

Psychosexual development in genetic males assigned female: the cloacal exstrophy experience

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Part 1. The social experience of psychosexual development in aphallic males

Background

Psychosexual development is a species-centered conception of human development that encompasses sexual differentiation, sexual development, sexual behavior, gender-typical social affiliation, pairing, reproduction, and parental care. It is affected and effected by neurobiologic processes that interact with general and specific environmental influences [1–9]. Similarly, it affects and effects neurobiologic processes and the environment within which it is expressed. It is not specific to postnatal life: prenatal psychosexual development influences and is influenced by central nervous system development and its neurobiologic underpinnings within a given uterine environment [1–5,8–12]. Few comprehensive studies of psychosexual development in typical children exist; child psychiatry texts, for example, tend to focus on atypical gender behaviors [13,14]. This unidimensional approach ignores the broader and richer dimensions of human sexuality, which pervasively influences individual internal and external perspectives while permeating personal attitudes, interests, imagery, and behavior.

If typical psychosexual development has been understudied, research frequently focuses on children who have anomalous genitalia [3,9,11,15–22]. There is nothing particularly novel about an interest in genital anomalies. Voltaire reviewed the Ancients' discussions of genital atypia and contemporary French

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social and legal conundrums that were engendered by such anomalies. Similarly, Foucault [23] noted that until the end of the eighteenth century in Europe, those who had anomalous genitalia violated European civil and religious codes of “fulfillment of matrimonial relations” by virtue of their anatomic insufficiencies. Kuhnle and Krahle [24] discussed multi-culture social and legal conceptions of gender identity relative to genital anomalies over the last century.

Modern medical approaches to genital atypia are important to an understanding of psychosexual development in the experience of children who are born with anomalous genitalia. Termed “ambiguous genitalia” since the mid-twentieth century, genital atypia has its written origin in the ancient Greek myth of Hermaphrodite, whose birth esoterically represented not so much ambiguity as an ambivalence regarding the relative human values of virtue versus beauty [25]. Ironically, it is twentieth century intellectual ambiguity and ambivalence among groups who formulate varying gender constructs that have engendered intellectual distrust while influencing medical interventions. Recent historical sociopolitical conceptions in intersex studies sometimes straddled psychologic ideals or ideology rather than aspiring to explicate mechanisms of sexual dimorphism, whether in terms of form or of function [26,27]. Yet the indirect politicization of research has encouraged secondarily the studies of children with genital atypia.

Twentieth century sex-assignment of the newborn: historical shifts in the treatment paradigm

Whether driven partly by social forces or by specific intellectual curiosities, clinicians in the second half of the twentieth century began to formulate interventions for children who had intersex. Despite apparent philosophic acceptance of ethical guidelines from the Nuremberg Code, the Helsinki Accord, and the Belmont Paper [17,28–31], clinical formulations seem to have eschewed largely animal studies of sexual dimorphism [4,6,7] in favor of surgical interventions that focused on the children themselves. Such surgical constructions required that a sex be assigned.

In the 1950s, Money [11,12] observed that some intersex adults seemed to reaffirm a gender identity that was in concert with their sex of rearing, regardless of their genetic sex. He reasoned that sex of rearing, if assigned early enough in childhood and if anatomically correlated with appropriate genitalia, is the most important determinant of gender identity. Influenced initially by Money’s work, varying clinical algorithms gradually evolved into a treatment paradigm [12,18, 20,26,32–36]:

- Assign neonates who have anomalous genitalia to female sex-of-rearing if they are genetically female or if they are genetically male but do not have an adequate phallus
- Perform feminizing genitoplasty
- Instruct parents to inform no one of this birth history

The paradigm emphasized the clinical usefulness of sex assignment; feminizing genitoplasty was easy, whereas virilizing genitoplasty was not feasible. A sense of urgency or emergency in sex assignment and surgical intervention was added to the algorithm later.

There are no life-threatening concerns that are relative to sex assignment; outcomes of sex assignment for genital form or function and for psychosexual development were not known and could not have been predicted [17,20,26, 28–31,37,38]. In addition and despite the developing clinical paradigm, indications for sex assignment varied among academic centers. Finally, the ramifications of such neonatal interventions arguably impact the adolescent or the adult more profoundly than the child.

