

INTRODUCTION

Variations/Differences of sex development/characteristics (formerly called DSD Disorders of Sex Development or Ambiguous Genitalia) is a term to encompass a broad range of conditions in which child's genital, gonadal or chromosomal presentation is not easily categorized as "typically female" or "typically male". For example, congenital adrenal hyperplasia can lead to virilization of an XX infant, whereas androgen insensitivity syndrome may lead to undervirilization of an XY infant, yet both may have the same external genital appearance.

Variations in sex characteristics can present prenatally with a mismatch between karyotype and fetal ultrasound, at birth or infancy (where infants are identified, diagnosed and managed in the immediate newborn period), or later in life. Variations in sex characteristics can appear as a lack of breast development, primary amenorrhea, unexpected virilization, lack of axillary/pubertal hair, micropenis, or significant hypospadias.

Patients presenting with variations in sex characteristics are managed by our GUPPE team – a multi-disciplinary team that includes Genetics, Urology, Psychology, Psychiatry and Endocrinology. Our clinicians are experienced in variations in sex characteristics and closely collaborate to develop the best possible treatment plan for each patient and family by providing comprehensive medical and psychological support.

INITIAL EVALUATION AND MANAGEMENT

INITIAL EVALUATION:

- Obtain targeted history:
 - Include neonatal exposure to any medications including topical hormone creams, maternal history of any virilization during pregnancy, birth history, any family history of genital concerns, pubertal disorders, infertility or sudden death
- Perform targeted physical exam:
 - Include genital exam, noting the following:
 - Palpate for gonads: note location (e.g., inguinal canals/labioscrotal area) or that they are non-palpable
 - Note the size of the clitorophallic structure
 - Assess if the urethral opening can be visualized, and where it is located (e.g., visualized or not visualized at tip/base/mid length or the clitorophallic structure)
 - Check if labioscrotal folds are fused
 - If karyotype is already known, provider can describe findings associated with the chromosomal sex (i.e., labial folds, vaginal opening, clitoris, scrotum, penis)

INITIAL MANAGEMENT:

- If the evaluation is suggestive of a variation in sex characteristics, please provide family with education and reassurance.
 - Refer families to [Clinic for Variations of Sex Development | Connecticut Children's](#): Clinic page with additional resources
- If patient has any variations of sex characteristics found on physical exam AND failure to thrive and/or poor feeding, please proceed with emergent referral to Endocrinology (see **When to Refer**).
 - Do not delay referral to Endocrinology by obtaining electrolytes

WHEN TO REFER	<p>EMERGENT REFERRAL to Endocrinology:</p> <ul style="list-style-type: none"> • If patient has any variations of sex characteristics found on physical exam AND failure to thrive and/or poor feeding, call Connecticut Children's One-Call (Phone: 833.733.7669) and ask to be connected to the Endocrinologist on call • If family needs to speak with the GUPPE family support clinician, reach out to the GUPPE clinic directly at 860-545-9395, who will then contact the family <p>ROUTINE REFERRAL to GUPPE Clinic (within 1 month):</p> <ul style="list-style-type: none"> ▪ Please refer to GUPPE Clinic as soon as a variation of sex characteristics is suspected. Early diagnosis and management leads to better medical and psychological outcomes for these families.
HOW TO REFER	<p>Referral to GUPPE Clinic in Department of Urology via CT Children's One Call Access Center Phone: 833.733.7669 Fax: 833.226.2329 For more information on how to place referrals to Connecticut Children's, click here.</p> <p><i>Information to be included with the referral:</i></p> <ul style="list-style-type: none"> ▪ Specify that you wish to have an appointment in the GUPPE Clinic ▪ Send relevant medical history/physical/diagnostics and growth chart; describe specific physical examination findings in the referral evaluation portion if possible (see Initial Evaluation) ▪ Provide patients/families with the GUPPE clinic link Clinic for Variations of Sex Development Connecticut Children's
WHAT TO EXPECT	<p>What to expect from CT Children's Visit:</p> <ul style="list-style-type: none"> ▪ History and physical exam by Endocrinology and Urology ▪ Patient and family support ▪ Evaluation of prior lab testing and growth charts ▪ Additional labs and imaging, if appropriate ▪ Recommendations regarding treatment as needed