MEDICAL ACADEMY NAMED AFTER S.I. GEORGIEVSKY OF VERNADSKY CFU

NAME –PAWAN

GROUP ;LA1-202(2)

TOPIC; PRENATAL DIAGNOSIS.MEDICAL GENETIC COUNSELING.

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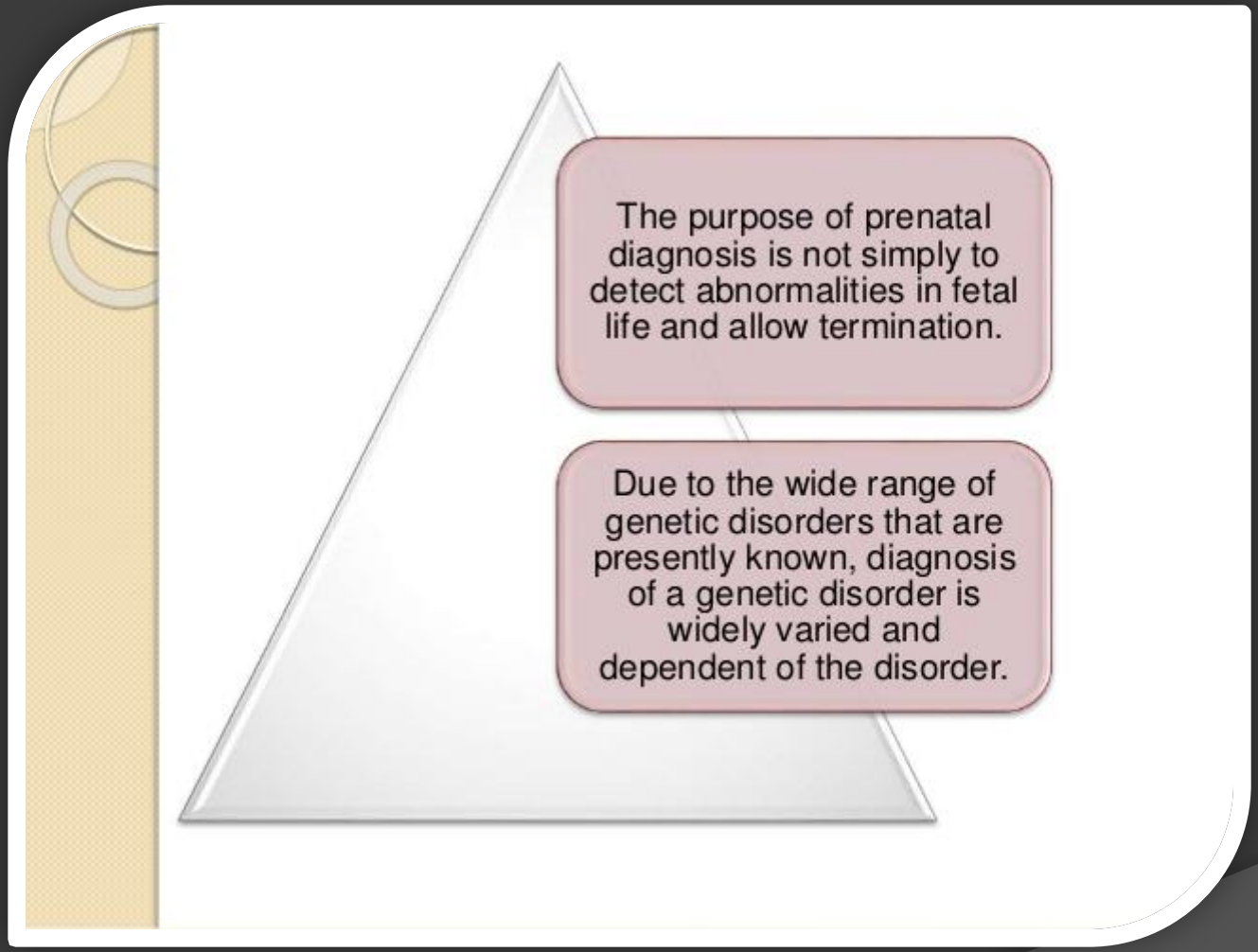
GENETIC DISORDERS

A **genetic disorder** is a genetic problem caused by one or more abnormalities in the genome, especially a condition that is present from birth (congenital). Most genetic disorders are quite rare and affect one person in every several thousands or millions.


