Screening for Chromosome Abnormalities

- Screening tests are offered early in pregnancy to see if patients are at increased risk for chromosomal abnormalities

Screening for Chromosome Abnormalities

- Testing options available to patients are based on their level of risk
- While the risk for chromosome abnormalities increases with the mother's age at delivery, most babies with these defects are actually born to younger women
- Screening is optional and should only be performed after a thorough discussion of available tests with your provider

Screening for Chromosome Abnormalities

- Low risk individuals may undergo one of two tests:
  - Sequential Screen (Ultrasound and blood draw before 14 weeks)
  - Quadruple Marker Screen (Blood draw after 15 weeks)

Low Risk Screening
Sequential Screen

- Combines:
  - Mother's age at the time of delivery
  - Ultrasound
  - Blood tests
- Determines the risk for chromosome abnormalities, including Down syndrome and trisomy 18, and a group of birth defects known as open neural tube defects, which includes spina bifida

Low Risk Screening
Sequential Screen

- Ultrasound examination must be performed between 11 and 13-6/7 weeks gestational age
- Measures nuchal translucency, a fluid filled space behind the baby's neck

Low Risk Screening
Sequential Screen

- Detects 90-92 percent of cases of Down syndrome and 90 percent of cases of trisomy 18, with a false positive rate of 5 percent
- Identifies approximately 80 percent of babies with open neural tube defects, and when combined with ultrasound in the 2nd trimester, it detects virtually all of these defects

Low Risk Screening
Quadruple Marker Screen

- Available between 15 and 21 weeks gestational age
- Blood test that measures the level of certain hormones in the mother's blood

Low Risk Screening
Quadruple Marker Screen

- Can detect 75-80 percent of cases of Down syndrome and 60-75 percent of cases of trisomy 18
- Can also detect 80 percent of cases of spina bifida
- False positive rate is 5-7 percent

High Risk Screening
Non-invasive Prenatal Testing

- New test that evaluates baby's DNA in the mother's blood
- Can be performed any time after 10 weeks' gestational age, but it is usually performed in conjunction with an ultrasound in the first trimester and assessment of nuchal translucency

High Risk Screening
Non-invasive Prenatal Testing (NIPT)

- Women should consider NIPT if any of the following apply:
  - Age of 35 or over at the time of delivery
  - Abnormal screening test (sequential or quad screen)
  - Abnormal ultrasound findings
  - Chromosome abnormality in previous pregnancy

High Risk Screening
Non-invasive Prenatal Testing (NIPT)

- Can detect approximately 99 percent of cases of Down syndrome and trisomy 18, and up to 92 percent of cases of trisomy 13
- Can also detect some abnormalities involving the sex chromosomes and reveal the gender of the baby
- False positive results are rare but possible

Diagnostic Testing

- Chorionic villus sampling or amniocentesis are still needed to confirm an abnormal NIPT result
- May be indicated if there are abnormalities seen on ultrasound, as NIPT does not test for all chromosome abnormalities