



# Genetic Screening

## Chorionic Villus Sampling

### What is the chorionic villus?

The term chorionic villus refers to a specific section of the developing placenta.

### What is chorionic villus sampling?

Similar to an amniocentesis, chorionic villus sampling (CVS) involves having a long needle inserted into the uterus, withdrawing a minute piece of the developing placenta. This is done under local anesthetic, with the aid of ultrasound to minimize risk to the baby. Some women find it painless, while others feel discomfort or pain. If your blood type is Rh negative, you will then be given a WinRho shot.

The withdrawn tissue contains cells genetically identical to the baby. These are analyzed for their chromosomal makeup. You can expect results within 1-3 weeks.

CVS can be done anytime between 10 and 13 weeks of pregnancy.

### Who is eligible for CVS?

There is a risk of pregnancy loss with this procedure of 1 in 200. Because of this, it is only offered to those whose risk of diagnosis of a fetal abnormality is greater than the risk of inducing an abortion. This includes:

- Women over 40 (carrying one baby)
- Women over 35 (carrying more than one baby)
- Women who have a personal or family risk of chromosomal abnormalities

### What can CVS tell me?

The CVS results will include a written report on how many chromosomes, as well as what sex chromosomes, your baby has. Note: if you do *not* wish to know the sex of your baby make sure to remind your caregiver when s/he gives you the results.

In British Columbia, a standard CVS procedure not only counts the number and type of chromosomes, but also exams genes inside the chromosomes for approximately 40 common genetically inherited abnormalities. It is still possible that your baby may have a condition not tested for.

At 99% accurate, CVS is considered extremely precise for the conditions it tests for, but it is not foolproof.

CVS also cannot give any information about how severe a genetically inherited condition might be. For example, with Down's syndrome there is a variance from highly functional both mentally and physically, to total incompatibility with life.

### What are the risks of CVS?

- There is a 1/100 to 1/200 risk of CVS causing a miscarriage.
- Even with ultrasound used during the procedure, there is still a 1 in 3000 chance of damage to the baby.



- Some women also experience cramping and/or bleeding after the procedure. These symptoms may range from mild to severe. Because of this, it is a good idea to take the day off work and plan to rest after the procedure.
- This procedure carries a 0.1% chance of uterine infection.
- In about 3% of cases, the CVS needs to be repeated due to culture failure.

### Are there any alternatives?

After 15 weeks of pregnancy, you may be eligible for an amniocentesis. This is a similar procedure that takes a sample of amniotic fluid instead of the placenta.

Between 15 and 20 weeks of pregnancy, you can choose to have a Quadruple Marker Screen which is a blood test that screens for Trisomy 21 (Down's syndrome), Trisomy 18 (Edward's syndrome) and open neural tube defects (spina bifida, anencephaly). It does not carry a risk of pregnancy loss, but has a high false positive rate and cannot provide an absolute diagnosis without further testing, i.e. amniocentesis.

If you are between 10 and 14 weeks, you are eligible for the Serum Integrated Screen or SIPS (two blood tests, one each in the first and second trimesters).

Depending on your age and any risk factors you may have, you may also be eligible for the Integrated Pregnancy Screen or IPS (the same two blood tests plus a first trimester ultrasound). Both the SIPS and IPS tests screen for the same things as the Quadruple Marker Screen, but do so with more accuracy and fewer false positives.

All women are offered a routine ultrasound at 19 weeks of pregnancy. Ultrasound is used to diagnose major congenital abnormalities, environmental or genetic, including serious neural tube defects. It can also pick up indicators or "markers" of genetically inherited conditions such as Trisomy 18 or Trisomy 21, but it cannot provide absolute diagnoses of these conditions.

*Please refer to the website of the BC Prenatal Genetic Screening Program for more details, including age-related risks, detection rates and false positive incidence:*

[www.bcprenatalscreening.ca](http://www.bcprenatalscreening.ca)