ILLUMI SCREEN

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Highlights From ACOG-SMFM

Practice bulletin #163 on screening for fetal aneuploidy

INTRODUCTION

The American Congress of Obstetricians and Gynecologists (ACOG) and the Society for Maternal-Fetal Medicine (SMFM) issued a joint Practice Bulletin on screening for aneuploidy (PB #163) in May 2016. This bulletin reviews current information for fetal aneuploidy screening options. PB #163 replaces the previous PB #77 from January 2007.

This information sheet highlights key points related to cell-free DNA (cfDNA) screening, which is the technology used in noninvasive prenatal testing (NIPT).

WHAT IS NEW IN cfDNA SCREENING?

Practice Bulletin #163 now recognises that cfDNA screening is an option for determining the risk of fetal aneuploidy in all pregnant women, regardless of maternal age, and highlights follow-up recommendations for patients with a positive result or a test failure.

"Women whose cfDNA test results are not reported, are indeterminate, or are uninterpretable (eg, a no-call test result) should receive further genetic counselling, and the offer of comprehensive ultrasound evaluation and diagnostic testing because of an increased risk of aneuploidy."

KEY FACTS ABOUT cfDNA SCREENING

- cfDNA screening is not a substitute for diagnostic testing.
- All patients with positive cfDNA test results should be offered confirmatory diagnostic testing before any irreversible decision, such as pregnancy termination, is made.
- The positive predictive value (PPV) of cfDNA screening is dependent on the prevalence of the condition.

GENERAL RECOMMENDATIONS

- All women, irrespective of maternal age, should be offered aneuploidy screening and diagnostic testing.
  - Ideally, testing options should be discussed at the first prenatal visit.
  - The risk of fetal aneuploidy, and the benefits, risks, and limitations of the different testing options should be reviewed with the patient—aneuploidy testing should be an informed patient choice.
- It is not cost-effective to perform multiple screening tests in parallel.
- Patients who conceive after preimplantation genetic screening (PGS) for aneuploidy should be offered aneuploidy screening and diagnostic testing because of the potential for false-negative results with PGS.

To learn more about NIPT using the Illlumiscreen prenatal test, visit **illumiscreen.co.nz**