The purpose of prenatal diagnosis is not simply to detect abnormalities in fetal life and allow termination.
Due to the wide range of genetic disorders that are presently known, diagnosis of a genetic disorder is widely varied and dependent of the disorder.

PURPOSE OF DIAGNOSIS

- Provide a range of informed choice to the couples at risk of having a child with abnormality
- Provide reassurance and reduce anxiety, especially among high-riskgroups •Allow couples at high risk to know that the presence or absence of the disorder could be confirmed by testing
- Allow the couples the option of appropriate management(psychological,pregnancy/delivery, postnatal)
- To enable prenatal treatment of the affect

INDICATIONS FOR PRENATAL DIAGNOSIS

- Advanced maternal age
- Previous child with a chromosome abnormality
- Family history of chromosome abnormality
- Family history of singe gene disorder
- Abnormalities identified in pregnency

METHODS OF PRENATAL DIAGNOSIS

NON INVASIVE TECHNIQUES

- Fetal visualization
  1. ULTRASONOGRAPHY
  2. FETAL ECHOCARDIOGRAPHY
  3. MAGNETIC RESONANCE IMAGING (MRI)
- Maternal serum screening
- Separation of fetal cells from the mother's blood

INVASIVE TECHNIQUES

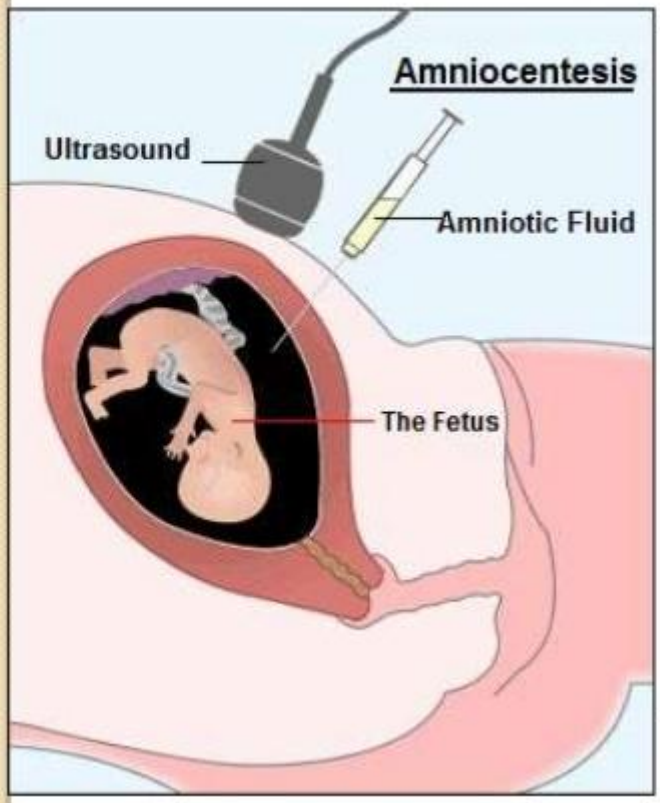
- Fetal visualization
- Fetal tissue sampling
- Cytogenetics
- Molecular genetics

AMNIOCENTESIS

Amniocentesis is an invasive, diagnostic antenatal test. It involves taking a sample of amniotic fluid in order to examine fetal cells found in this fluid.

