Review Article

USG Guided Chorionic Villus Sampling

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Abstract

Treatment of most genetic disorders is either not possible or is very expensive. The current trend is to prevent the birth of children with genetic disorders. The ability to take sample from the fetus in early pregnancy has opened up new vistas in the management of genetic disorders. In this sense, Chorionic Villus Sampling (CVS) can play a very crucial and pivotal role to determine chromosomal or genetic disorders in the fetus. CVS entails getting a sample of the chorionic villus (placental tissue) and testing it. It can be done 10-12 weeks after the last menstrual period (LMP). It is the preferred technique before 15 weeks. Among the approaches for fetus sampling, CVS is capable to provide the earliest and reliable form of prenatal diagnosis of fetal abnormalities with high level of accuracy (98-99%). It is the most popular and efficient procedure of fetus sampling, because it can be done earlier in pregnancy. Results of CVS sampling can be available sooner than amniocentesis results. This allows to know the health of the baby and make an earlier decision whether to continue or terminate the pregnancy. In this way, CVS helps to eradicate the curse of genetically disordered babies and thereby, paves the way to build up a healthy society.

Key words: Chorionic Villus Sampling (CVS), Chorionic Villus, Amniocentesis.

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Introduction:

Chorionic villus sampling is the removal of a small piece of placental tissue from the uterus during early pregnancy to screen the baby for genetic defects. The chorionic villi are the tiny units that make up the placenta and have the same chromosomes as the fetus. CVS is usually done 10 to 12 weeks after the last menstrual period, earlier than amniocentesis (usually done at 15 to 20 weeks). CVS is ideal for DNA, cytogenetic and enzyme analysis. DNA-based diagnoses of Mendelian conditions, such as cystic fibrosis, hemophilia, muscular dystrophy and hemoglobinopathies can be made by direct analysis of uncultured chorionic villus cells (a more efficient method than culturing amniocytes).[1-4]

CVS can be done through the cervix (transcervical) or through the abdomen (transabdominal). The techniques are equally safe when done by a provider with experience, although miscarriage rates are slightly higher when done through cervix. The health care provider will use ultrasound to choose the safest approach and as a guide during sampling.

An abdominal ultrasound is performed to determine the position of the uterus, the size of the gestational sac and the position of the placenta within the uterus.

The transcervical procedure is performed by inserting a thin plastic tube through the vagina and cervix to reach the placenta. Ultrasound is used to guide the tube into the appropriate area and remove a small sample of chorionic villus tissue.

The transabdominal procedure is performed by inserting a needle through the abdomen and uterus and into the placenta. Ultrasound is used to help guide the needle and a small amount of tissue is drawn into the syringe. The sample is placed in a dish and evaluated in a laboratory.[5-7]

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Decision to terminate pregnancy is never taken lightly in any society. Though several reports suggest reservations among Muslim patients in terminating a pregnancy for a genetic disorder, in Muslim countries like Pakistan, CVS is being performed successfully. A religious verdict (Fatwa) on the subject from a known religious scholar in Pakistan, says that Islam permits termination of pregnancy for a genetic disorder, if the disorder for which termination is being sought is of serious nature and the termination is done before 120 days (17 weeks) gestation. In Pakistan, prenatal diagnosis for thalassaemia was introduced in May 1994. Since then, over 5000 CVS procedures have been done at various centers. The experience has shown that over 90% of the couples who request prenatal diagnosis also terminate the pregnancy when it is required.[8]

History:

For the first time, CVS was tested by Italian Biologist Giuseppe Simoni, scientific director of Biocell Center, in 1983. CVS had become widely used, worldwide by the early 1980s. The World Healh Organization (WHO) sponsors an International Registry of CVS procedures. Data in the International Registry probably represent less than half of all procedures performed worldwide.[9,10]

Indications:

Possible reasons for having a CVS can include:

- Patients are known carriers of a genetic disorder like thalassaemia.
- Mother's age, 35 years or more.

  (In the United States, an estimated 40% of pregnant women more than or equal to 35 years of age underwent either amniocentesis or CVS in 1990).
- Abnormal first trimester screen results.
- Increased nuchal translucency or other abnormal ultrasound findings.
- Family history of a chromosomal abnormality or other genetic disorder such as, Down's syndrome, Haemophilia, Sickle cell disease, Tay Sachs disease, Cystic fibrosis.
- Previous pregnancy affected by a genetic abnormality.[11-14]

Contraindications:

CVS is not recommended for women who:

- Have an active infection (i.e. STD)
- Are carrying twins
- Have experienced vaginal bleeding during pregnancy.

Transcervical CVS is not recommended for women who:

- Have uterine fibroids
- Have a tilted uterus which impedes the catheter.

