

**PRACTICAL PEARLS: Prenatal Screening**

<p><b>INTRODUCTION</b></p>	<ul style="list-style-type: none"> <li>• 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</li> <li>• Analysis of maternal blood for cell free (floating) fragments of DNA (cfDNA) from the placenta.</li> <li>• cfDNA is sequenced and counted. Complex algorithms detect pregnancies at high risk for trisomies 21, 18, or 13.</li> <li>• Newer versions can detect other chromosome abnormalities such as microdeletion syndromes (22q11).</li> <li>• 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 <a href="http://www.acmg.net/docs/ACMG_NIPS_FINAL_STATEMENT_GIM_Oct%202016.pdf">http://www.acmg.net/docs/ACMG_NIPS_FINAL_STATEMENT_GIM_Oct%202016.pdf</a> ACOG Committee Opinion Number 640, 2015. <a href="http://www.acog.org/Resources-And-Publications/Committee-Opinions/Committee-on-Genetics/Cell-free-DNA-Screening-for-Fetal-Aneuploidy">http://www.acog.org/Resources-And-Publications/Committee-Opinions/Committee-on-Genetics/Cell-free-DNA-Screening-for-Fetal-Aneuploidy</a></li> </ul>
<p><b>INITIAL EVALUATION AND MANAGEMENT BY PRIMARY CARE</b></p>	<ul style="list-style-type: none"> <li>• NIPS is offered to high risk women (advanced maternal age, previous pregnancy with trisomy, fetal ultrasound abnormalities).</li> <li>• 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).</li> <li>• 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.</li> </ul>
<p><b>WHEN TO REFER</b></p>	<ul style="list-style-type: none"> <li>• Pre or post-conception patients interested in genetic testing options.</li> </ul>
<p><b>HOW TO REFER</b></p>	<ul style="list-style-type: none"> <li>• (413) 794-KIDS</li> <li>• (413) 794-8890</li> </ul>
<p><b>WHAT TO EXPECT FROM BAYSTATE CHILDREN'S HOSPITAL VISIT</b></p>	<ul style="list-style-type: none"> <li>• Our Reproductive Genetic Counselors carefully consider indications for genetic testing, interpret genetic test results and deliver informed and compassionate genetic counseling.</li> </ul>

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