



# **Genetic Steroid Disorders: Chapter 3E. Ambiguous Genitalia in Newborns**

*Sowmya Krishnan, Amy B. Wisniewski*

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Ambiguous genitalia can be associated with disorders of sex development (DSD). DSD occurs when a person is born with discordant genetic, gonadal, or anatomic sex. Here we discuss typical-appearing external genital appearance in unaffected males and females followed by descriptions of ambiguous genitalia in newborns with 46,XY DSD, 46,XX DSD, syndromes associated with multiple congenital anomalies including, but not limited to, ambiguous genitalia, ovotesticular DSD, and mixed gonadal dysgenesis in newborns who possess a Y chromosome. We provide guidance to proceed with a clinical work-up to differentiation between types of DSD that result in ambiguous genitalia at birth. Finally, we discuss how gender assignments are made for newborns with DSD including ambiguous genitalia.

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