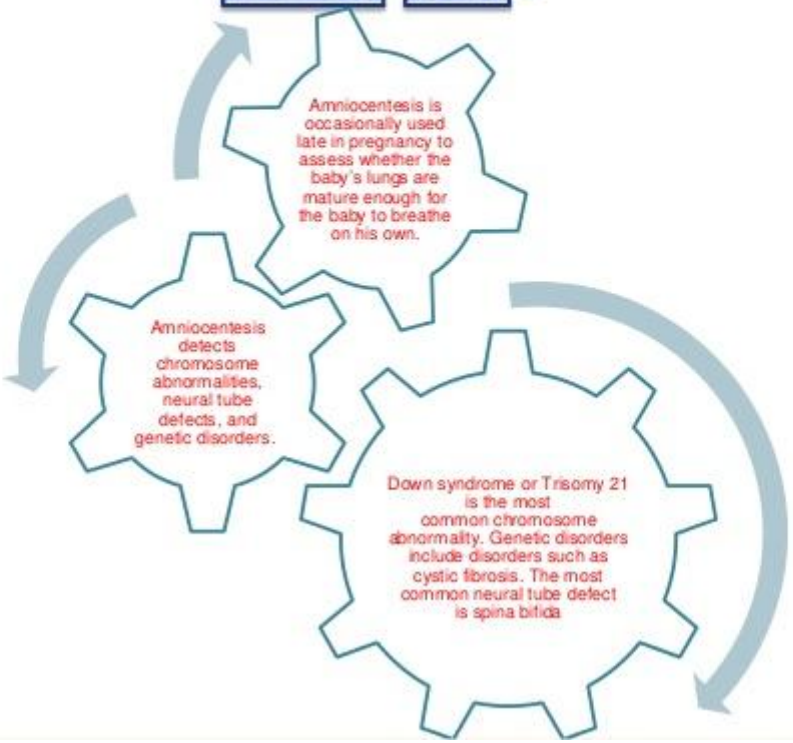
Because it carries a slightly increased risk of miscarriage amniocentesis is usually reserved for those women considered at higher risk of carrying a fetus with a chromosomal abnormality.

How Is Amniocentesis Performed?



An ultrasound is used as a guide to determine a safe location for the needle to enter the amniotic sac, so the fluid may be safely removed. A sample of amniotic fluid is collected through the needle. The procedure takes about 45 minutes, although the collection of fluid takes less than five minutes.

What does amniocentesis look for?



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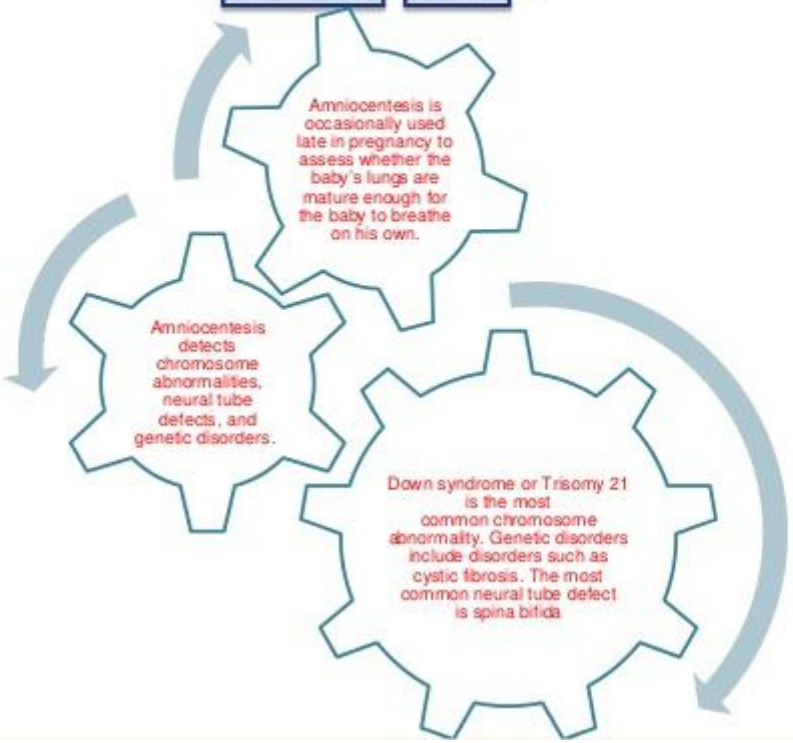