Some studies did not support the concept that form and function plus the appropriate environment were necessary, or even sufficient, determinants of gender identity or gender role. For example, in 1979 Imperato-McGinley et al [39] reported that genetic males who had 5 α -reductase deficiency and were reared female commonly could transition to male during puberty. Grumbach's [40] group noted that genetic males who had micropenis fared tolerably well when reared male. Berenbaum et al [1–3,41] reported male-typical behaviors and attributes, but not gender identity, in genetic females who were exposed to prenatal androgens from congenital adrenal hyperplasia. Diamond and Sigmundson [42] and Diamond [43] argued that sexual identity could not be created. Sexual orientation toward females in children who were exposed prenatally to androgens is not uncommon, regardless of karyotype or sex-of-rearing [15,16,32,40,42, 44,45]. There were sporadic reports of children or adults who self sex-reassigned from female to male; this contradicted their sex of rearing [26,32,42,46].

Cloacal exstrophy: the clinical intersex paradigm is expanded

Assessment of children who have anomalous genitals can be problematic. First, most studies involve nonhomogeneous patient populations who have variable diagnoses and poorly-defined phenotypes. Second, population diagnostic variability often precludes precise method design. Third, interventions generally have not been defined precisely.

The syndrome of cloacal exstrophy can provide assessment data of children with interventions for anomalous genitalia. It is a small, but homogeneous, population that has severely inadequate genitalia and experience similar interventions. As a devastating pelvic field defect that results from early abnormal embryogenesis, cloacal exstrophy is characterized by (1) a wide pubic symphyseal diastasis; (2) an omphalocele; (3) foreshortened small bowel with ensuing short-gut syndrome; (4) an enterovesical (or “cloacal”) exstrophic admixture (ie, a markedly foreshortened hindgut situated between and two hemibladders all with failure of anterior fusion, so that the intraluminal surfaces of hindgut-bladder plus urethra open to the external body wall); (5) imperforate anus; (6) lower vertebral deformities; and (7) severe inadequacy or absence of genitalia. Before 1959, survival was apparently nil; with vast improvements in neonatal

medical and surgical care in the last 30 years approximately 90% or more babies who are born with this defect survive into adulthood [47]. In the male, severe genital structural abnormalities occur despite normal male prenatal sex chromosome activity, testicular development, and androgen activity [48,49]; ultimate clinical success (that is, routine survival) demanded addressing the aphallia in the males.

Cloacal exstrophy is rare; it occurs in approximately 1 in 400,000 live births with about a 2:1 male:female ratio. In the United States, therefore, one might expect the birth of about 10 children who have cloacal exstrophy yearly; six or seven of these would be males. Because the penis is seriously inadequate or absent, clinical recommendations for the past 25 years have included sex assignment of males at birth to females socially, legally, and surgically through neonatal bilateral gonadectomies and surgical feminizing genitoplasty. These recommendations remain common today [50,51] but outcome studies are unavailable.

These genetic males experience a male-typical prenatal hormonal milieu [48] and sex chromosome-related properties of neuronal expression [10,52]. Thus, neonatal intervention in these children allows the study of psychosexual development—not in terms of intersex, these children experience an induced postnatal hypogonadal state—in terms of genetic and hormonal prenatal males who are castrated at birth and reared female. Studies of these children are incomplete, methods are not adequate, and there is considerable longitudinal assessment that has yet to be completed. Data about psychosexual developmental patterns in these children is especially important in the context of clinical experience and clinical research about genital atypia and relates to the clinical treatment decision-making conundrum that exists [47,49,53].

Early clinical experience with aphallia and cloacal exstrophy

Before the 1950s little was written about or done surgically for the rare child who had aphallia. Clinicians were daunted by the thought of any interventions; de novo penile construction generally was futile and sex assignment to female had not reached clinical consciousness [36]. Generally, multi-disciplinary approaches were unavailable. Initial interventions in children who had cloacal exstrophy focused on the general health of the neonate, and, if the child survived, on pelvic organ reconstruction. Survival as a reality—the first survivor apparently was born in 1959—forced clinicians and parents to focus on the aphallic status of the male. By the early 1970s, clinical expediency seemed to indicate sex assignment to female.

