

# Seven studies address scientific, cultural considerations for kids born with ambiguous genitalia, mismatched chromosomes

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Disorders and differences of sex development, or DSD, has replaced "intersex" to describe a spectrum of conditions related to defects of human sex development. The new terminology follows an explosion of research investigating challenges related to DSD, including varying physical presentations and repercussions. Now, in a special journal issue, 36 researchers offer a comprehensive look at DSD from its molecular etiology to its effect on society as a whole.

Naveen Uli, MD, associate professor of pediatrics and Michiko Watanabe, PhD, professor of pediatrics at Case Western Reserve University School of Medicine served as editors for the special issue of *Birth Defects Research Part C: Embryo Today*.

The issue provides a much-needed scientific overview of current sex development understanding, and guidance for those treating individuals with DSD. According to the introduction, DSD care teams can include "endocrinologists, obstetrician-gynecologists, psychiatrists, psychologists, surgeons, urologists, sociologists, as well as the primary care pediatrician and family members." Updates relevant to all care team members are provided across seven review articles.

"I believe every article in this issue sheds light on a unique aspect of DSD," said Uli. "The reviews highlight the complexities in the basic biologic process of sex development, and provide examples of situations

in which the answer to 'is it a boy or a girl?' is not so straightforward."

According to the special issue, 1 in every 4500 to 5000 individuals is born with a DSD. Children with DSD may be born with ambiguous genitalia, or have sex chromosomes that do not match internal or external genitalia. The condition may be diagnosed pre- or post-natally. Diagnoses are accompanied by myriad questions, including clinical and cultural concerns. A DSD diagnosis also triggers a cascade of tough decisions, which may involve gender assignment or surgical interventions. In the special issue, leading researchers bring readers up to date on key aspects of DSD including:

- Complex steps in sex development
- Common genetic causes of DSD
- "Epigenetic" molecular tags on DNA that influence sex development
- Sociocultural implications of DSD worldwide
- Sex-specific effects of chemical exposure on developing fetuses
- Challenges in diagnosing and managing DSD
- Advances in DSD molecular diagnostics

Together, the articles offer a comprehensive perspective on DSD "through the distinct lenses of authors from all over the world with their different scientific, cultural and societal backgrounds," write Uli and Watanabe.

Said Uli, "My hope is that the broad scope of articles in this special issue will provide a frame of reference to the reader, which should serve as a good starting point for further exploration."

Provided by Case Western Reserve University

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