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Series of New Studies Shed Light on Sex Determination in Human Development

Teratology Society Journal Issue Advances Physical and Cultural Understanding of Sex Development Disorders

RESTON, VIRGINIA— “‘Is it a girl or a boy?’ For parents of children with Disorders/Differences in Sex Development (DSD), the question can provoke great anxiety,” writes Michiko Watanabe, PhD, and Naveen Uli, MD, co-editors of a special issue of [Birth Defects Research Part C: Embryo Today](#), which examines challenges surrounding sex determination of babies born with defects of the genitalia.

Historically referred to as “intersex,” the Differences/Disorders of Sex Development (DSD) means babies born with DSD can have sex chromosomes that don’t match internal or external genital anatomy that are expected for a boy or a girl or there are ambiguous genitalia. The scientific journal released today (see the [Overview](#), doi: 10.1002/bdrc.21149) includes studies shedding light on how changed molecular structure of DNA could predict certain DSD in the baby.

“I was struck by the [review by Mendonca et al.](#) in which they followed many patients and started with a thorough and detailed phenotyping of patient anatomy. They then delved deeply into the details of the associated 3-D molecular structures that caused the changes in anatomy,” said Dr. Watanabe, who is also a professor of pediatrics at Case Western Reserve University. “This highly detailed explanation of how DSD arose could have big implications in devising potential treatments for the future,” she added.

Dr. Watanabe points out that while the research included in the journal surrounding DSD focuses primarily on physical birth defects of the genitalia, she does see it as a gateway for potential answers to questions surrounding the transgender community. “The issue of transgender is certainly getting a lot of heated public discussion,” she said. “This is a brain function issue since the brain is an important sex organ. It is very likely linked to molecular changes during brain development and differentiation. We are just beginning to understand this complex and fascinating story.”

The heated social implications toward DSD are also alive and well in the communities around the world, as noted by Anastasia Ediati, PhD, lead author of another [review](#) included in the issue who also serves as a faculty member of the Clinical Psychology Department at Diponegoro University, Semarang, Indonesia. “Having a genetic diagnosis can be important for patients with a DSD since it is often met with stigma.” She points out the strong social and cultural reactions to DSD and how the families of individuals with DSD may be influenced by the social and cultural environment. “They’re making important decisions for medical treatment that may not necessarily be in the best interest of the individual with DSD due to these influences.” However, Dr. Ediati also points out that many efforts had been taken to promote acceptance from the families of those affected by DSD, but less efforts have

been done to work on the community level. “Social support environments for individuals with DSD may be a good place to start, but we still have a long way to go.”

About the Teratology Society

[The Teratology Society](#), an international professional group of scientists hailed as the premier source for cutting-edge research and authoritative information related to birth defects and developmentally-mediated disorders, publishes *Birth Defects Research* with John Wiley & Sons.

The Teratology Society is made up of more than 700 members worldwide specializing in a variety of disciplines related to birth defects research, including developmental biology and toxicology, reproduction and endocrinology, epidemiology, cell and molecular biology, nutritional biochemistry, and genetics as well as the clinical disciplines of prenatal medicine, pediatrics, obstetrics, neonatology, medical genetics, and teratogen risk counseling. Scientists interested in membership in the Teratology Society are encouraged to visit www.teratology.org.

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