

PRACTICAL PEARLS: Prenatal Screening

INTRODUCTION	<ul style="list-style-type: none">• Noninvasive prenatal screening (NIPS or NIPT) is a test for abnormal chromosome number that uses a sample of blood from a pregnant woman after about 8 weeks gestation• Analysis of maternal blood for cell free (floating) fragments of DNA (cfDNA) from the placenta.• cfDNA is sequenced and counted. Complex algorithms detect pregnancies at high risk for trisomies 21, 18, or 13.• Newer versions can detect other chromosome abnormalities such as microdeletion syndromes (22q11).• This is a screening test; positive results need to be confirmed with a diagnostic test (chromosome analysis via CVS, amniocentesis, postnatal blood testing). False positives and false negatives occur! ACMG Statement, Genet Med 18:1056-1065, 2016 http://www.acmg.net/docs/ACMG_NIPS_FINAL_STATEMENT_GIM_Oct%202016.pdf ACOG Committee Opinion Number 640, 2015. http://www.acog.org/Resources-And-Publications/Committee-Opinions/Committee-on-Genetics/Cell-free-DNA-Screening-for-Fetal-Aneuploidy
INITIAL EVALUATION AND MANAGEMENT BY PRIMARY CARE	<ul style="list-style-type: none">• NIPS is offered to high risk women (advanced maternal age, previous pregnancy with trisomy, fetal ultrasound abnormalities).• First trimester screening remains standard of care aneuploidy screening for low risk women (ACOG 2015). However, NIPS can be appropriate for low risk women (ACMG 2016).• Importantly, the positive predictive value of an abnormal test in a low risk woman may be quite low, so pre- and post-test counseling are extremely important.
WHEN TO REFER	<ul style="list-style-type: none">• Pre or post-conception patients interested in genetic testing options.
HOW TO REFER	<ul style="list-style-type: none">• (413) 794-KIDS• (413) 794-8890
WHAT TO EXPECT FROM BAYSTATE CHILDREN'S HOSPITAL VISIT	<ul style="list-style-type: none">• Our Reproductive Genetic Counselors carefully consider indications for genetic testing, interpret genetic test results and deliver informed and compassionate genetic counseling.

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Genetics

June 17

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