Money's work and early influence

It was from a clinical vacuum that Money initiated his work on profiles of adults who had intersex in his Harvard doctoral study [54]. Later, with broadening clinical exposure at the Johns Hopkins Hospital, Money [12] initiated

studies of children who had intersex and others who had a variety of diagnostic sexual categories. He formulated the idea that gender identity was fluid, perhaps even plastic, at least during childhood. Even if some gender behaviors probably were influenced by prenatal androgens, children could be assigned gender during early childhood; a child's growth and development in this assigned gender would be successful if the environment prescribed was coupled with correspondingly appropriate genitalia.

One of Money's first clinical cases, however, involved a male toddler who did not have intersex but had a traumatic loss of the phallus. Reports of success with this case encouraged the application of the early clinical paradigm to cloacal exstrophy. There was clinical frustration with aphaallic neonates and clinical anecdotes had been circulating that adult aphaallic males would be sexual misfits, chronically psychosexually frustrated, or suicidal; if assigned female, the children would grow up and do well. The veracity of these claims was unquestioned. Some studies cast doubt about Money's idea; they were, however, apparently disregarded by the developing paradigm [39,40]. This new approach seemed a boon to clinicians; construction of genitalia that were female in form and in function allowed clinicians, parents, and the children to meet developmental "requirements" of gender and genital identification as well as normal sexual function, sexual relationships, and marriage.

The experiences and influence of the clinicians

The clinical paradigm expanded well beyond Money's tenets. The notion that gender-typical behaviors and gender identity could be products of surgical and social technique was appended to the clinical teaching repertoire as spoken truths of clinical practice. This required a conception of the newborn as a "tabula rasa," or blank slate, and of gender identity and gender-related traits as largely social domains. Additionally, a sense of urgency or emergency was appended to the paradigm and often is emphasized in the literature and clinical teaching [32,51,55]. Data remained sparse.

Earlier clinical frustration at the inability to intervene with aphaallic male newborns was transformed into a utilitarian clinical approach. The blank slate conception became the floor of a clinical premise; the success of the surgical and social prescription of sex assignment of males to female would be demonstrated by successful surgical outcome and by the transformation of a male infant into a female throughout child and adult life. This intervention model—although it preceded research, so that the model was in search of data [56]—became definitive treatment. It also transformed a broad psychologic conception into an inflexible clinical concept.

This new science filled clinical, parental, and related social vacuums as it fulfilled parental needs and the needs of the child. Or did it? Is the penis so critical that a male who grows up without one cannot function or adapt psychosocially as a male? Is a female so simplistic in nature that surgical and social prescriptions can create one?

Evolving hypotheses and paradigms: early clinical experience and clinical research

Sex assignment of genetic males who have cloacal exstrophy to female often is said to be the only realistic approach [47,51]. This implies that sex assignment is successful—that the requisites of form and function are achieved. The report by Schober et al [53] that discussed briefly the psychosexual development in their cohort of 14 genetic male subjects who had cloacal exstrophy and were sex-assigned female, might seem to corroborate success. In a phone interview, their subjects reported having a female gender identity; however, the investigators admitted that their subjects had a male-typical gender role and were “too young” to identify sexual orientation or sexual activities adequately. The methods that they used in their study are not provided.

Most clinicians’ early experiences with reconstructed patients might support the notion of successful sex assignment. During the first few years of the patient’s life, when clinical involvement is most intense, the clinician essentially is dealing with a preverbal child. Additionally, the literature commonly refers only to Money’s early case [12,34]. The first survivor of cloacal exstrophy, from 1959, who preceded the clinical paradigm is alive, healthy, educated, working, and married [57]. His outcome has neither been studied nor described.

