

Disorders of Sex Development (DSD)

The DSD Center at Cincinnati Children's Hospital Medical Center (CCHMC) uses an interdisciplinary approach with compassionate care to meet the complex needs of patients diagnosed with disorders of sex development (DSD).

The term DSD refers to congenital conditions affecting the normal development of the sex chromosomes, sex glands and hormones and genital appearance of a person. DSD typically occur early in pregnancy when the development of reproductive structures has been interrupted or altered. While some DSD are diagnosed at birth, others may be found later, during childhood, puberty, or even adulthood. Many types of DSD are not life-threatening, but some can have associated health issues. Treatment of DSD can be complex, and usually involves the combined expertise of doctors from several different specialties.

Interdisciplinary Team

The DSD team at CCHMC includes physicians from five specialty divisions including: Endocrinology, Gynecology, Human Genetics, Psychology/Behavioral Medicine, and Urology. Combined with nurses, social workers and genetic counselors, our team works in collaboration to provide patients/families:

- Initial diagnosis and assessment
- Comprehensive, interdisciplinary care, with close communication among the specialists in making treatment plans
- Personalized medical, psychosocial and/ or surgical treatment, and support services
- Coordination of care (referrals, appointments, tests, treatment) in a single, convenient setting
- Education and empowerment of patients and families to receive the best holistic care possible with optimal short and long-term outcomes

Conditions Treated

The DSD Center provides care for all ages, from counseling before birth to treatment through adulthood. The goal of the team is to meet the needs of our patients and families as partners of the healthcare team. The DSD conditions treated at CCHMC include, but are not limited to:

Disorders of sex chromosomes

- Mixed Gonadal Dysgenesis
- Sex chromosome mosaicism
- Disorders resulting from abnormal number or appearance of X and Y sex chromosomes, including Klinefelter syndrome

Disorders of sex glands and sex hormones

- Congenital adrenal hyperplasia (CAH)
- Gonadal dysgenesis (complete or partial)
- Ovotesticular DSD
- Testosterone synthesis defects
- Androgen insensitivity (complete or partial, CAIS or PAIS)

Disorders of internal and external genitalia

- Severe hypospadias (penoscrotal hypospadias)
- Cloacal anomalies/ cloacal exstrophy
- Absent vagina/ Mayer-Rokitansky-Kuster-Hauser syndrome (MRKH)
- Mullerian duct abnormalities

Our mission is to provide excellent, innovative care improving the medical and quality-of-life outcomes of patients and families living with complex DSD conditions.



Referrals

Health care providers may refer a patient by calling the DSD Center at CCHMC at **513-803-4DSD (4373) or 1-800-344-2462, ext 4373.**

Physician Priority Link (PPL) a physician to physician hotline for referring physicians is available 24 hours a day, seven days a week. PPL operators can put physicians in contact with appropriate on-call specialty services (endocrinology, genetics, urology) for urgent referrals. **Call 636-7997 or 1-888-636-7997.**

Parents or patients may self-refer or make an appointment by contacting the DSD Center at CCHMC at: **513-803-4373 (4DSD) or 1-800-344-2462, ext. 4373.**