



INTRODUCTION	<p>Disorders of Sexual Development (DSD) may lead to ambiguous genitalia - a rare condition where it is unclear whether the external and/or internal genitalia are entirely male or female. 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 ambiguous appearance on exam.</p> <p>Infants ambiguous at birth are identified, diagnosed and managed in the immediate newborn period. This guideline also applies to DSDs that may not have features of ambiguity in infancy, but present later in life. In presumed females this could include lack of breast development, primary amenorrhea or unexpected virilization. In presumed males this could include lack of axillary/pubescent hair, micropenis or significant hypospadias.</p> <p>Our patients with DSD receive care from our multidisciplinary GUPPE team, which includes subspecialists from Genetics, Urology, Psychology, Psychiatry, and Endocrinology. Our experienced clinicians are familiar with the various aspects of each unique condition as well as working with families. Each subspecialist brings their unique expertise and perspective, and this collaborative effort allows us to develop the best possible treatment plan for each patient.</p>
INITIAL EVALUATION AND MANAGEMENT	<p>Initial Evaluation:</p> <ul style="list-style-type: none">● Obtain targeted history:<ul style="list-style-type: none">○ Include neonatal exposure to any medications including topical hormone creams, maternal history of any virilization during pregnancy, birth history and any family history of genital concerns, pubertal disorders, infertility or sudden death.● Perform targeted physical exam:<ul style="list-style-type: none">○ Include genital exam.<ul style="list-style-type: none">▪ Overvirilized XX▪ Undervirilized XY <p>Initial Management:</p> <ul style="list-style-type: none">● If the evaluation is suggestive of a DSD, please provide family with an overview of DSD and reassurance
WHEN TO REFER	<p>Routine Referral (within 1 month)</p> <ul style="list-style-type: none">● Please refer as soon as a disorder of sexual development is suspected. Early diagnosis and management leads to better medical and psychological outcomes for these families.
HOW TO REFER	<p>Referral to the DSD clinic</p> <ul style="list-style-type: none">● A referral to the DSD clinic can be made to the Department of Urology: Fax: (860) 545-9036; Phone: (860) 545-9520<ul style="list-style-type: none">▪ <i>Please specify that you wish to have an appointment for GUPPE</i>● With the referral, please send relevant medical history/physical/diagnostics and growth chart● If making a referral, please provide patients/families with a DSD clinic brochure
WHAT TO EXPECT FROM CONNECTICUT CHILDREN'S VISIT	<ul style="list-style-type: none">● History and physical exam by endocrinology and urology● Comprehensive patient evaluation by psychology/psychiatry● Evaluation of prior lab testing and growth chart● Additional labs and imaging if appropriate● Recommendations regarding surgery and hormone therapy as needed