Cloacal exstrophy research

The author’s long-term study of genetic males who have cloacal exstrophy began in 1993 after extensive surgical experience with children who had genital anomalies. Twenty-nine genetic males have been assessed and followed in studies of gender identity. Twenty-seven have cloacal exstrophy and two have classic exstrophy which is a less severe anomaly with involvement of only the anterior half of the pelvic structures so that there is a large pubic symphyseal diastasis and the bladder, urethra, and penis are exstrophic (the penis is broad and markedly short and the males are nearly always reared male). The ages of the patients range from 2 to 22 years and the length of follow-up ranges from 2 to 9 years. The subjects who have cloacal exstrophy have been reported on elsewhere [58]. Twenty-four of the 29 subjects were sex-assigned female at birth (including both who have classic exstrophy), whereas parents of the other five subjects refused sex assignment to female. Of the 24 subjects who were sex-assigned female, surgery with feminizing genitoplasty occurred in the first month of life in 22 of the subjects and by 3 months of age in the other 2 subjects. All subjects experienced surgical reconstruction of the pelvic organs between 1 and 6 months of age; all but one underwent anti-incontinence surgery by age 16 years.

All initial interventions occurred at a small number of academic pediatric medical centers. Each subject ultimately was referred to one of the author’s two institutions for urologic assessment. Each subject-family consented to join the study at that time. Twenty-seven subjects remain in the study longitudinally; one

Table 1
Female sex assignment and declared gender

	Number of subjects	Age range (years)	Sex surgically assigned 14 days	Sex surgically assigned 60–90 days	Gender at initial assessment (number)	Gender at follow-up (number)	Declared gender at present (number) ^a
Cloacal exstrophy (reared female)	22	5–20	20	2	Female (19) Male (3)	Female (12) Male (9)	Female (11) Male (10)
Cloacal exstrophy (reared male)	5	2–19	NA	NA	Female (0) Male (5)	Female (0) Male (5)	Female (0) Male (5)
Classic exstrophy	2	5–22	2	0	Female (0) Male (2)	Female (0) Male (2)	Female (0) Male (2)

^a One is undeclared at age 15 years.

dropped out after 2 years and one died at age 16 years. Parents of all subjects reported that they had been instructed to tell no one, especially the subject, about the sex assignment history. Twenty of 24 subjects who were sex-assigned female have sisters. Parents reported that they reared the subjects and their sisters as similarly as feasible, in terms of temperament and personality.

The five subjects who were reared male live as male. Of the 24 subjects who were sex-assigned female, 13 have declared themselves male; however, the parents of two of these subjects persistently reject their child's declarations, but both subjects identify as male when they are away from home. Seven of the female-assigned subjects declared themselves female and two others have refused persistently to declare gender identity (although both, at age 15 years, recently agreed to estrogen administration). One additional subject, who is 7 years old has been in transition to male identity and has stated male-preference repeatedly; one female-assigned subject is deceased. Gender identity refers to a subject's persistent specific gender identification. Table 1 lists the subjects' sex assignment, transition, and declared gender identity.

Six of the 24 female-assigned subjects spontaneously declared male identity without knowledge of birth status, whereas seven others declared themselves male after being informed by their parents that they were born male (a breach of treatment protocol). One additional female-assigned subject who was told of her birth status by her parents refuses to discuss gender identity, although she recently agreed to estrogen administration (at age 15 years).

Psychosexual development in qualitative terms

Several psychosexual developmental features stand out in the study subjects who were raised male. All 29 subjects demonstrated marked male-typical toy choice, games, activities, interests, attitudes, and preference of friends (by gender). Seven subjects (all declaring male) who were old enough or willing to identify sexual orientation stated attraction to females only; no subject who declared female gender identity discussed sexual orientation. All subjects who are living as male use male names, male facilities, and male legal registrations, whereas all subjects who live as female use female names, facilities, and legal registrations. All subjects who live as male or declared themselves to be male (except for the 2-year-old) stated a strong preference for a penis.

