## Infant with ambiguous genitalia

<table>
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<tr>
<th>Suggestive history and physical findings</th>
<th>Initial laboratory and/or radiologic work-up can include:</th>
<th>When to refer</th>
<th>Items useful for consultation</th>
<th>Additional information</th>
</tr>
</thead>
</table>
| **History:** poor feeding, lethargy. Family history of SIDS | **Blood tests:**  
- Sodium  
- Potassium  
- Glucose  
- Cortisol (7-8 am)  
- 17-OH progesterone  
- Karyotype | **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. | 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** |
| **Physical findings:** Low or high blood pressure | **Other tests to consider after consultation with Pediatric Endocrinologist**  
- Hormone studies  
- FISH for SRY  
- Abdominal/pelvic ultrasound | | | |
| Ambiguous genitalia: including bilateral cryptorchidism, hypospadias with unilateral cryptorchidism, posterior labial fusion  
Hyperpigmentation | | | | |
| **Differential Diagnosis** | | | | |
**Etiology: Can be broadly classified into:**

<table>
<thead>
<tr>
<th>XY DSD</th>
<th>XX DSD</th>
<th>Syndromes with multiple congenital abnormalities</th>
<th>Ovo testicular DSD</th>
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</thead>
<tbody>
<tr>
<td>Partial gonadal dysgenesis</td>
<td>Abnormal fetal androgen production</td>
<td>VATER syndrome</td>
<td>46,XX with translocation of SRY</td>
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<tr>
<td>Deficiency of testosterone biosynthesis</td>
<td>Excess maternal androgen production</td>
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<tr>
<td>5 alpha reductase-2 deficiency</td>
<td>Placental aromatase deficiency</td>
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<tr>
<td>Abnormal androgen receptor activity</td>
<td>Drugs administered to mother during pregnancy</td>
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**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:


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