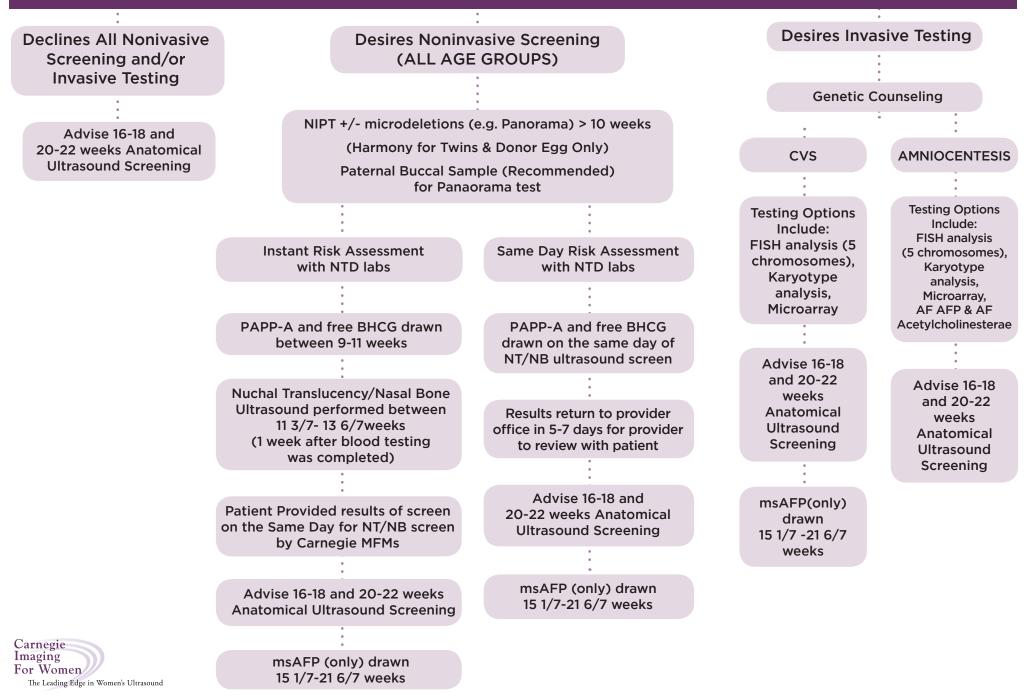
Guidelines for Fetal Aneuploidy Testing

ALL Pregnant Women Offered Invasive Testing OR Noninvasive Screening For Fetal Genetic/Chromosomal Conditions



Relevant Facts Associated with Aneuploidy Screening

Risk of miscarriage 1/300 to 1/500 per ACOG Opinion 88, 2007 for Either Amniocentesis or Chorionic Villous Sampling

SCREENING

	Maternal Serum Screening (MSS)	NIPT		
Timing	11-13 weeks and/or 15-22 weeks	>10 weeks		
False Positive Rate	5%	0.1% Panorama 1.0% with Microdeletions		
Number of Chromosome Conditions Tested	2-3 Conditions T21 T18 (T13)	Chromosomes/ Conditions T21 T18 T13 Triploidy X,Y Aberrations	Microdeletions 22q11.2 deletion/DiGeorge** 1p36 deletion Angelman Cri-du-Chat Prader Willi	

** 22q11.2 microdeletion Panorama screen identifies 87% of DiGeorge Syndrome. PPV of 22q11.2 microdeletion is estimated to be 1:18

Nuchal Translucency	Aneuploidy	Fetal Death	Live Born without Abnormality	Cardiac Abnormalities
>95th %tile -3.4 mm	4%	1.3%	93%	2.5%
3.5-4.4 mm	21%	2.7%	70%	10%
4.5-5.4 mm	33%	3.4%	50%	18.5%
5.6-6.4 mm	51%	10.1%	30%	24.2%
>6.5 mm	65%	19%	15%	46.2%

Reasons for discrepancy between Invasive Testing & NIPT: True Fetal mosaicism, Confined placental mosaicism, Aneuploidy dizygotic twin demise ("vanishing twin"), Nonmosaic maternal chromosome abnormality (eg. 47XXX), maternal somatic cell variation, maternal malignancy, Lab error, Low fetal fraction