



Proceeding

Volume 3 Suppl 1 – March 2017
DOI: 10.19080/AJPN.2017.03.555630

Acad J Ped Neonatol

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Disorders of Sex Development; A Structured Approach on Diagnosis and Management

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Submission: March 05, 2017; **Published:** March 20, 2017

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Abstract

Genital ambiguity is defined as difficulty in determining sex. It occurs in 1 in every 10000 live births but about 1 in every 350 is still born. Knowledge of embryology and differential diagnosis is important. The proposed revised nomenclature for Disorders of Sex Development (DSD) includes: 46, XY DSD, 46, XX DSD, ovo testicular DSD, 46, XX testicular DSD and 46, XY gonadal

disgenesis. A systematic approach based on whether the gonads are palpable or not and on laboratory investigations is important for prompt diagnosis. A multidisciplinary team is needed for proper diagnosis and management but one contact person for the parents is preferred. Physicians must consider the situation as « urgent » and a proper decision needs to be made. It is important to consider psychosexual outcome and not only fertility.



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DOI: [10.19080/AJPN.2017.03.555630](https://doi.org/10.19080/AJPN.2017.03.555630)

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