Of the 24 subjects who were raised female, all reported an aversion to strictly feminine attire; parents of all 24 subjects reported considerable difficulty in dressing their children in feminine clothing after the age of approximately 3 years. Parents reported that by ages 3 to 4 years, female-assigned subjects were beginning to display notable male-typical behaviors and interests that were distinct from their female siblings (if present). The subjects who were reared as male had no history of gender atypical behaviors. Parents of genetic females who have cloacal exstrophy reported no atypical gender behaviors.

Subjects who were reared as male or who transitioned to male have been open about their sexual and gender identity and role, sexual activity and behavior, and

sexual interests, even if their parents were present. Six of the 7 oldest subjects who were living as male, but none who were living as female, have dated girls. Each of the subjects who live as male desires a penis as do the two whose parents reject their declarations of male identity. Transitions to male have gone well except for one 12-year-old, whose testosterone was withheld by his family until his incarceration at age 18 for a strong-armed robbery. Nevertheless, he was sexually active with girls beginning at age 15 despite incontinence and feminized external genitalia.

Parents of the subjects who live as male noted that their children are happy. Of those who transitioned to male, parents noted that their children were happier than before and seemed to be more socially involved as time passed. The children described feeling more comfortable socially, whereas they had difficulty fitting in with girls before the transition. Parents of subjects who live as female noted that their children are happy. These subjects were not as socially active as their male counterparts, especially into mid-adolescence. Generally, they reported having difficulty fitting in with girls as they grew older.

Research directions: implications for intersex studies

Psychosexual development in the study subjects who had been sex-assigned female has been a complex process. Despite the absence of postnatal and pubertal testosterone surges, many seem to have recognized a male sexual identity independent of genitalia, social and legal constructs, and child rearing.

Of the subjects who continue to be reared as girls, parents have been conscious of their child's male-typical behaviors and attitudes. They have observed no noticeable change in behaviors or attitudes since the initiation of estrogen therapy. Many parents are worried that the subjects will discover their genetic-male birth history, which will lead to their child's transitioning to male. Many parents actively maintain secrecy.

Fetal exposure to inappropriate androgens (in genetic females) or inadequate or ineffective androgens (in genetic males) leads to unpredictable degrees of masculinization of gender role [3,41,58]. Even more unpredictable is sexual identity and gender preference. Although cloacal exstrophy is not an intersex condition, for several reasons these studies have implications for psychosexual development in intersex, especially in children who were sex-assigned female: (1) the cloacal exstrophy population essentially was uniform, with diagnostic homogeneity, and experienced uniform interventions; (2) the sample size was substantial; (3) the sample is being followed longitudinally; (4) genetic, epigenetic, and major environmental factors have been arguably similar; and (5) postnatal androgens were precluded. Within study limitations, data imply that prenatal androgens influenced these study subjects' activities, attitudes, and interests, and, for many, their sexual identity, sexual activities, and sexual orientation.

Until systematic studies are available for children who have anomalous genitalia, informed consent requires parental education about the knowledge of prenatal androgen effects and an understanding of our lack of knowledge of its

global effects on child, adolescent, and adult development. Purely reductionistic or restrictive views of the affected child (eg, how the genitalia appear or might appear after surgical interventions or whether the child may be fertile in a given sex assignment) need to be subordinated to quality-of-life outcomes. Psychosocial and psychosexual development must be considered major parameters of health care-related quality of life in these children. Systematic multi-method outcome studies need to include diagnosis, standardized treatment protocols, and quality-of-life outcomes [33,59]. Finally, clinician and parents may have to deal with what appears to be clinical ambiguity, at least until the child can communicate their identity and its ramifications.

Part 2. The child's experience of psychosexual development: genetic males who have cloacal exstrophy

The continuum of development

Development as a whole is beyond the scope of this article, but certain generalities are important for this discussion. First, genetic and epigenetic processes underlie developmental potentialities [60]. For example, aspects of central nervous system genetics are necessary for appropriate development of brain structure and physiology as well as for interneuronal connectivity and signaling systems. Second, the brain is a systems organ; interconnectivity is crucial to specific aspects of function (eg, sexual function), dimension (eg, personality development), and even to some psychiatric vulnerabilities. Third, interactions of genetic processes with prenatal and postnatal environmental effectors, mediators, and moderators often determine whether genetic potentials are achieved or altered, sometimes predictably but often unpredictably. Uncertainty is plentiful and may contradict inflexible clinical approaches. Outcome studies can help to gauge predictability and translate data into new clinical algorithms or, at least, treatment guidelines.

