

CHAPTER 7

ENDOCRINE SYSTEM

7.1 DISORDERS OF SEX DEVELOPMENT (DSD)

Q52.9/ Q55.9

DESCRIPTION

The current terminology for neonates or children presenting with incomplete differentiation of the external genitalia is "disorder of sex development".

DIAGNOSTIC CRITERIA

Clinical

- DSDs present with one or more of the following:
 - > varying degrees of hypospadias,
 - > maldescent of one or both gonads,
 - > atypical size of the phallus,
 - > scrotalisation of the labia, and
 - > a urogenital sinus.
- Isolated hypospadias is not a DSD.

Suspect congenital adrenal hyperplasia in an infant with non-palpable gonads and DSD.

Investigations

- Urgent urea/electrolytes, venous blood gas and blood glucose to identify possible adrenal insufficiency.
- Elevated 17-hydroxyprogesterone level to confirm a diagnosis of adrenal hyperplasia (to be done after day 3 of life for an accurate interpretation of the result).
- Further investigations (in discussion with referral centre):
 - > Genitourinary imaging (e.g. ultrasound).
 - > Genetic evaluation.

GENERAL AND SUPPORTIVE MEASURES

- Gender assignment in these infants should only be undertaken after extensive counselling and evaluation by a multidisciplinary team.
- Stabilise all neonates suspected of having congenital adrenal hyperplasia with a salt-losing crisis prior to urgent referral, as a crisis may be life threatening.

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Katrin Zwirglmaier



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