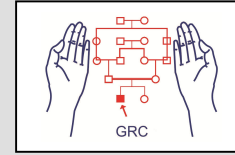


# Fetal Sampling

Genetics Resource Centre (GRC)



Getting sample from the fetus is an essential requirement for prenatal diagnosis. The two most common methods of fetal sampling include Chorionic Villus Sampling (CVS) and Amniocentesis.

## Chorionic Villus Sampling (CVS)

CVS is done under ultra-sound guidance either by the trans-abdominal or the trans-cervical route. At GRC and the other places in Pakistan the trans-abdominal approach is used. The CVS needle is inserted through the abdomen into the placenta under ultrasound guidance and the placental tissue is aspirated. The whole procedure takes about 10-15 minutes and the mothers can go home after the test.

## Amniocentesis

Amniocentesis is usually done after 15 weeks gestation. The aspirated fluid is used for culture of amniotic cells or extraction of DNA. However, the yield of DNA is low. It may be useful for cytogenetic analysis. The sharp needle used in amniocentesis may injure the fetus if the operator is not careful. The pregnancy loss in amniocentesis is around 1-2%.

### When is the right time for CVS?

The CVS can be done at any time after 10 weeks gestation. The best time to do the CVS is around 12 weeks. However, in very obese women and those with very low lying posterior placenta it may be advisable to delay the sampling till 13 or 14 weeks. Keeping in view the fatwa on termination of pregnancy we discourage any type of fetal sampling after 16 weeks. The procedure must not be attempted before 9<sup>th</sup> weeks as it can result in fetal malformations.

### Does the patient need any preparation?

No specific preparation is required. The patients who come empty stomach are more likely to develop dizziness or fainting. Full bladder is often required for visualizing uterus in the early gestation period. If the patient is taking Aspirin then the drug must be stopped at least 72 hour prior to the procedure.

### In what type of diseases CVS can be useful?

CVS is only a method for fetal sampling. It should be done only with a specific indication in mind. The indication must be ascertained before requesting CVS. It is commonly used for prenatal diagnosis of single gene disorders like thalassaemia, cystic fibrosis, Duchenne Muscular Dystrophy etc. It can also be used for broader spectrum genetic testing like cytogenetics. Since cytogenetic testing takes longer time, these days many labs use quick PCR based testing of trisomies like 13, 18 and 21.

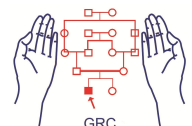
### What are the common complications of CVS?

CVS is a safe technique for the mother as well as the fetus. The women often complain of pain due to uterine cramps. The patient may be given paracetamol for pain relief. Infection or miscarriage is the most feared complication that may require hospitalization. In our experience of over 3500 procedures the miscarriage rate is <2%. In suspected Rh-D feto-maternal incompatibility Anti-D prophylaxis (250ug i/m within 72 hours of the procedure) should be given.

### Does the patient need prophylactic antibiotics?

Prophylactic antibiotics after the procedure are usually not required.

### Can the patient travel long-distance after the CVS?



The patients are usually advised to have bed rest with restriction of strenuous activities. Since many patients come from out-station they are usually curious to know about the effect of travelling. Journey on a bumpy road can be hazardous and should be avoided as far as is possible.

**What is the better choice, CVS or Amniocentesis?**

CVS is the best choice for fetal sampling because it can be done as early as 10 weeks whereas amniocentesis is usually done after 15 weeks gestation. Early amniocentesis is unlikely to yield enough fluid (fetal cells) for DNA testing. Amniocentesis is more often used for cell culture and cytogenetic analysis. We do amniocentesis in patients who have very low lying posterior placenta that is not accessible through the abdominal route.

**How fetal sampling is done in a twin pregnancy?**

We need to take two separate samples in non-identical (di-zygotic) twins. If the twins are confirmed to be identical (mono-zygotic) then one sample is considered enough.

**What can be done in discordant twins?**

Occasionally in a twin pregnancy one twin is affected while the other is not. In such cases selective feticide of the affected twin can be done by intra-cardiac injection of KCl.

