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When sex development goes awry: Is it a girl or a boy?

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Few conditions underscore the importance of bedside manner quite like differences and disorders of sex development (DSD). Doctors faced with DSD patients must collaborate to recommend tailored and timely management plans covering medical, psychological, and social cofactors of the most intimate nature. Standards of care are ever evolving, but now front-line pediatricians from Case Western Reserve University School of Medicine have assembled the latest data about the varied causes of DSD, complete with clinical vignettes and appropriate management plans. Their comprehensive review was included in a special issue of *Birth Defects Research Part C: Embryo Today*.

The review's lead author is Naveen Uli, MD, associate professor of pediatrics at Case Western Reserve University School of Medicine. "Our aim was to provide an overview of the complexity of the medical decisions, investigations, and treatment approaches involved in the diagnosis and management of this spectrum of disorders," said Uli. "We wanted to highlight a variety of disorders, as well as presentation during the newborn period, and later."

In the review, researchers outline the complex process of sex development that typically leaves a baby girl with two X chromosomes, and a baby boy with an X and a Y. Sex chromosomes are a major driver of gender-specific genitalia, hormones, and developmental timelines. But as Uli explains, an abnormal number of sex chromosomes can have consequences that are not immediately apparent.

"DSD comprise a wide range of molecular etiologies, with great variation in presentation," said Uli. "Diagnosis in individuals may occur any time from the newborn period to adulthood."

Management plans for DSD can require very stressful, life-altering decisions, including gender assignment. In one case study in the review, a baby born with ambiguous genitalia completes many rounds of testing before a DSD is identified. Although the baby leaves the hospital after birth without a gender, doctors work with the family to follow up with hormone therapy and reparative surgery before he reaches one year old. An included discussion offers support for medical professionals faced with similar situations. The authors provide strategies to balance proper psychological development, clinical findings, and family concerns—all within in a tight timeline.

Said Uli, "These conditions have wide ranging implications on health, behavior, psychosocial function, family dynamics, and society as a whole. Appropriate recognition is key to optimizing evaluation, intervention, and outcomes."

Diagnostic tools have evolved substantially for DSD over the past decade, lowering costs and testing turn-around times. According to Uli, as molecular diagnostic technology advances, the science underlying DSD will become more precise which will inform treatment plans. Until then, the review article offers a comprehensive resource for doctors navigating these changes to treat the spectrum of individuals living with DSD.

Provided by Case Western Reserve University