CHRONIONIC VILLUS SAMPLING

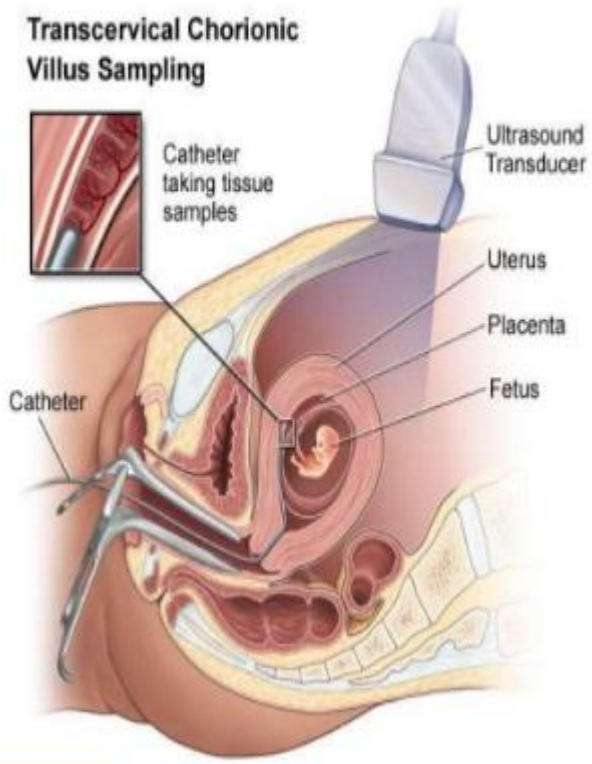
It enables diagnosis in first trimester(10-11 week of gest.)under ultrasound guidance by transcervical or transabdominal aspiration of chorionic villi

•These are fetal cells from the outer layer of trophoblast.

•Results can be obtained in one to three days, so a diagnosis in first trimester in addition that villi provide are rich source of DNA

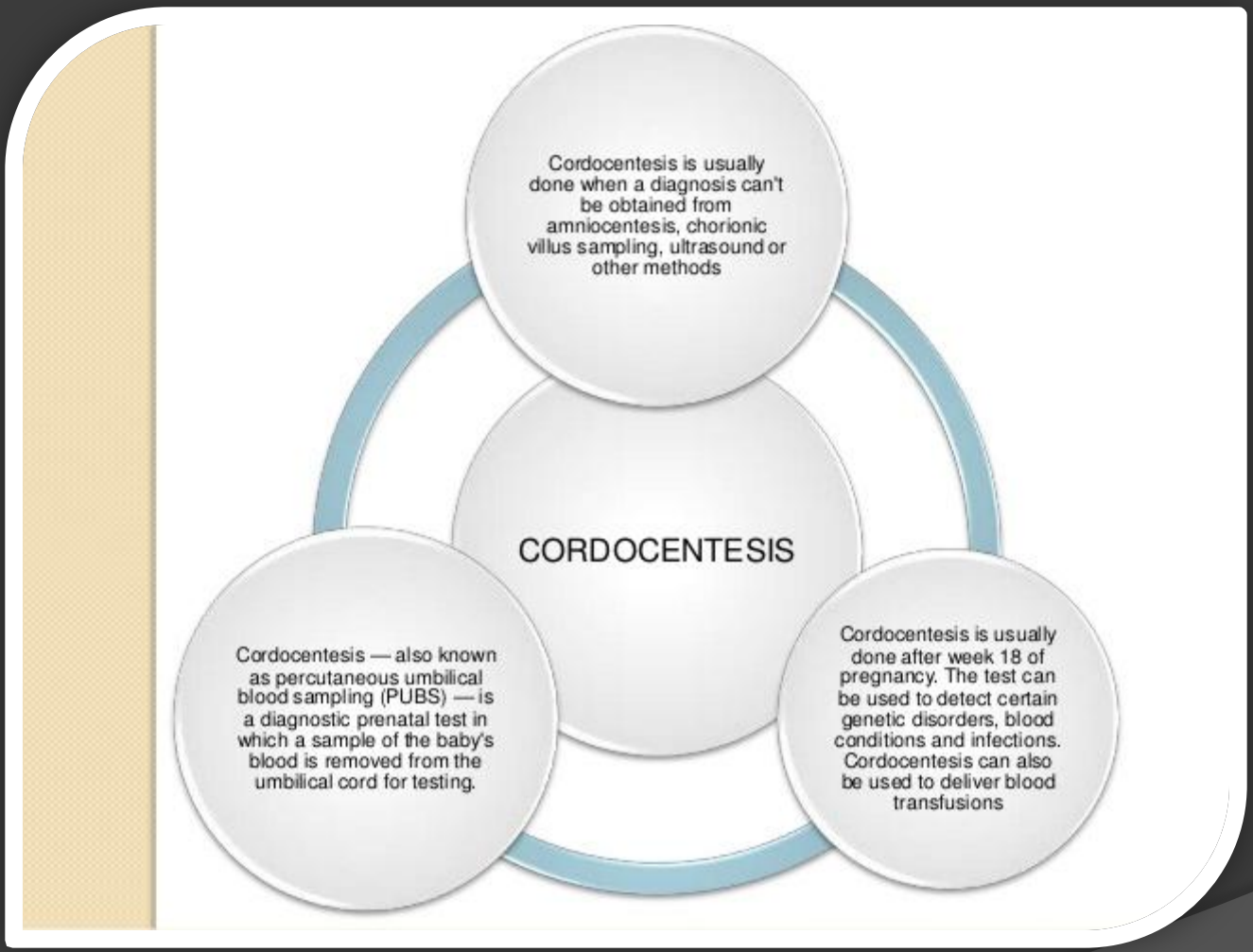
•Disadvantage is in higher risk of abortion(2-3%)and limb abnormalities if carried before the 9 weeks of gestation