Technique: (Transabdominal approach):

- At the time of booking, ultrasound is done to ascertain the gestation, position of placenta and the number of fetuses. The right time for sampling is decided.
- The procedure is done under ultrasound guidance by using 3.5 MHZ convex probe. The key to successful sampling lies in selecting the right spot for entry. Anterior placenta is easier to approach.
- A suitable site for introducing the needle on the anterior abdominal wall is selected. Abdominal skin is cleaned with betadine. About 5-10 ml of 2% xylocain is injected. The whole tract of the CVS needle from the skin to the uterine serosa is infiltrated with xylocain.
- The aspiration can be done with a simple spinal needle or with a special co-axial needle set (Rocket Medical, Uk, http:// www.rocket medical.com) The co-axial needle set consists of an outer 17 or 18G guide needle, 16.5 cm in length and an inner 20 cm 19 or 20G aspiration needle. The needle sets can be reused after sterilization and sealing in a sterile pack.
- The CVS outer needle is introduced through the puncture site used for the local anesthetic. The USG probe is held in the left hand and the CVS needle is maneuvered with the right hand. The needle tip should remain visible at all times. Care is required to avoid entering the peritoneal cavity.
- After piercing the uterine wall, the needle is pushed with a jerky movement to enter the placenta in its longitudinal plane. The entry of needle into the placenta is marked by loss of resistance. A 50 ml disposable syringe is attached to the CVS inner needle and it is rinsed with sterile normal saline. The stillet of the outer needle is removed and the inner needle is introduced through the outer needle.
- Once the inner needle is in place, the plunger of the syringe is pulled back to create suction force. The position is maintained by locking the plunger with the four fingers of the right hand.

- The aspiration syringe and the inner needle in the locked position are jiggled to and fro a few times to cause localized disruption of the placenta.
- The disrupted villi are sucked into the needle. The sample is flushed into a sterile Petri dish containing normal saline. Adequate amount of grayish white placental villi confirm a successful aspiration.
- Post aspiration USG scan is done to see the fetal well being and formation of placental haematoma.[14-17]

Problems:

(1) Sampling may be difficult in very obese patients. In such patients it is better to delay the sampling till 13-14 weeks when the placenta is clearly visible.

(2) Flabby abdomen with very lax skin may also pose difficulty. Previous C-section scars are often very difficult to pierce. Effort should be made to choose an area of skin away from the scar.

(3) Occasionally the needle may enter a placental lake resulting in blood tap from the needle when the stillet is removed. In such cases, aspiration should be done quickly before the blood clots in the needle.

Follow-up:

1. The patient should be kept under observation for 30 to 60 minutes after the procedure.
2. The patients are usually advised to restrict movements like climbing stairs, traveling on a bumby road etc. Bed rest may be advised for those who develop placental haematoma.
3. Some degree of pain or discomfort usually persists for a couple of days. Mild analgesics may be prescribed as a routine.
4. All Rh negative mothers undergoing CVS should be given 250 mg anti-D prophylaxis.[18]

Side effects and risks:

Although CVS is considered to be a safe procedure, it is recognized as an invasive diagnostic test that has some temporary side effects and does pose some potential risks. Patient may have mild pain or cramps, like period pain, and spotting (light bleeding form the vagina) for a few hours afterwards. Studies show that the CVS-related miscarriage rate is approximately 0.5%-1.0% (1/200-1/100). Apart from risk of miscarriage, there is a risk of amniotic fluid leakage. The resulting amniotic fluid leak can develop oligohydramnios. Infection has occurred rarely after the procedure. In one study, no episodes of septic shock were reported after 4,200 CVS procedures. Over all, infection rates have been 0.1% after either CVS or amniocentesis. Patient can also develop placental haematoma. There is a risk of CVS causing limb/digit reduction defects in the fetus if performed before 10 weeks (0.07%-0.10%). More recent studies provide reassurance that there is no increased risk to limb defects when CVS is performed at 10 weeks or later. Additionally, if the mother has HIV, there is an increased risk of transmission of the virus to the baby during CVS.[15, 19-28]

Limitations:

A small percentage (1-2%) of pregnancies will have confined placental mosaicism, where some but not all of the placental cells tested in the CVS will be abnormal, even though the pregnancy is unaffected. Cells from the mother can be mixed with the placental cells obtained from the CVS procedure. Occasionally, if these maternal cells are not completely separated from the placental sample, this can lead to discrepancies with the results. This phenomenon is called Maternal Cell Contamination (MCC).[28] CVS does not detect neural tube defects.[5]

Conclusion:

Genetically disordered babies put up tremendous pressure on the family and the society as a whole. Being a member of third world country, we are not yet capable of taking proper care of these babies. In such a situation, CVS can play a vital role to alleviate the severity of this problem. But disappointingly, CVS has not yet been introduced in our country. Recently, a workshop on CVS was organized by Dhaka Shishu Hospital Thalassaemia Center and Obstetrics and Gynecology Department of Bangladesh Medical College Hospital on 20th March, 2010, where CVS on 4 cases was performed successfully in Radiology and Imaging Department. It is a matter of great regret, that laboratory facilities for CVS are also very poor and meager in our country. Only BSMMU(Bangabandhu Sheikh Mujib Medical University) with very limited scope is providing the services. So, Government and private initiatives are urgently required towards creating awareness about CVS, motivating the people and to generate trained and skilled doctors with proper technological expertise. We are looking forward towards expeditious and effective implementation of CVS system in Bangladesh that would, in turn, usher in new hope for the concerned people.

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