Clinical Reports and Narratives

Genital Ambiguity at Birth: Ethical Issues in the Management of Children with Differences of Sexual Development, also Known as Intersex

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ABSTRACT

The "best interest of the child" is the primary principle in medical decision making for infants and minor children. In infants born with disorders of sexual development (DSD), early genitoplasty (plastic surgery to the genitals) in the absence of medical or surgical indication is not in the best interest of the child. Infants with DSD have the right to an open future, which can only be supported if they can participate meaningfully in decision making. In this clinical report, we present the case of a newborn with DSD and use three basic principles of bioethics to support our recommendations against nontherapeutic early genitoplasty.

CLINICAL PRESENTATION

Baby A was born at full term and admitted to the neonatal intensive care unit (NICU), as planned, due to a prenatal diagnosis. On physical examination the baby was found to have ambiguous genitalia.

CASE DISCUSSION AND BACKGROUND

Baby A was prenatally diagnosed with mosaic Turner syndrome, with the majority of cells with Y (male) chromosome after amniocentesis. Mosaic Turner syndrome is a genetic condition in which the karyotype is 45 XO/46XY.[1] On examination, there was a 3-cm-long clitoro-phallic structure (like an underdeveloped penis) and a urethral opening at the tip with mild hypospadias (an opening at the base of the clitoro-phallic structure) that suggested virilized genitalia (that is, genitalia influenced by the male hormone testosterone). A separate vaginal opening was not visualized. No gonads were palpable on examination.

Genetic workup in the NICU confirmed the prenatal diagnosis. The infant had normal electrolytes and normal levels of cortisol, testosterone, follicle-stimulating hormone, luteinizing hormone, and estrogen. On pelvic ultrasound, the uterus could be seen, but gonads were not visible, which suggested gonadal dysgenesis (atypical development of the gonads in which reproductive tissue is replaced by functionless, fibrous tissue), which would require surgical removal due to cancer risk. The medical team planned to do magnetic resonance imaging (MRI) when the child turned two, to check for the presence of gonads.

The medical team recommended not assigning any sex to the Baby A. They suggested it would be best for the baby to forgo a non-urgent surgical intervention such as genitoplasty and be raised in a gender-neutral manner until the child could declare a preferred gender.

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The parents were overwhelmed by the ambiguous genitalia of their newborn. They belonged to a very tightly knit religious community. A commitment to a gender was considered essential in their family and community. They were worried about being unaccepted by and alienated from their community if they decided to raise a child who had ambiguous genitalia. Most importantly, they were fearful and concerned about stigma and bullying for their child within their community if the child was reared without a definitive gender.

Additionally, early genitoplasty in children with ambiguous genitalia cannot be considered to be in their best interests in the absence of sufficient data that support the benefits of early genitoplasty.

Baby A was their third child after two female children. The parents wanted the medical team to complete their workup, decide on a sex assignment for the baby, and then pursue surgical intervention for genital modification to conform to the chosen gender. The parents did not want to take their baby home with ambiguity about the baby's gender.

The medical team requested an ethics consultation to help in deciding the best possible course of action. After discussing the ethical dilemma with the medical team, the ethics team set up a care conference with the family and the medical teams (endocrine, genetics, neonatology) and the allied healthcare teams (social work, spiritual care, psychology, and ethics).

During the care conference, a review of the baby's medical condition and genetic, endocrine, and anatomic information was presented to the family. The infant's blood karyotype was reviewed, which showed the majority of cells with 45X (female chromosome, 86.5 to 87.5 percent), and additional cell lines with 46XY (3 to 7 percent) and 47XYY (6.5 to 9.5 percent). The family understood that the karyotype was different from the amniocentesis. The results of an extensive endocrine workup for DSD (differences of sexual development) remained pending. The medical team expressed uncertainty in making decisions regarding the baby's sex solely based on karyotype.

CHART NOTE AND RECOMMENDATIONS

There was uncertainty regarding predicting the appropriate gender for the child. The ethics team recommended the following:

1. In the absence of a medical or surgical indication for urgent intervention, the assignment of sex for the child should be delayed until the child developed the capacity to participate in decision making and could express an opinion regarding gender, and whether the child wished to pursue a surgical modification.
2. The medical team was under no obligation to offer medically unnecessary surgical procedures. In fact, when the procedures would be harmful, as in this case, the team had an obligation to protect the child's bodily integrity.

REASONING

The ethics team's recommendation was built upon the principles of respect for the patient's right to self-determination and the right to participate in decision making for medical treatment. Additionally, these recommendations were based on the patient's right to bodily integrity and quality of life, and the right not to be harmed by unnecessary interventions. These principles are further discussed below.

Best Interests and Nonmaleficence

The concept of best interests in the management of a child with DSD focuses on the child's psychosocial well-being and the effectiveness of the treatment.[2] In the prepubertal phase, gender identity and gender role development do not correlate with the appearance of external genitalia.[3] Therefore, a child's well-being cannot be confirmed automatically by the determination of unambiguous external genitalia.