The cloacal exstrophy experience

Children who have cloacal exstrophy experience a prolonged, complex developmental trajectory that encompasses pathophysiologic realities that typically transform the child/mother relationship into a dependency that is unique, even for children who have chronic illness. In the first year or so of life, the child's specific needs relate mostly to survival. Typically, these needs influence early parenting role and parenting style. Similarly, parental needs and anxieties influence the child's environment and parents and patients develop intricate relationships with their physicians. Parental and child anxieties tend to be high. The roles that these and other environmental issues play in the child's overall psychosocial development are unclear.

Issues of parent-child dependence often may become muddled in the uniquely interwoven threads of the realities of their lives. The parental influence in raising the genetic male as a female is difficult to know or interpret. Parent-child interactions may be unusual or unexpected. Although there may be value in assessing the parenting of these children as female, there is no systematic approach for measuring this retrospectively.

Parents of all subjects in these studies reported good social functioning, in general. The subjects tended to have friends, even if most of their friends were male; they tended to do fairly well in school; and they showed little disruptive behavior. For the children who transitioned to male, parents and children reported improvement in overall psychosocial functioning. This included improved mood and affect, relations with girls as a group and individually, participation in social activities, and acceptance of medical and health-related demands. Illness behavior may have begun to favor more physical and social independence developmentally among those transitioning to male, after transition.

Choice versus recognition: sexual identity

What may be most interesting in subjects who transitioned to male is the ease with which the transition seemed to progress. Each child who transitioned, plus the two whose parents refused such transition, seemed, in some manner, to recognize their male identity. The six who spontaneously declared themselves as male, without birth information, could not explain fully how they recognized their male identity: “I just know” was the remarkably similar statement from each. All 13 subjects who transitioned to male made a declaration of identity that refuted social and genital indications and familial constraints, apparently as an act of self-awareness; these children seem to have intuited their maleness.

What was ease of transition for the children was emotional turmoil and social angst for their parents. The complexity of social and extended family relationships, intermingled with parental dimensional realities and nuclear family demands, created periods of unease and occasional personal upheaval, especially for the mothers. Mothers of most sex-assigned subjects expressed guilt over their child’s life-course as they feared that their child would discover his/her birth status. Parents of those who transitioned to male admitted that they were completely unprepared for their child’s declaration or their transition to male identity, although most had considered the possibility.

The children themselves seemed to proceed with their transition despite, of perhaps without fully recognizing, their parents’ turmoil. By parental observations, at times the children seemed to be almost empowered by their declaration. The following transcript is a small part of the follow-up audiotape of a 13-year-old patient. His initial assessment in Baltimore had taken place 10 months earlier. Two months later his parents informed him (the night before school started) that he was born male. Following this disclosure, he transitioned

to male, including choosing a boy's name, almost immediately. The discussion demonstrates a scenario that is typical of many of these children:

Q: What does that tell you, that you can figure out that you are a boy even though everyone expected you to be a girl? You were raised a girl?

BJ: I had a boy's mind. . . . [By the way,] how did you all get me to go over there? What did you tell me that I was going over there for?

Q: You mean to Baltimore? I don't know. Your mom and dad did that. I don't know what they said. Were you scared?

BJ: I thought it was just like a little vacation to go talk to a doctor. I don't remember what they told me.

Q: What was it like? Going to Baltimore and talking about all the stuff that we talked about.

BJ: It was weird. You were asking me all of these things and what did that have to do with anything? I did not know. I did not have the slightest clue. It was silly.

Q: It really did not fit into anything to you? It just did not make sense?

BJ: If you had told me [I was born a boy] and then you had talked about it, then it would have made sense to me.

Q: I could not tell you.

BJ: Well, it did not make sense to me. . . .

