



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.

Disclaimer: This summary is NOT intended to highlight the benefits and limitations of all screening and diagnostic options for pregnant women. This summary is also NOT intended to review all the recommendations and discussion included in Practice Bulletin #163, and is NOT intended to make recommendations relating to the practice of medicine or to substitute for the independent professional judgment of a licensed physician.

Next generation prenatal testing delivered by Healthscope NZ.

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