Additionally, early genitoplasty in children with ambiguous genitalia cannot be considered to be in their best interests in the absence of sufficient data that support the benefits of early genitoplasty. The potential harm of impaired urological and sexual function, dissatisfaction with the appearance of the genitalia, and repeated surgeries outweigh the reported benefits of improved wound healing, decreased anxiety, and technical ease of surgery in early genitoplasty.[4] There is increasing evidence that sex hormones influence the developing brain prenatally.[5] Undergoing gender assignment surgery in infancy could result in discordance between the appearance of a person's genitalia and a person's affirmed gender later in life, which can cause irreparable psychological damage. Also, any inherent medical risks for any surgery, including the potential detrimental impact of anesthesia on the developing brain, cannot be ruled out.[6]

As in this case, an infant child's wishes cannot be determined with certainty. No one should make decisions regarding permanent genital sex assignment surgery except for the patient. Hence, minor interventions for genital sex assignment should be deferred at least until the age of five or six years, that is, around the time when a child can express a gender preference; major interventions should be deferred until the age of 12 to 14 years.[7]

Autonomy and Informed Consent

The concept of autonomy in pediatrics is unique because the patient is not usually autonomous. In the case of early genitoplasty, the child is too young to provide assent, so the parents act as surrogate decision makers. Even so, these decisions must comply with the best interests' standard, and remain above the threshold of harm. It is inappropriate for parents to determine their child's gender identity by advocating for irreversible genital gender assignment surgery when the child cannot express or articulate a gender. Such a decision would be a violation of the child's autonomy.[8] The child should have an "open future,"[9] that is, genitoplasty should be delayed until the child reaches the capacity to decide on a gender after weighing the potential risks and benefits.

In newborns, after excluding conditions that require an urgent intervention, such as functional disorders of the urinary tract or recurrent urinary tract infection, ambiguous genitalia do not represent a surgical emergency.[10] Any therapeutic decision that is not intended to prevent imminent harm to a child's health and well-being must be carefully weighed for benefits and risks. Due consideration should be given to the different possible options and should be reviewed thoroughly by multidisciplinary healthcare team members and parents.

RESOLUTION OF THE CASE

After extensive discussion, Baby A's parents decided to raise the baby as a male and to delay all permanent interventions for genital sex assignment for the time being. The parental decision to raise their baby as a male was based on the appearance of the baby's external genitalia and the parents' understanding of the amniocentesis results. The parents mentioned that they would let the child decide the right gender when older, and, as parents, they would be comfortable if the child chose to be a female in the future.

To address their fear of how their community and the family would receive their child, the parents had the opportunity to meet with the social work team, the spiritual care team, and the psychology team. They received counseling on the potential effects of stigma and strategies to cope with it. They were advised to connect with parent-to-parent self-help groups as sources of information and social support.

BLINDING OF THE CASE

Details of this case have been altered to protect the privacy of the patient and the patient's family.

NOTES

1. M. Rasouli, K. McDaniel, M. Awadalla, and K. Chung, "Mosaic turner syndrome presenting with a 46, XY karyotype," *Case Reports in Obstetrics and Gynecology* (11 April 2019): 1-3.

2. A.A. Kon, "Ethical issues in decision-making for infants with disorders of sexual development," *Hormone and Metabolic Research* 47, no. 5 (2015): 340-3.

3. U. Thyen, H. Richter-Appelt, C. Wiesemann, P.-M. Holterhus, and O. Hiort, "Deciding on gender in children with intersex conditions," *Treatments in Endocrinology* 4, no. 1 (2005): 1-8.

4. R.M. Harris and Y.-M. Chan, "Ethical issues with early genitoplasty in children with disorders of sex development," *Current Opinion in Endocrinology, Diabetes and Obesity* 26, no. 1 (2019): 49-53; I.A. Hughes et al., "Consensus statement on management of intersex disorders," *Journal of Pediatric Urology* 2, no. 3 (2006): 148-62.

5. S. Rowlands and J.-J. Amy, "Preserving the reproductive potential of transgender and intersex people," *European Journal of Contraception & Reproductive Health Care* 23, no. 1 (2018): 58-63.

6. Harris and Chan, "Ethical issues with early genitoplasty," see note 4 above.

7. C. Wiesemann, S. Ude-Koeller, G.H.G. Sinnecker, and U. Thyen, "Ethical principles and recommendations for the medical management of differences of sex development (DSD)/intersex in children and adolescents," *European Journal of Pediatrics* 169, no. 6 (2010): 671-9.

8. D.B. Gorduza, C.A. Quigley, A.A. Caldamone, and P.D.E. Mouriquand, "Surgery of anomalies of gonadal and genital development in the 'post-truth era,' " *Urologic Clinics* 45, no. 4 (2018): 659-69; J. Garland and S. Slokenberga, "Protecting the rights of children with intersex conditions from nonconsensual gender-conforming medical interventions: The view from Europe," *Medical Law Review* 27, no. 3 (2019): 482-508.

9. Harris and Chan, "Ethical issues with early genitoplasty," see note 4 above.

10. A.F. Saltzman et al., "Patients with disorders of sex development and proximal hypospadias are at high risk for reoperation," *World Journal of Urology* 36, no. 12 (2018): 2051-8.