Sem – VI (UG)

CC-13: Developmental Biology

C13T: Unit -5, Implications of Developmental Biology

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Amniocentesis

What is Amniocentesis? (Amniotic Fluid Test)

Medical procedure used for prenatal genetic testing for chromosomal abnormalities (presence or absence of certain chromosomes, genes or enzymes), fetal infections as well as for sex determination to obtain a sample of amniotic fluid from a pregnant woman. A long sterile needle is inserted through the abdominal wall into the amniotic sac to obtain the fluid.

Amniotic fluid—the substance that fills the amniotic sac and surrounds the developing fetus—contains fetal cells (e. g. multipotent mesenchymal, hematopoietic, neural, epithelial, and endothelial stem cells) that can be used for genetic testing.

Why is an Amniocentesis performed?

This process is performed to look for certain types of birth defects. Because amniocentesis presents a small risk for both mother and her baby, the prenatal test is generally offered to women who have a significant risk for genetic diseases, including those who:

- Have an abnormal ultrasound or abnormal lab screens
- Have a family history of certain birth defects

- Have previously had a child or pregnancy with a birth defect
- Had an abnormal genetic test result in the current pregnancy

Amniocentesis does not detect all birth defects, but it can be used to detect the following conditions if the parents have a significant genetic risk of:

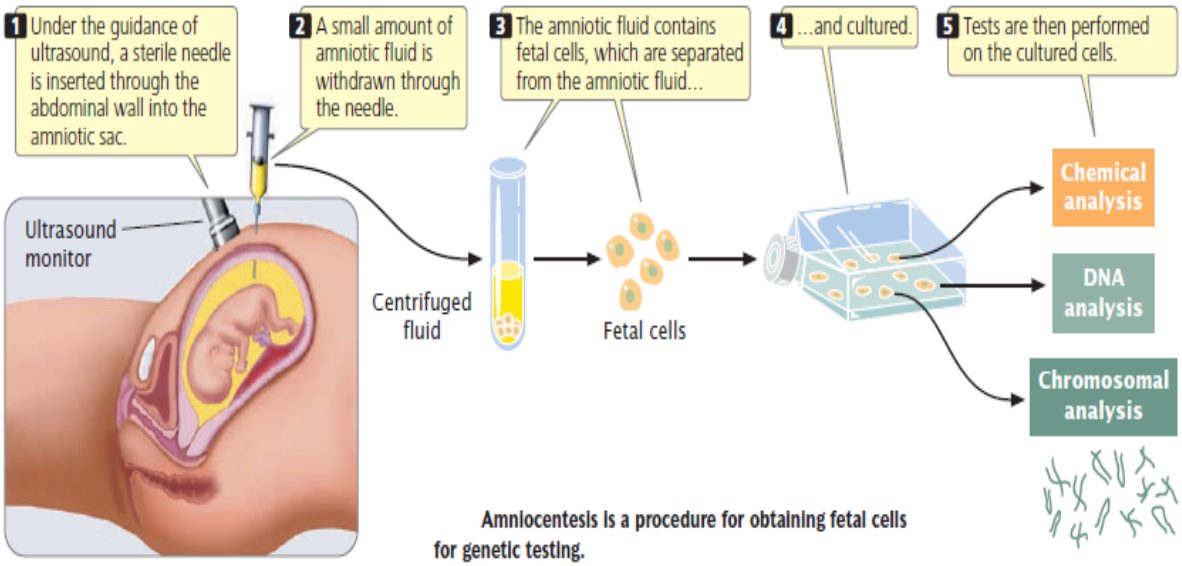
- Down syndrome
- Sickle cell disease
- Cystic fibrosis
- Muscular dystrophy
- Tay-Sachs disease

Procedure:

Amniocentesis is routinely performed as an outpatient procedure either with or without the use of a local anaesthetic.

