A Screen Positive Result

What does it mean and what do I do now?

BC Prenatal Genetic Screening Program

Perinatal Services BC
An agency of the Provincial Health Services Authority

You have learned that the result of your prenatal genetic screening is "screen positive." The screening was to find out the chance of your baby having Down syndrome, trisomy 18 or an open neural tube defect.

What does a "screen positive" result mean?

This result does not mean your baby has Down syndrome, trisomy 18 or an open neural tube defect. In fact, most women with this result do not have a baby with one of these conditions. The result means that the chance of your baby having Down syndrome, trisomy 18 or an open neural tube defect is high enough that further testing is offered to determine if your baby has any of these three conditions.

As part of your screen result, you have been given a number that estimates the chance your baby has one of the conditions screened.

Here is an example of a screen positive result:

The *chance* of Down syndrome is 1:100. This is the same as saying:

- There is a 1% chance your baby will have Down syndrome **or**
- There is a 99% chance your baby will not have Down syndrome **or**
- If 100 women had the same result, one of them would have a baby with Down syndrome and the other 99 would not.

According to your screen result, you screened positive for:

_____

The chance your baby has this condition is:

1 in _____ which is _____%

What happens now?

Your health care provider may need to confirm how far along you are in your pregnancy. It is important to make sure the pregnancy dating used for the prenatal screen result is correct.

If you have a screen positive result for an open neural tube defect, you will be offered a detailed ultrasound and an appointment with a maternal fetal medicine doctor or a genetic counsellor at one of BC's medical genetics clinics in Vancouver or Victoria. If your baby has an open neural tube defect, this is usually seen on the ultrasound scan.

If you have a screen positive result for Down syndrome or trisomy 18, you will be offered another blood test called NIPT (for non-invasive prenatal testing) or an amniocentesis.

What is Non-Invasive Prenatal Testing (NIPT)?

It is a more accurate screening test for Down syndrome and trisomy 18 than SIPS/IPS/Quad and it is done through a blood test. It detects almost all babies with Down syndrome and trisomy 18 with very few false positive results.

NIPT will give you a new risk (very low or very high) of having a baby with Down syndrome, trisomy 18 or trisomy 13. If the risk is very low, no further testing would be recommended. If the risk is very high, an amniocentesis would be needed to confirm the result. The NIPT test result is available in 10 days. For women with a positive SIPS/IPS/Quad screen, NIPT is funded by MSP.

What is an amniocentesis?

It is a diagnostic test. It tells you for sure if your baby has Down syndrome or trisomy 18. It is done by putting a very fine needle into your belly to remove a few teaspoons of amniotic fluid from around the baby. The needle is guided by ultrasound so it does not touch the baby. The baby's cells that are in the fluid sample are looked at to find out whether your baby does or does not have Down syndrome or trisomy 18. When an amniocentesis is done for a positive screen, results are available in 3 days.

Amniocentesis has a 1 in 200 (0.5%) chance of pregnancy loss. In other words, if 200 women have an amniocentesis, one would lose the pregnancy as a result of the procedure.

Should I have extra testing and if so, which one? NIPT or amniocentesis?

It is your choice. Not every woman who has a screen positive result wants extra testing. Think about these questions to help make up your mind.

- Do I think the chance that is given on my screen result is high or low?
- Do I want to know if my baby has Down syndrome or trisomy 18 before the baby is born?
- Will I be reassured if my NIPT result shows a very low risk of Down syndrome, trisomy 18 and trisomy 13?
- Do I need a result that is 100% accurate (amniocentesis and diagnostic test) even if it means taking the 1 in 200 chance of pregnancy loss?
- If I find out that my baby has Down syndrome or trisomy 18, what will I do with this information? Will I end the pregnancy? Will I continue the pregnancy? Will I use the information to prepare for a child with special needs? Will I make an adoption plan for the baby?
- Will I feel too worried for the rest of my pregnancy if I do not have the amniocentesis or NIPT?

What if the result of the amniocentesis or detailed ultrasound shows that the baby has one of these conditions?

Your health care provider, as well as medical geneticists or genetic counsellors, are there to discuss your choices with you and to help you make a decision that is right for you.

Please take this home to read, think about and talk over with your partner and family. If you have more questions, or feel a genetic counselling appointment to talk about your options would be helpful, talk to your health care provider.