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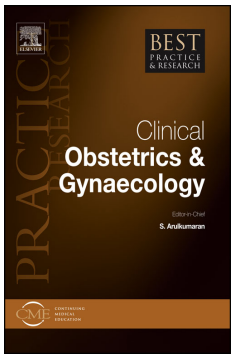

Genetics of gynaecological conditions

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I. FEMALE GENITAL TRACT MALFORMATIONS

A. SPORADIC (MULTIFACTORIAL) FEMALE GENITAL TRACT MALFORMATIONS (FGTA)

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Abstract:

From genomic imbalances associated with developmental abnormalities of the female genital tract to the molecular mechanisms underpinning endometriosis and uterine leiomyomatosis, new technologies have allowed exploration of the genetic contribution and mapping the molecular pathways underpinning common and rare gynaecological conditions.

While some of these conditions have historically been considered sporadic, recent research has demonstrated their heritable nature linked to single genes or copy number variants. The phenotypic variability including non-penetrance indicates their multifactorial, complex aetiology encompassing genetic, epigenetic and environmental influences.

Although genetic tests are not routinely undertaken in gynaecological practice, there is an increasing body of evidence suggesting that, in appropriate cases, molecular investigations such as array CGH analysis may be an important part of the diagnostic algorithm. The subtlety of clinical features, especially in the context of syndromic diagnoses, requires the practitioner to become familiar with those conditions and how to approach diagnostic investigations.

This chapter combines the recent research output related to gynaecological disorders with a holistic, clinical genetics approach aiming to highlight the multisystem character of some of these conditions, their implications for management, reproductive risks and options, as well as the importance of genetic counselling.