What does a normal CVS or amniocentesis result mean?

Most commonly, chorionic villous or amniotic fluid sample are used for examination of the karyotype. A normal result in this case means the fetus has normal chromosomal constitution, thereby excluding Down's syndrome, Edward's syndrome, Patau's syndrome and all other chromosomal aberration. However, the samples are not usually examined for all other genetic defects unless there is such an indication and prior arrangement has been made. A normal result therefore also cannot exclude all possible genetic defects. A normal karyotype cannot also exclude structural malformations or other anomalies that are not related to chromosomal constitution.

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Knowing Chorionic Villous Sampling and Amniocentesis

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What is chorionic villous sampling (CVS)?

Chorionic villous sampling involves getting a small sample of the chorionic villous (placental tissue) (Figure 1). The procedure is usually performed between 11-14 weeks of gestation.

Figure 1:
Chorionic villous sampling

What is involved in the procedure of CVS?

Before the procedure, an ultrasound examination is performed to locate the site of the placenta. Then local anaesthetic is injected to the mother's tummy. After the medication has taken effect, a fine needle is passed under ultrasound guidance through the mother's tummy and her uterus to the placenta, where a small sample of tissue is obtained by suction. The sampling process takes around 1 minute.

What is amniocentesis?

Amniocentesis involves withdrawing a small sample of amniotic fluid from the mother's uterus (Figure 2). Amniotic fluid is the watery fluid that surrounds the fetus and it contains cells that the fetus has shed. The procedure is usually performed between 16-20 weeks of gestation.

Figure 2:
Amniocentesis

What is involved in the procedure of amniocentesis?

Before the procedure, an ultrasound examination is performed to locate the best site for sampling. During the procedure, a fine needle is passed through the mother's tummy into the amniotic fluid cavity and a small amount of the fluid is withdrawn. The sampling process takes around 1 minute.

Are there any risks associated with CVS or amniocentesis?

Both are safe procedures in general although they carry a small miscarriage rate of 0.5-1% even under experienced hands. Other complications such as wound infection, uterine infection, haemorrhage, amniotic fluid leakage and failed procedure are very rare.

What are the indications for CVS or amniocentesis?

Chorionic villous and amniotic fluid samples can be used for examination of the fetal karyotype (the chromosomal constitution), or for diagnosis of certain genetic conditions. Those pregnancies with increased risks for chromosomal aberration or genetic disease are candidates for consideration of either procedure. One common indication is an abnormal screening test for Down's syndrome.

How is CVS compared to amniocentesis?

The advantage of CVS is that it can be performed earlier than amniocentesis and hence any prenatal diagnosis can be made earlier in pregnancy. It is controversial whether amniocentesis carries a slightly lower risk of miscarriage compared to CVS. In any case, the risks of miscarriage are low. The accuracy of both procedures is also comparable. There are conditions where the location of the placenta will make CVS procedure very difficult, in which case an amniocentesis would be a better alternative. The choice of any procedure can be made after discussion with your obstetrician.