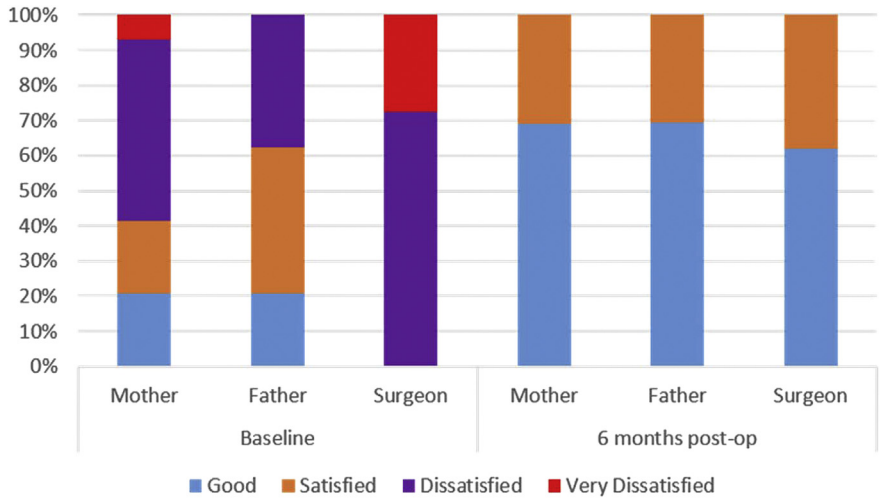
http://dx.doi.org/10.1016/j.jpurol.2016.08.017



Summary Fig postoperative cosmesis ratings at baseline and 6 months.

Introduction

Disorders or differences of sex development (DSD) are conditions in which the chromosomal, gonadal or phenotypic sex is atypical [1]; atypical external genitalia are often the presenting feature. The incidence is reported to be 2 per 10, 000 births [2]. It is recommended that genital surgery for a child raised as a female only be considered in cases of severe virilization (Prader 3—5) and that surgery of the clitoris not be performed for reasons of cosmetic appearance alone [1]. For patients with congenital adrenal hyperplasia (CAH) with severe virilization raised as a female, it is recommended that 'clitoral and perineal reconstruction be considered in infancy' and those with a low vaginal confluence undergo vaginoplasty at an 'early age'; the appropriate timing is less certain for those with a higher vaginal confluence [3]. Whether or not to perform clitoroplasty for children with a large clitoris raised as a female is controversial, and the rates of and rationale for clitoroplasty across institutions are unknown. Recommendations for masculinizing genitoplasty are vague [4]. Hypospadias repair is more successful if performed in pediatric rather than adult patients [5]. The current rates of surgery in the United States of America (USA) among children with atypical genitalia are unknown, as there have been no multicenter, prospective studies on this topic. The frequency of surgery and types of procedures performed may differ from prior eras; this is due to changes in attitudes regarding surgery and sex assignment, and because preferences for different procedures and advances in surgical techniques also change over time.

Cosmetic outcomes after genitoplasty are variable [6—12]. Most studies have included only cosmetic and functional outcomes rated by a physician [7,8,10—12], and satisfaction ratings may differ between patients and physicians. Studies that include cosmetic ratings by affected adults are scarce [9,12], and fewer exist among adolescents [7] or children [11], with most being small, retrospective and single center [7,9,10,12].

The present prospective, observational, multi-center study aimed to: 1) describe the frequency of sex assignment and types of surgery performed in a cohort of patients followed with moderate-to-severe genital ambiguity at DSD centers in the USA; and 2) prospectively determine cosmesis ratings by parents and surgeons before and after genital surgery. It was hypothesized that there would be no difference in baseline ratings between parents and surgeons, but that ratings by surgeons would be more favorable than those by parents at the 6-month postoperative visit.

Materials and methods

Participants

Participants included children from 11 children's hospitals in the USA with programs that specialize in DSD care, they were: Children's Hospital, Colorado; University of Oklahoma Health Sciences Center; St. Louis Children's Hospital; University of California, San Francisco; New York Presbyterian Hospital; Boston Children's Hospital; Lurie Children's Hospital of Chicago; Women and Children's Hospital of Buffalo; Children's Hospital of Philadelphia; Children's Hospital of Michigan; and Cincinnati Children's Hospital Medical Center.

Inclusion criteria were: moderate-to-severe genital atypia, as defined by a Prader rating [13] of 3—5 in a 46,XX child; or a Quigley rating [14] of 3—6 in a child with 46,XY or 45,X/46,XY chromosomal complement (see Supplemental Fig.); age <2 years; and no prior genitoplasty at the time of enrollment. Exclusion criteria were: infants and children with malformations of organ systems other than urogenital, and families with a limited comprehension of either English or Spanish.

If surgery was performed, the baseline visit occurred prior to surgery, and the postoperative visit was 6 months after surgery (or after the initial surgery if multiple or staged procedures were planned). If no cosmetic surgery was performed, the follow-up visit occurred 6 months after the baseline visit. The first baseline visit was performed in 2013, and baseline data on parent psychosocial functioning have previously been reported [15].