

# 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 2<sup>nd</sup> 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