University of North Carolina at Chapel Hill
Department of OB/GYN
Partner Clinic Information

Genetic Screening and Testing

Updated: May 3, 2013

	Any time after 10 weeks	**First Trimester**		**Second Trimester**		
	Screening Test	*Screening Test*	*Diagnostic Test*	*Screening Test*		*Diagnostic Test*
Name	NIPT (Non-invasive prenatal testing)	First trimester screening – *traditional**	CVS	Quad Screen	Genetic Sonogram	Amniocentesis
Available to	Patients with a risk factor[¥]	All patients	All patients	All patients	Patients with a risk factor[¥]	All patients
Timing	After 10 Weeks	11-1/7 to 13-6/7 Weeks	10 to 13-6/7 Weeks	15 to 22-6/7 Weeks	18-20 Weeks	After 15 Weeks
Detection Rate	~99% for Down syndrome 98-99% for trisomy 18 79-92% for trisomy 13	85% for Down syndrome 90% for trisomy 18 and trisomy 13	>99% Accuracy (for chromosomal abnormalities)	80% for Down syndrome 60% for Trisomy 18 80% for NTDs	50% for Down syndrome ≥90% for Trisomy 18 and Trisomy 13 90% for NTDs (in qualified centers)	>99% Accuracy (for chromosomal abnormalities) >97% for NTDs
Risk	Non-invasive	Non-Invasive	0.5% Miscarriage Risk	Non-Invasive	Non-Invasive	0.2-0.3% Miscarriage Risk
Results	2 Weeks	3-5 days	10-14 days	1 Week	Immediate	10-14 days

[¥]Risk factors for Down syndrome, trisomy 18, or trisomy 13 include maternal age ≥35 at EDD, a positive first or second trimester screening test, abnormal ultrasound findings, or a prior affected pregnancy.

*First trimester screening *with instant results* is also available. In this event, blood can be collected by referring provider after 9-0/7. Patient can then get results instantly after her ultrasound. If your group wishes to be set-up for first trimester screening *with instant results* please call 919-966-2229 to discuss.

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References

1. ACOG Practice Bulletin #77 – Screening for Fetal Chromosomal Abnormalities, January 2007
2. Perni SC, Predanic M, Kalish RB, et al. Clinical use of first-trimester aneuploidy screening in a United States population can replicate data from clinical trials. *Am J Obstet Gynecol*. 2006; 194(1): 127-130.
3. Spencer K, Nicolaides KH. A first trimester trisomy 13/19 risk algorithm combining fetal nuchal translucency thickness, maternal serum free Beta-hCG and PAPP-A. *Prenat Diagn*. 2002; 22:877-879.
4. Palomaki GE, Kloza EM, Lambert-Messerlian GM, Haddow JE, Neveux LM, Ehrich M, van den Boom D, Bombard AT, Deciu C, Grody WW, Nelson SF, Canick JA. DNA Sequencing of Maternal Plasma to Detect Down Syndrome: An International Clinical Validation. *Genet Med.* Vol 13:No 11. Nov 2011.
5. Palomaki GE, Deciu C, Kloza EM, Lambert-Messerlian GM, Haddow JE, Neveux LM, Ehrich M, van den Boom D, Bombard AT, Grody WW, Nelson SF, Canick JA. DNA sequencing of maternal plasma reliably identifies trisomy 18 and trisomy 13, as well as Down syndrome: An international collaborative study.*Genet Med.* 2012 Mar 1.
6. Canick JA, Kloza EM, Lambert-Messerlian GM, Haddow JE, Ehrich M, van den Boom D, Bombard AT, Deciu C, Palomaki GE. DNA sequencing of maternal plasma to identify Down syndrome and other trisomies in multiple gestations. Prenat Diagn. 2012 May 14:1-5

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Notification to Users

These algorithms are designed to assist the primary care provider in the clinical management of a variety of problems that occur during pregnancy. They should not be interpreted as a standard of care, but instead represent guidelines for management. Variation in practices should take into account such factors as characteristics of the individual patient, health resources, and regional experience with diagnostic and therapeutic modalities.

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