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Management of infants with ambiguous genitalia reassessed

by Kenneth C. Copeland, M.D., FAAP, Jorge J. Daaboul, M.D., and William Reiner, M.D.

Few pediatric problems elicit more consternation than an infant born with ambiguous genitalia. “Is it a boy or a girl?” is the first question friends and family are likely to ask, and to be unable to respond with certainty creates anxiety in even the most confident physician.

Although uncommon, most pediatricians in clinical practice will encounter an infant with ambiguous genitalia several times during their career, and most busy pediatric endocrinologists will encounter several such affected infants each year. For several generations, pediatric endocrinologists and pediatric urologists in this country have been trained that the management of “intersex” followed a reasonably well-defined algorithm. Although passage through the decision-making tree usually required a battery of tests and procedures, and although this process took some time, ultimately the “proper” gender was discovered. Parents then could be told “what sex your child really is.”

Compassionate specialists sincerely thought they knew how to determine what was best for the patient and family. Traditionally, the presence of a uterus, tubes and ovaries defined fertility potential; thus, female gender was assigned. Similarly, the absence of a penis “adequate” for intercourse presumably doomed the infant to a life as an inadequate man, and usually a female gender was assigned. In an otherwise normal-appearing female infant with a large clitoris, genital surgery in infancy was felt to be beneficial so the visual appearance did not confuse parents, grandparents and friends, and somehow interfere with proper socialization and gender identification of the female infant.

Over the last five or six years, specialists have been rethinking these approaches to such infants. Several national and international meetings have been convened over the last four years to deal with the topic, combining representation from both medical and lay communities. Evident from this interdisciplinary discussion is the reality that there exists a dearth of long-term data on the ultimate outcome of children born with anomalies of the external genitalia. Furthermore, it has become evident that some of the presumptions on which medical decisions were made, such as “fluidity” of gender identity if assigned in early infancy, were based on faulty assumptions.

Recently, the pediatric endocrine and urology communities have come to recognize that traditional approaches need to be modified somewhat. These discussions can be summarized as follows:

Is ambiguous genitalia a ‘social emergency’? Physicians usually feel a sense of urgency to assign gender to a newborn. This may be fueled in part by a belief that parents cannot tolerate uncertainty in their infant’s sex assignment. There simply is no evidence to support this claim. Most parents are able to assimilate complex medical information from other areas of newborn care and, given appropriate education and support, do grasp intellectual and emotional ambiguities.

Ambiguous genitalia generally does not involve the urgency associated with birth at 28 weeks of gestation nor does it carry the weight of life and death decisions. Pediatricians should assume that parents of a newborn infant with ambiguous genitalia are capable of accepting a delay in gender assignment and participating in an open discussion of available options.

Who should decide the baby’s gender? We presume that parents best represent the interests of their children. Physicians should provide information on risks, benefits and alternatives to available treatment; assure adequate parental understanding of the situation; make appropriate recommendations; and help parents to choose what best fits their circumstances. Parents must be permitted to disagree with the gender assignment, even when physicians believe recommendations are based on “subjective” facts and experience. Excluding families from full participation in gender assignment is illogical and unethical, especially in light of the paucity of data on the long-term outcomes of children assigned a sex by their treating physicians.

Under moral and legal precepts of the doctrine of informed consent, health care professionals have a clear obligation to disclose all relevant information about diagnosis, treatment options and expected outcomes. By contemporary standards, good medical decision-making is decision-making shared with parents and/or valid surrogates.

What are the critical factors to be considered in the process of gender assignment in infancy? What matters most is the patient’s gender identity and quality of life. Ultimate personal gender identity “trumps” everything else: fertility potential, chromosomal sex, what gender baby the parents wanted. If a female with ovaries, tubes and uterus chooses never to have heterosexual intercourse or become pregnant or an XY male considers himself exclusively male regardless of the absence of a penis and the presence of a perfectly constructed vagina, then “potential” function becomes irrelevant.

Quality-of-life outcome can be defined only by the patient. Unfortunately, since genital sexual function primarily is adolescent and adult in timing, patients are likely to judge their quality of life (with regard to genital function) as most important during these times. Adults, adolescents and even young children are capable of recognizing sexual identity, and this sexual identity may be discordant with assigned sex, sex-of-rearing, genetic sex or predicted sexual identity. Thus, surgical interventions in the neonatal and infancy periods that preclude desired genital reconstruction later are to be discouraged.

Clearly, some surgical interventions must be directed by clinically significant realities (malignancy risk, etc.) independent of age.

By recognizing these issues, physicians can work to help parents caught in this dilemma.

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Section working to spark interest, funding in urology research

by Barry A. Kogan, M.D., FAAP

The AAP Section on Urology realizes the future care of our patients depends greatly on research advances. It is because of this that we are working on many fronts to increase interest and funding in pediatric urology research.

One positive step has been the follow-up of an National Institutes of Health (NIH) sponsored workshop on congenital hydronephrosis. This has resulted in a program announcement (#PA-03-076), requesting both clinical and basic science grants related to congenital obstructive uropathy. The announcement was released through Feb. 1, 2006, and is a great opportunity for anyone interested in this area. Proposals are encouraged for both traditional (R01) grants and for exploratory development project (R21) grants. The latter require very little preliminary data. Details are available at http://grants.nih.gov/grants/guide/pa-files/PA-03-076.html.

Progress also has been made toward a multicenter clinical trial in pediatric urology funded by the NIH. Most likely the project will be in the field of urinary tract infections and vesicoureteral reflux. Several proposals are in the early stages of being considered. Almost certainly, the final result will be multidisciplinary with input from general pediatricians, pediatric nephrologists and pediatric emergency physicians in addition to pediatric urologists. How rapidly this project moves forward will depend on the constraints of this year’s budget, but the National Institute of Diabetes & Digestive & Kidney Diseases has recognized a need in this area.

Two other concepts remain in the early planning stages. We all feel it is important to perform a formal research progress report in pediatric urology. This report would document strengths, weaknesses and opportunities in pediatric urology research so the NIH could target the most appropriate areas in the future. Progress reports in diabetes and bladder dysfunction have led to increased recognition and funding for research in these areas. In addition, there is a recognized need for higher quality clinical research in pediatric urology. We are hopeful the NIH will host a workshop to highlight skills in this area. A previous workshop in nephrology was highly successful.

Overall, we are making slow but steady progress in this area, but more effort is needed. All of us in pediatrics realize that children do not vote, and hence, it is particularly important for us to advocate for research on their behalf.

Dr. Kogan is chair-elect of the AAP Section on Urology executive committee.
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