Answers to Medical Quiz

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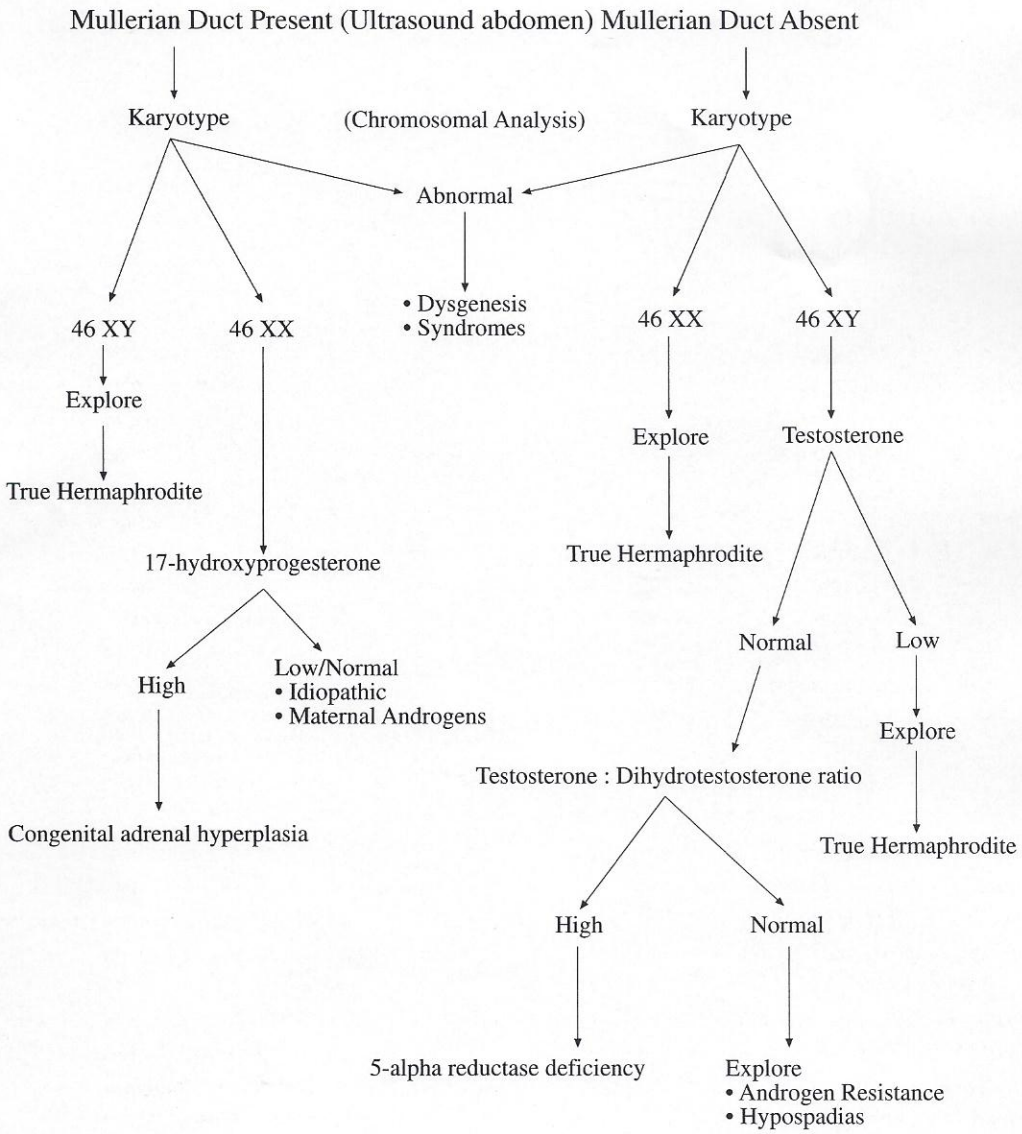
A1. Ambiguous Genitalia

A2. Chromosomal, Genetic, Structural (see below)

A3. The differential diagnosis of the condition includes various endocrinopathies or chromosomal disorders. The work up to reach the diagnosis is shown in Figure 1.

Figure 1: Diagnostic algorithm for Ambiguous Genitalia



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The two primary investigations include an abdominal ultrasound (US) and chromosomal analysis. When the US suggests the presence of mullerian duct, the possibility of a female gender is more likely. On the contrary, when mullerian duct is absent, the male sex is the possible diagnosis. The next step is to do karyotyping. If it is abnormal, the diagnosis of dysgenesis or associated syndromes should be entertained.

In a patient with the presence of mullerian duct and a karyotype of 46 XY, or no mullerian duct with a karyotype of 46 XX, the diagnosis of true hermaphrodite is considered and the patient should be explored to confirm that.

When mullerian duct is absent with 46 XY status:

Check the testosterone levels. If low, explore for true hermaphrodite. If normal, obtain a testosterone dihydrotestosterone ratio. If the ratio is high the diagnosis is 5 alpha reductase deficiency and if the ratio is normal the diagnosis could be hypospadias or androgen resistance.

When mullerian duct is present with 46 XX status:

Obtain the 17-hydroxyprogestrone level. If high, the diagnosis is congenital adrenal hyperplasia. If low or normal, the diagnosis could be idiopathic or secondary to maternal androgens.

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