Ethical aspects of gender assignment in ambiguous genitalia-congenital adrenal hyperplasia A case report

by Nur Rochmah

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Case Report

Ethical aspects of gender assignment in ambiguous genitalia-congenital adrenal hyperplasia: a case report

Nur Rochmah[1,2], Muhammad Faizi[1,2], Adwina Nurlita Kusuma Wardhani[2]

Congenital adrenal hyperplasia (CAH) is an autosomal recessive disorder commonly caused by mutation of the CYP21A2 gene, resulting in deficiency of an enzyme required for cortisol synthesis in the adrenal cortex. In 90-95% of cases, the deficient enzyme is 21-hydroxylase (21-OH), with an incidence ranging from 1 in 5,000 to 15,000 live births across various ethnic and racial backgrounds. In classical 21-OH deficiency (21-OHD) CAH, excessive androgen exposure in the fetus results in virilization at birth.[1]

The management of ambiguous genitalia in children with CAH presents a unique and ethically challenging decision-making dilemma for the medical team. Insensitive and poorly informed statements made in the delivery room may cause long-term psychological problems for the families. It is important to refrain from assigning gender until sufficient diagnostic information can be gathered. Parents, as guardians, and the supporting medical team must make decisions on behalf of the child, with the goal of enabling the child to grow into a healthy and happy adult with his or her assigned gender.[2,3] We report a case of a child with CAH, focusing on the ethical challenges in management of ambiguous genitalia. **[Paediatr Indones. 2021;61:356-8 ; DOI: 10.14238/pi61.6.2021.356-8].**

Keywords: *ethical approach; congenital adrenal hyperplasia; child; ambiguous genitalia; gender identity*

The Case

A 14-month-old girl who had been diagnosed with salt-wasting CAH since the age of three months was brought to Dr. Soetomo General Hospital, Surabaya, Indonesia, with complaints of vomiting at every feeding, about four times per day, for the previous three days. The child's body weight was 7.4 kg, body length was 66 cm, and head circumference was 37 cm. Her routine medications were hydrocortisone, fludrocortisone, and salt supplementation.

On physical examination, her vital signs were normal. She had craniosynostosis and ambiguous external genitalia with enlarged clitoris, partly fused and rugated labia majora, and no palpable gonads (**Figure 1**).

Laboratory examination revealed a 17-hydroxyprogesterone (17-OHP) level of 73.00 ng/mL (reference value 0.11-1.70 ng/mL) and a karyotype of 46XX, supporting the diagnosis of CAH. Thyroid function tests showed a free thyroxine (FT4) level of 2.19 ng/dL (reference value 0.9-2.3 ng/dL) and a thyroid-stimulating hormone (TSH) level of 14.68 uIU/mL (reference value 1.7-9.1 uIU/mL), consistent with compensated hypothyroidism. As shown in **Figure 2**, her bone age was appropriate for age, based on Greulich and Pyle.[4] The patient was diagnosed with 46XX classical salt-wasting CAH, with microcephaly, craniosynostosis, and severe stunting.

Department of Medical Science[1] and Department of Child Health[2], Universitas Airlangga Medical School, Surabaya, East Java, Indonesia.

Corresponding author: Nur Rochmah, Department of Child Health, Universitas Airlangga Medical School/Dr. Soetomo General Hospital. Jalan Mayjend Prof. Dr. Moestopo No. 6-8, Surabaya 60286, East Java, Indonesia. Phone: +6281703501118; Email: drnurrochmah@gmail.com.

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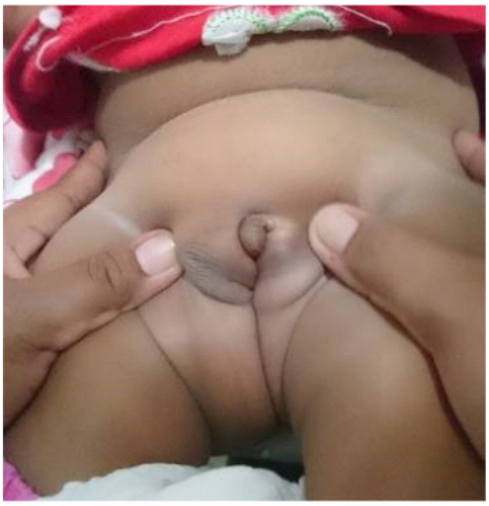

Figure 1. CAH female patient with ambiguous genitalia

Discussion

Congenital adrenal hyperplasia (CAH) is the most common cause of genital ambiguity due to 21-OHD in cortisol synthesis. Females with classic 21-OHD have ambiguous genitalia at birth: clitoromegaly, fused and rugated labia majora, and a common urogenital sinus consisting of the urethra and the vagina,1 as was present in this patient.

