Infant with ambiguous genitalia

Suggestive history and physical findings	Initial laboratory and/or radiologic work-up can include:	When to refer	Items useful for consultation	Additional information
History: poor feeding, lethargy. Family history of SIDS **Physical findings:** Low or high blood pressure Ambiguous genitalia: including bilateral cryptorchidism, hypospadias with unilateral cryptorchidism, posterior labial fusion Hyperpigmentation Differential Diagnosis	**Blood tests:** • Sodium • Potassium • Glucose • Cortisol (7-8 am) • 17-OH progesterone • Karyotype Other tests to consider after consultation with Pediatric Endocrinologist • Hormone studies • FISH for SRY • Abdominal/pelvic ultrasound	**Urgent: All cases of ambiguous genitalia are considered an emergency and should be promptly referred** to a pediatric endocrinology team with a multidisciplinary approach to these patients. Gender assignment is not done until evaluation is completed by the multidisciplinary team Find a Pediatric Endocrinologist	Results of newborn screen Pertinent medical records Recent laboratory and radiologic studies	Additional Information Congenital Adrenal Hyperplasia: A Guide for Families Genetic Testing: A Guide for Families References

Etiology: Can be broadly classified into:

XY DSD	Partial gonadal dysgenesis Deficiency of testosterone biosynthesis 5 alpha reductase-2 deficiency Abnormal androgen receptor activity
XX DSD	Abnormal fetal androgen production Excess maternal androgen production Placental aromatase deficiency Drugs administered to mother during pregnancy
Syndromes with multiple congenital abnormalities	VATER syndrome CHARGE Syndrome
Ovo testicular DSD	46,XX with translocation of SRY

Additional Information:

While evaluating a child with ambiguous genitalia the primary concern should be

1. Is this associated with a life threatening illness? Congenital adrenal hyperplasia is associated with adrenal insufficiency (and may also be associated with salt wasting), which if not recognized and treated urgently can lead to mortality in the infant.
2. Gender of rearing: Ideally a decision about gender of rearing should be made as early as possible **but only after appropriate work up in the setting of a multidisciplinary team** has been done. It is very traumatic for family and the patient to change the gender of rearing later on in life. So care should be taken to avoid calling the baby: baby boy or baby girl until appropriate work up is done and a decision has been made. Certain factors that go into this decision include the underlying etiology, potential for fertility, need for multiple surgeries and what is known about the long term outcome of individuals with this condition. Medical team should explain the process of sex determination and differentiation to the parents and they should be given time to think about their choices.

Suggested References and Additional Reading:

- Wisniewski AB, Krishnan S. Ambiguous Genitalia in the Newborn. 2012 Nov 15. In: De Groot LJ, Beck-Peccoz P, Chrousos G, Dungan K, Grossman A, Hershman JM, Koch C, McLachlan R, New M, Rebar R, Singer F, Vinik A, Weickert MO, editors. Endotext [Internet]. South Dartmouth (MA): MDText.com, Inc.; 2000-. Available From http://www.ncbi.nlm.nih.gov/books/NBK279168/ PubMed PMID: 25905391.
- Hughes IA, Houk C, Ahmed SF, Lee PA. LSWPE/ESPE consensus group. Consensus statement on management of intersex disorders. Arch Dis Child 2006, 91:554-563.

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