Q: How long did it take you when your mom and dad actually told you [that you were born a boy, 2 months after the initial assessment], how long did it take you to realize that they were telling you the truth and all of that kind of stuff?

BJ: Right when they told me, because now everything made sense.

Q: From your past you mean? Why you like girls [sexually] instead of boys and why you did not wear dresses and things?

BJ: I knew that it was true. Even if it had been another person that had told me I would have believed them because it made sense to me.

Q: You mean if it had not been somebody in your family?

BJ: I would have believed them.

Q: And you would have gone home and said, what's going on here, the nurse said that I'm a boy, and I think it's true?

BJ: Yeah.

Q: Let me ask you a little bit more. So, the first week when you did not get to go back to school, what was going on in your head? Didn't you go back to school the second week?

BJ: The first week I stayed off of school and the second week I went to school.

Q: What was going on in your head that first week? Were you excited, scared, worried, nervous?

BJ: It [the initial upset] was nothing about them [my parents] telling me, it was just all about my friends is all.

Q: Telling your friends.

BJ: Yeah. I was not sad about me being a boy, it was just telling my friends that got me down.

Q: Sad or worried?

BJ: Yeah.

Q: Did you cry a lot? Sometimes? Have you cried very much since that time?

BJ: No.

Q: When you cry is it usually because you have gotten sad?

BJ: Yeah.

Q: How often does that happen? How often do you get sad?

BJ: Most often when I was being a girl. I would think about stupid things. . .
My mom painted my room.

Q: Your room looks great. I forgot to mention it when I came in.

BJ: It was pink. I was tearing off the flowers on it.

Q: Before or after your mom and dad told you [you were born a boy]?

BJ: Before. It was white at the top with a flower trim and then pink at the bottom. I was tearing off the flowers. It was ugly. [Then questions about girlie magazines and whether I have any.]

Q: [Pause] So what has it been like now that you are changing? You did not do much changing when you went from female to male, fast, boom.

BJ: Cut my hair. That's all I did.

This intuitive and often rapid recognition of male identity is striking. It seems to be an internal, personal awareness that somehow is related to a largely male psychosexual development, despite having a female name, genitalia, and social and legal status.

Interdisciplinary collaboration and interventions: roles for mental health professionals

Developing a plan for the child

Given a genetic male child who has cloacal exstrophy and the present knowledge of the etiology of gender identity, how do we assign sex in the neonatal period? First, developing a plan for the child is critical. This plan should include the parents and the family because it is likely to impact the child and family dynamics for many years and should include long-term monitoring.

The plan should be interdisciplinary because of the complexity of cloacal exstrophy. A dedicated child psychiatrist, psychologist, nurse, and social worker can be vital elements of a successful treatment plan for these children and families, for global planning and for specific interventions. With medical insights that are drawn from studies of genetics, embryology, anatomy, physiology, and pharmacology and with clinical training in child development, family therapy, and care of pediatric and surgical patients, a child psychiatrist is an ideal candidate to be the “point player” on the treatment team. In this position s/he would coordinate roles of the team members with patient and parental needs. Some members of the team will be involved for years or decades; others may have intermittent or sporadic involvement as clinically indicated. The child psychiatrist is likely to always be a valuable consultant during child and family growth and development throughout the medical experience.

The team approach should include early team meetings with and without the parents. Short- and long-term goals should be established early and modified whenever necessary. Routine broad and specific developmental assessments, illness education, and discussions and question periods with the patient and the

parents should be accompanied by written summaries for parental reflection. Preventive measures will become more apparent as our understanding of vulnerabilities of the child and family matures.

To assist the parents and the child, I have devised “5-Year Plans” with an additional separate plan for Year 1, that deal with initial survival and intense surgical issues. (General 5-year plans are available from the author.) These plans describe typical clinical and family scenarios during Year 1 and by approximately 5-year blocks thereafter. These materials discuss surgical reconstruction; medical vulnerability; somatic deformities; sexual potential, in terms of function and fertility; incontinence of urine; and social relatedness. Charts, graphics, and written materials enhance comprehension and compliance. Such materials help parents to focus on current problems without losing sight of intermediate and long-term goals of overall treatment and development.

