Prenatal Screening Tests

What is a screening test?

- An ultrasound and/or bloodtest
- It reports the likelihood, chance or risk that your baby has a birth defect
- A screening test **does not** give a "yes" or "no" answer
- A negative/normal result = your baby has low risk of having a birth defect
- A positive result = there is a higher-than-average chance your baby has a birth defect
- Women with an increased risk are offered further information and testing which may include a detailed ultrasound and/or diagnostic testing, such as an amniocentesis

Is there routine screening in the pregnancy?

- Yes! The routine ultrasound booked for all pregnant patients between 18-20 weeks of your pregnancy is also a screening test
- This ultrasound looks at baby's physical features, so any abnormalities will be reported to your doctor

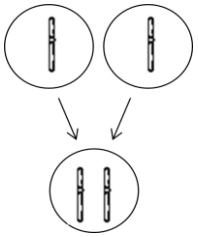
What are the benefits of prenatal screening?

- Early screening gives peace of mind to many families
- The screening risk estimate can be used to assist women in making a more informed choice about diagnostic testing
- Some major birth defects can be detected from screening scan, but not all

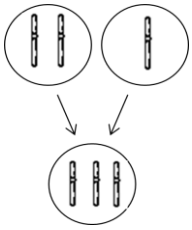
What birth defects can be screened?

There are 2 types of birth defects that can be screened:

1. An extra chromosome



Normal Chromosome Pair



Abnormal Chromosome Trisomy

Down Syndrome

- The most common chromosome condition – an extra chromosome 21
- Babies will have delayed physical and intellectual development
- There is a slightly greater chance for having a baby with Down syndrome as a woman ages

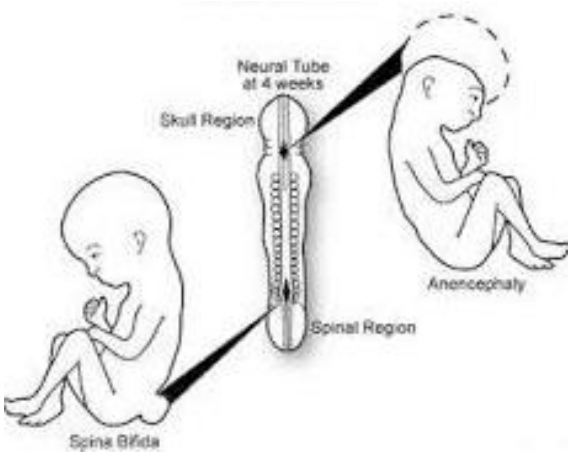
Trisomy 13 and Trisomy 18

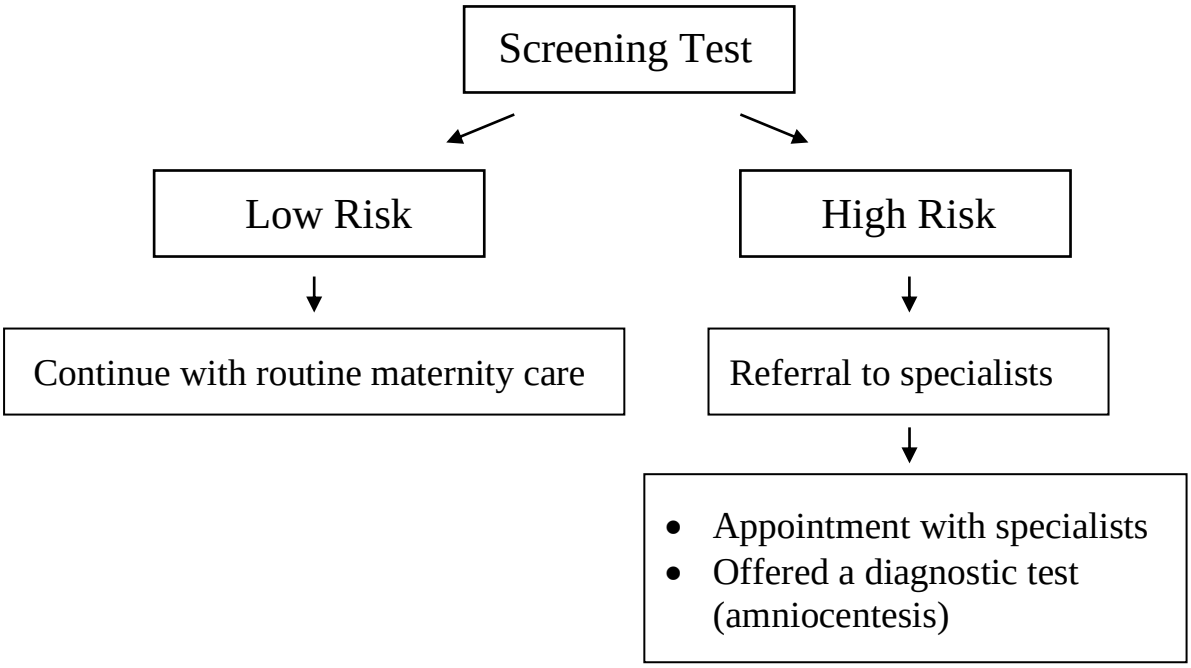
- Babies are typically small and have many physical abnormalities and problems with internal organs
- Most pregnancies with trisomy 13 or 18 will miscarry or die when they are infants

2. A problem with your baby's spine development

Spine/Neural Tube Defects (NTDs)

- The neural tube is a part of the baby that forms very early in development
- The upper part of the neural tube forms the baby's brain and the lower part forms the spine
- Examples of NTDs are anencephaly (brain) and spina bifida (spine)





What screening tests are available in pregnancy?

	First Trimester/ Nuchal Translucency	Maternal Serum Screen (MSS)	Non-Invasive Prenatal Screening (NIPS)
Cost	Free	Free	~ $140 - $500
How is the test done?	Ultrasound + blood test (same day)	Blood test	Blood test (Need a kit)
Gestational age	11 – 13wks	15 – 20wks	From 10wks
Detection rate	How many babies with the condition will be detected by the test?		
	85-90/100	80-90/100	99/100
Conditions screened	Down syndrome Trisomy 13 Trisomy 18	Down syndrome Trisomy 18 NTDs	Down syndrome Trisomy 13 Trisomy 18 Sex chromosomes

What are the limitations of prenatal screening?

- Screening tests do NOT give a yes or no answer
- About 1 in 20 women will receive an increased-risk result
    - It is normal to be worried if you hear you are at an increased risk
    - Most women with an increased risk result have healthy babies
- An increased-risk result does not mean that the baby has a chromosome condition and a reduced-risk result does not guarantee a healthy baby

What happens after the screening test?

- The doctor will explain the results of the test to you
- If the risk score is normal/negative: routine maternity care
- If the risk score is high/positive: there appears to be an increased risk of your baby having a condition
    - A referral is made to the hospital specialists to review the results
    - Further testing options will be offered such as an Amniocentesis or Chorionic Villus Sampling (CVS) to DIAGNOSE your baby

Websites

- https://myhealth.alberta.ca/Health/pages/conditions.aspx?hwid=aa21828&lang=en-ca
- https://www.invitae.com/en/individuals/reproductive-genetic-testing/non-invasive-prenatal-screening/