Guidelines for Fetal Aneuploidy Testing

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

Declines All Nonivasive Screening and/or Invasive Testing

- Advise 16-18 and 20-22 weeks Anatomical Ultrasound Screening

Desires Noninvasive Screening (ALL AGE GROUPS)

- NIPT +/- microdeletions (e.g. Panorama) > 10 weeks
  (Harmony for Twins & Donor Egg Only)
  Paternal Buccal Sample (Recommended) for Panaorama test

Instant Risk Assessment with NTD labs

- PAPP-A and free BHCG drawn between 9-11 weeks
- Nuchal Translucency/Nasal Bone Ultrasound performed between 11 3/7- 13 6/7weeks (1 week after blood testing was completed)
- Patient Provided results of screen on the Same Day for NT/NB screen by Carnegie MFMs
- Advise 16-18 and 20-22 weeks Anatomical Ultrasound Screening
- msAFP (only) drawn 15 1/7-21 6/7 weeks

Same Day Risk Assessment with NTD labs

- PAPP-A and free BHCG drawn on the same day of NT/NB ultrasound screen
- Results return to provider office in 5-7 days for provider to review with patient
- Advise 16-18 and 20-22 weeks Anatomical Ultrasound Screening
- msAFP (only) drawn 15 1/7-21 6/7 weeks

Desires Invasive Testing

- Genetic Counseling

CVS

- Testing Options Include: FISH analysis (5 chromosomes), Karyotype analysis, Microarray
- Advise 16-18 and 20-22 weeks Anatomical Ultrasound Screening
- msAFP(only) drawn 15 1/7 -21 6/7 weeks

AMNIOCENTESIS

- Testing Options Include: FISH analysis (5 chromosomes), Karyotype analysis, Microarray, AF AFP & AF Acetylcholinesterae
- Advise 16-18 and 20-22 weeks Anatomical Ultrasound Screening

Carnegie Imaging For Women
The Leading Edge in Women's Ultrasound

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