



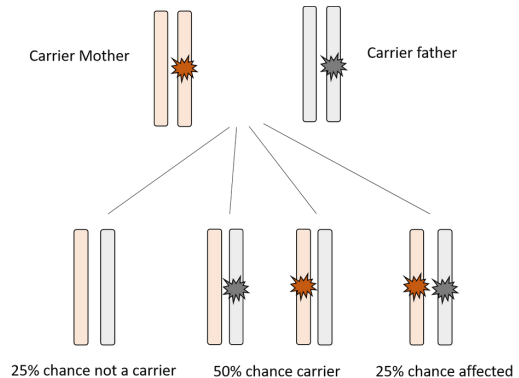
### GENETIC SCREENING AND TESTING IN PREGNANCY

#### CARRIER SCREENING

- A blood test to determine if you and your partner are at increased risk to have a baby with a specific genetic disorder
- Most of these conditions are autosomal recessive - if both parents are carriers for the same genetic condition there is a 25% chance of having a child with this condition
- Some conditions are X-linked - if a mother is a carrier of an X-linked condition, sons have a 50% chance of being affected

Examples of conditions included on carrier screening:

- cystic fibrosis
- sickle cell anemia
- alpha thalassemia
- Fragile X syndrome



#### SCREENING FOR CHROMOSOMAL CONDITIONS

##### NON INVASIVE PRENATAL TESTING/SCREENING (NIPT/NIPS/CELL FREE DNA SCREENING)

- Blood test that screens for Down syndrome (Trisomy 21), Trisomy 13, Trisomy 18 and the sex chromosomes
- Can be done as early as 9-10 weeks gestation
- Does not replace diagnostic testing.

##### NUCHAL TRANSLUCENCY (NT) SCAN

- Sonogram at ~11-14 weeks gestation
- Measures pocket of fluid at back of fetus' neck
- An increased NT measurement is considered to increase the risk for genetic conditions

#### DIAGNOSTIC GENETIC TESTING OPTIONS

- Provides diagnostic (definitive) information vs screening provided by NIPT
- Can provide additional information on less common genetic syndromes not included on NIPT
- Involves an invasive procedure

##### CVS

- Can be done at approximately 10-13 weeks gestation
- Sample taken from the placenta for genetic testing

##### AMNIOCENTESIS

- Can be done as early as 15-16 weeks gestation
- Sample of amniotic fluid taken for genetic testing

Discuss with your doctor if genetic counseling is right for you