HOW IS CVS PERFORMED?

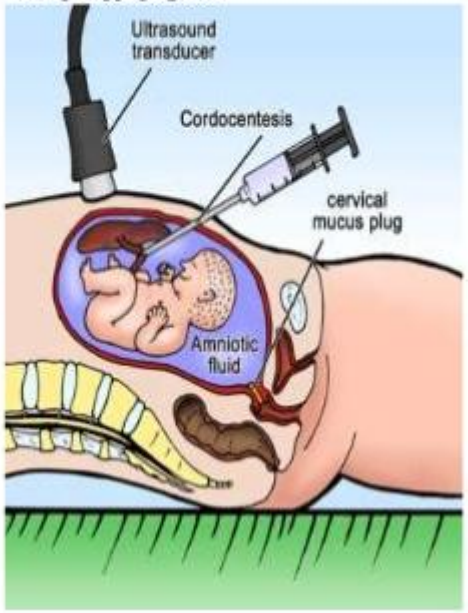


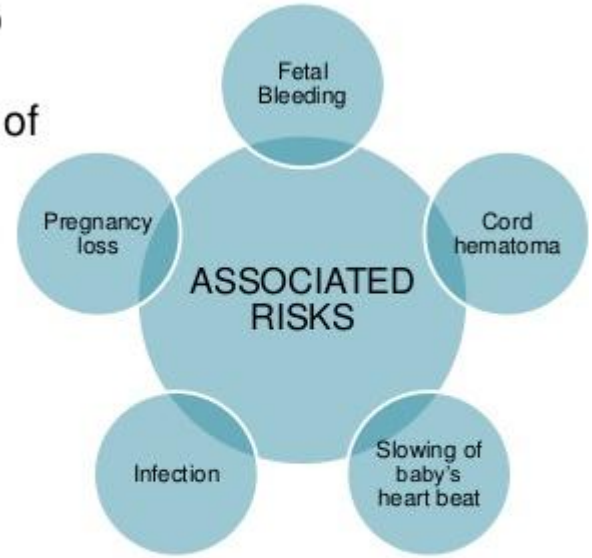
CVS is usually carried out between the 11th and 14th weeks of pregnancy, although it's sometimes performed later than this if necessary.

transabdominal CVS – a needle is inserted through your tummy (this is the most common method used)
transcervical CVS – a tube or small forceps (smooth metal instruments that look like tongs) are inserted through the cervix (the neck of the womb)


Cordocentesis is usually done when a diagnosis can't be obtained from amniocentesis, chorionic villus sampling, ultrasound or other methods
CORDOCENTESIS
Cordocentesis — also known as percutaneous umbilical blood sampling (PUBS) — is a diagnostic prenatal test in which a sample of the baby's blood is removed from the umbilical cord for testing.
Cordocentesis is usually done after week 18 of pregnancy. The test can be used to detect certain genetic disorders, blood conditions and infections. Cordocentesis can also be used to deliver blood transfusions

It is usually used in the management of Rhesus iso immunization and in some cases to solve the problem of mozaicism





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FETOSCOPY

Fetoscopy is an endoscopic procedure during pregnancy to allow access to the fetus, the amniotic cavity, the umbilical cord, and the fetal side of the placenta. A small (3–4 mm) incision is made in the abdomen, and an endoscope is inserted through the abdominal wall and uterus into the amniotic cavity. Fetoscopy allows medical interventions such as a biopsy or a laser occlusion of abnormal blood vessels or the treatment of spina bifida


When Perform?
18 -20 weeks
Fetoscopy
-IU surgery
-TTT syndrome
-Icthyosis

THANK YOU