Myths and realities of psychosexual development in children who have cloacal exstrophy

During the initial interactions and interventions with the parents, the child psychiatrist should explore parental understanding of psychosexual development while, over time, discussing the child’s sexual function potential. We do not know the full realities of typical pediatric psychosexual development, especially sexuality in the child who has chronic illness or birth defects. What we know and do not know should be expressed; what we might learn from the children themselves as they grow and develop should be emphasized. The importance of the team approach cannot be understated: these children are medically, surgically, psychosocially, and psychosexually complex.

The few male children who have cloacal exstrophy and were raised as male do well psychosocially and psychosexually. During childhood and adolescence, those who were raised male and those who transitioned to male seem to do better psychosexually than boys who have classic bladder exstrophy, a serious but far less severe or complex anomaly in which boys have a short penis but are reared male. Although causation is not clear, males who have cloacal exstrophy are more likely to masturbate, date, and be sexually active than boys who have classic exstrophy [58,61]. Masturbation commonly occurs after they locate a small, stunted, or subcutaneous perineal locus of erotically sensitive tissue.

The processes of child development and adaptation in genetic males who have cloacal exstrophy can lead to competent psychosocial functioning whether the child is reared male, reared female, or reared female but transitions to male. Data suggest that those who were reared or transitioned to male also have competent psychosexual functioning. Body image is important to these children; genital image is important as well, at least to those who live a male identity. Psychosexual development in those who live as male is complex and encompasses sexual curiosity, imagery, and activity that is not entirely unusual for typical males of the same age. As adolescents, they are more likely than those who live

as female to ask questions that stem from their sexual curiosity, especially about female anatomy and how to please a girl or a woman sexually; they are extremely interested in their own sexuality and sexual function. Such sensitivity may have positive ramifications for peer relations, social relatedness, and even sexual relationships in late adolescence and adulthood.

Conclusion: lessons from the untaught: the cloacal exstrophy experience

Males who were born with cloacal exstrophy, whether raised as male or castrated at birth and raised as female, demonstrate psychosocially and psycho-sexually dominant male-typical tendencies. It seems that prenatal androgen exposure (and perhaps sex chromosome-specific neuronal properties) influences gender role, and, at least sometimes, creates male sexual identity, even if males are castrated at birth and reared female. These children adapt to their lives with severe somatic anomalies, pathophysiologic vulnerabilities, and complex medical and surgical interventions from birth. They do not observe their lives; they live their lives. The intuitive recognition of male identity by many, whether they experienced sex assignment or not, attests to our own lack of knowledge about the formation of human identity.

These children adapt to neonatal surgical castration and feminizing genitoplasty, although data from this study do not imply that clinical indications justify such interventions. Genetic female as well as genetic male children who have cloacal exstrophy require genital construction. Genetic females have normal ovaries; genetic males have normal testes [48]. Surgical castration does not eliminate the need for genital construction; it induces a hypogonadal state that requires life-long exogenous sex hormone administration. Additionally, neonatal castration plus feminizing genitoplasty—coupled with social and legal assignment to female—does not seem to override the neurobiologic underpinnings of male sexual identity and male gender role preferences. Therefore, female sex assignment in genetically- and hormonally-male neonates risks personal internal confusion and social external conflict and contradiction.

Studies of males who have cloacal exstrophy underscore our lack of full understanding of the mechanisms of neonatal and childhood male psychosexual development. Yet we argue that evidence-based medicine must be at the core of medical decision-making. Data from these studies highlights the tenuousness of the intellectual foundations of our arguments for sex assignment in children who have cloacal exstrophy.

The study data imply an important role for prenatal androgen exposure in male-typical development, including male sexual identity. Clinical algorithms and paradigms in these children need to be re-evaluated. The implications of these data need to be studied in terms of children who have intersex. These children's inability to circumvent male-typical gender role and their frequent intuitive recognition of male sexual identity provide arguments that contradict the conception that a genetic and hormonal male can be successfully assigned to female.

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