Prenatal diagnosis should be performed in pregnant women who have previously given birth to a child with CAH. In such cases, the likelihood of bearing an affected girl is 1/8. Masculinization of the external genitalia begins at 8 weeks' gestation. Therefore, treatment must be started immediately after pregnancy is confirmed. Steroid treatment is commonly started at 6 weeks' gestation, using dexamethasone at a dose of 20 mg/kg/day, divided into three doses. Approximately 70% of prenatally treated females are born with normal genitalia or only a few male genitalia characteristics. Ethical gender assignment requires a multidisciplinary team.[5] Prenatal preventive treatment remains controversial because dexamethasone consumption in early pregnancy influences fetal neural development.[1,5] In countries with limited resources, such as Indonesia, prenatal screening tests for CAH are not routinely done.[6]

The healthcare team and parents of newborns with ambiguous genitalia face an enormous challenge

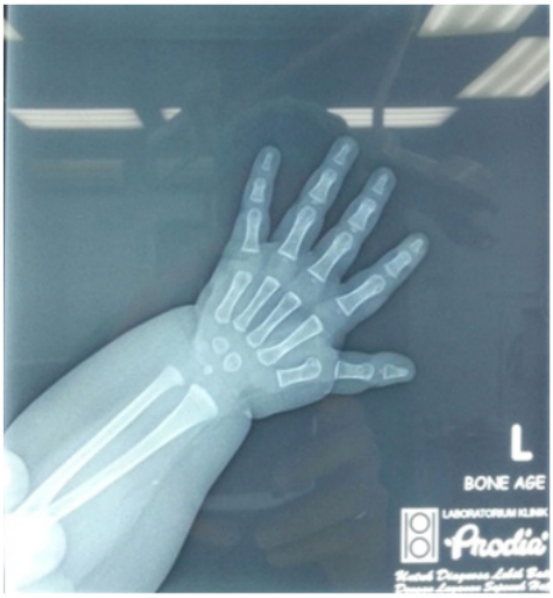

Figure 2. An x-ray revealed a bone age appropriate for a 14-month-old girl

against local cultural stigma. Early conversations need to use positive terms like "your baby" rather than "it," to promote greater bonding and humanize the baby. Both parents and professionals need to use gender-neutral terms until the decision is finalized, to prevent misunderstanding and confusion.[2] The medical

provider should emphasize the baby's potential to grow and develop into a healthy and happy adult if given appropriate treatment. In Indonesian society, parents may feel ashamed of having a child with ambiguous genitalia. Parents who seek help must be supported.[6]

The initial management of an infant diagnosed with ambiguous genitalia begins with gender assignment. There is agreement that patients should be assigned a gender as soon as clinical data are available. Factors influencing gender assignment include diagnosis, fertility potential, genitalia appearance, adult sexual function, surgical and hormone replacement options, family preferences, and cultural factors. Reduction of excess sex hormones and replacement of adrenal hormone deficiency remains the goal of CAH treatment. Administration of glucocorticoids and mineralocorticoids were given. For severely virilized females, genital surgery such as clitoroplasty and vaginoplasty may be necessary to produce successful development of gender which is programmed during the first few years of life through social conditioning.[2,3] A multidisciplinary team is important in the management of such cases and should be involved in discussions on sexuality, fertility, menstruation, and complementary surgery, as well as to provide information and options to families. The team should consist of pediatric endocrinologist, pediatric growth and development, gynecologist, geneticist, clinical psychologist, psychiatrist and pediatric surgeons/urologists.[3,7] After explanation of physical examination, laboratory results, multidisciplinary team allow parents to make gender assignment, in this case, parents choose gender assignment as a female.

The medical and surgical management of children with disorders of gender development (DSD) require several major decisions on behalf of child. Parents as guardians have rights and responsibilities. Parental preference is the main factor in the ethics-based decision-making process to guide treatment for their child's fertility and sexual relations in the future, while minimizing health and psychosocial risks as mentioned in UU No. 35 2014 about Child Protection, MUI Fatwa No. 03/Munas/VIII/2010 about Gender Assignment and Refinement, also Minister of Health Constitution No. 191/MENKES/SK/III/1989 about the implementation of hospital and expert team as the place of gender assignment surgery. The Halifax Resolution outlines the principles guiding decisions regarding the type and timing of surgery for children with DSD, including minimizing physical and psychological risks, preserving the capacity of satisfying sexual relations, providing open options for the future, and respecting parents' wishes and beliefs.[2]

Ethics-based decision-making in the management of infants with ambiguous genitalia is challenging in terms of complexity, stigma, and implications on the child's future. Advancements in diagnostic technology and medicine cannot substitute for ethics-based decision-making. The multidisciplinary team should use ethics-based guidelines in communicating and educating parents, stabilizing mental health, developing treatment plans, and setting overall priorities to improve the well-being of individuals with DSD over their lifetime.[3]

We conclude that ethics-based guidelines for treatment plans by multidisciplinary team are the most important factor for improving the well-being of CAH patients with ambiguous genital over their lifetime.

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