- Ultrasonography is used to locate the position and movements of the fetus, location of placenta, and characteristics of the amniotic fluid in the uterus.
- With the aid of ultrasound guidance, a long, sterile needle is inserted through the abdominal wall of the uterus at an angle through the muscle, then through the wall of uterus and finally into the amniotic sac.
- The physician then punctures the sac in an area away from the fetus and extracts approximately 20ml of amniotic fluid. This procedure can be performed with a single needle and double needle technique. These techniques have their own variations in how they are performed including guidance of needle insertion location, and angle of needle insertion.
- From the 20ml of amniotic fluid, the first 2ml is typically discarded due to mixture with maternal blood cells to ensure high quality fluid sampling.

- Fetal cells are separated from the amniotic fluid and placed in a culture medium that stimulates them to grow and divide, then fixed and stained. Under a microscope, the chromosomes are examined for abnormalities.
- After the procedure, the puncture seals and the amniotic sac replenishes the liquid over the next 24-48 hours.



Amniocentesis is a procedure for obtaining fetal cells for genetic testing.

Medical uses:

1) Genetic diagnosis:

Early in pregnancy, amniocentesis is used for diagnosis of chromosomal and other fetal problems such as:

- Down syndrome (Trisomy 21)
- Patau syndrome (Trisomy 13)
- Edwards' syndrome (Trisomy 18)
- Sex chromosome aneuploidies
- Neural tube defects (diseases where the brain and spinal column don't develop properly) – anencephaly (a baby born with an underdeveloped brain and an incomplete skull) and spina bifida/split spine (a birth defect in which there is incomplete closing of the spine

and the membranes around the spinal cord during early development), by alpha-fetoprotein levels

- Rare metabolic disorders

2) ***Infection:***

This process can detect infections via decreased glucose level, a Gram stain showing bacteria, or abnormal differential count of WBCs.

3) Lung maturity:

This can predict fetal lung maturity, which is inversely correlated to the risk of infant respiratory distress syndrome. Several tests are available, including the-

- Lecithin-sphingomyelin ratio (L/S ratio): if the result is less than 2:1, the fetal lungs may be surfactant deficient.
- The presence of phosphatidylglycerol (PG): indicates fetal lung maturity.
- The surfactant/albumin ratio (S/A ratio): the result is given as mg of surfactant per gm of protein. An S/A ratio <35 indicates immature lungs, 35-55 is intermediate, and >55 is mature surfactant production.

Note: Lungs require surfactant, a soap-like substance, to lower the surface pressure of the alveoli in the lungs. This is especially important for premature babies trying to expand their lungs after birth. Surfactant is a mixture of lipids, proteins, and glycoproteins; lecithin and sphingomyelin being two of them. Lecithin makes the surfactant mixture more effective.

Higher amounts of lecithin – in reference to albumin – is indicative of lung maturity (and thus survival of the neonate).

4) Decompression of polyhydramnios:

Polyhydramnios (the accumulation of amniotic fluids) can be relieved via decompression amniocentesis. Amniocentesis

can also be used to diagnose potential causes of polyhydramnios.

5) ***Rh incompatibility:***

This process can be used to diagnose Rh incompatibility, a condition when the mother has Rh-negative blood and the fetus has Rh-positive blood. Early detection is important to treat the mother with Rh immunoglobulin and to treat her baby for haemolytic anemia.

Risks:

Amniocentesis is performed between the 15th and 20th week of pregnancy; performing this test earlier may result in fetal injury. The term "early amniocentesis" is sometimes used to describe use of the process between weeks 11 and 13.

Complications of amniocentesis include preterm labor and delivery, respiratory distress, postural deformities, chorioamnionitis or intra-amniotic infection (an inflammation of the fetal membranes – amnion and chorion, due to a bacterial infection), fetal trauma and alloimmunisation or rhesus disease (an immune response to foreign antigens after exposure to genetically different cells or tissues) of the mother. Amniotic fluid embolism or AFE (a very uncommon childbirth emergency in which amniotic fluid enters the blood stream of the mother to trigger a serious reaction and results in cardiorespiratory collapse and massive bleeding) has also been described as a possible outcome. Additional risks include amniotic fluid leakage and bleeding. These two are of particular importance because they can lead to spontaneous abortion in pregnant patient.

- *The first amniotic stem cells bank in the US is active in Boston, Massachusetts.*