

A Screen Positive Result:

What does it mean and what do I do now?

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 diagnostic testing is offered. This type of testing tells you for sure 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 a test that tells you if the baby has either of these conditions for sure. This test is called an amniocentesis.

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 or another chromosome condition.

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.

The amniocentesis result is ready in 2 to 3 weeks.

Should I have an amniocentesis?

It is your choice. Not every woman who has a screen positive result wants to have an amniocentesis. 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 need to know for sure if my baby has Down syndrome or trisomy 18 before the baby is born?
- 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 put the baby up for adoption?
- Most women who have the amniocentesis do not lose their pregnancy, but do I think the 1 in 200 chance of pregnancy loss is too high for me to take?
- Will I feel too worried for the rest of my pregnancy if I do not have the amniocentesis?
- What would be more difficult for me? If I have the amniocentesis and lose the pregnancy or if I don’t have the amniocentesis and find out when my baby is born that the baby has Down syndrome or trisomy 18?

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.

www.bcprenatalscreening.ca