Practical Approach To Imaging Children With Ambiguous Genitalia

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INTRODUCTION

Ambiguous genitalia in neonate can be of varied etiologies and may be associated with a wide spectrum of outcomes ranging from normal female or male development to severe congenital anomalies requiring urgent intervention and surgical correction. Appropriate imaging is necessary to assess neurological and systemic anomalies, genitourinary tract anatomy and development, and sexual differentiation.

EPIDEMIOLOGY

The incidence of ambiguous genitalia is 1 in 10,000 live births. Causes of ambiguous genitalia can be classified into primary and secondary types. Primary causes include genetic errors (46XY, 46XX, 45X, etc.) and gonadal dysgenesis. Secondary causes include fetal exposure to androgens or estrogens, maternal diabetes, congenital infections, chromosomal abnormalities, and chromosomal mosaicism.

CLASIFICATION

According to anatomical classification, ambiguous genitalia may be present as Pure Testicular Tissue, Pure Gonadal Dysgenesis, and Mixed Gonadal Dysgenesis. The presence of testicular tissue, complete or incomplete, is important to differentiate between pseudohermaphrodite, true hermaphrodite, and gonadal dysgenesis.

IMAGING OF AMBIGUOUS GENITALIA

1. **Ultrasound (US)**: US is a useful initial imaging modality for evaluating the presence of testicular tissue, determination of fetal gender, and monitoring fetal growth. It can be performed in utero or in neonates.
2. **Magnetic Resonance Imaging (MRI)**: MRI is the modality of choice for evaluating the internal anatomy and development of the genitourinary tract. It can be performed in utero or in neonates.
3. **Computed Tomography (CT)**: CT is useful in evaluating the presence of calcification or neoplastic changes and can be performed in utero or in neonates.

DIAGNOSTIC APPROACH

A multidisciplinary approach involving geneticist, urologist, and radiologist is necessary for the evaluation of ambiguous genitalia. Imaging plays a crucial role in the diagnosis and management of these conditions.

REFERENCES