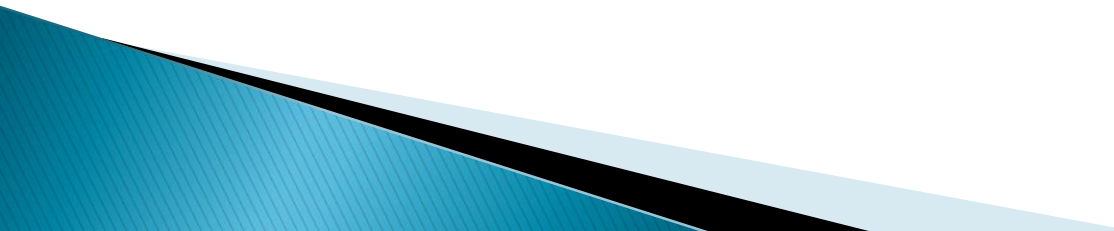


# Human Health & Sex Education

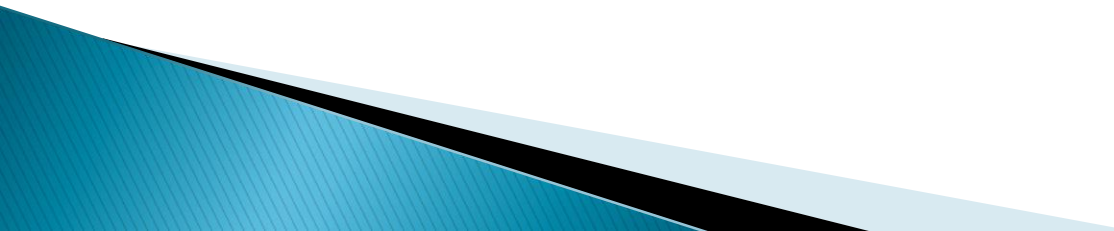
Open Course – V Semester

Dr. Swapana Johny  
Asst. Professor  
Dept of Zoology

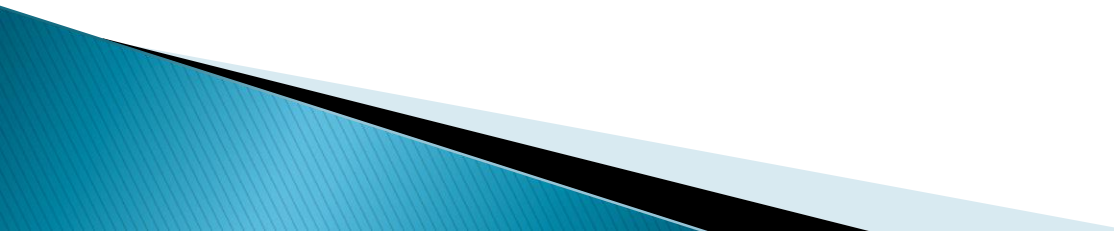
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# PRE-NATAL DIAGNOSIS

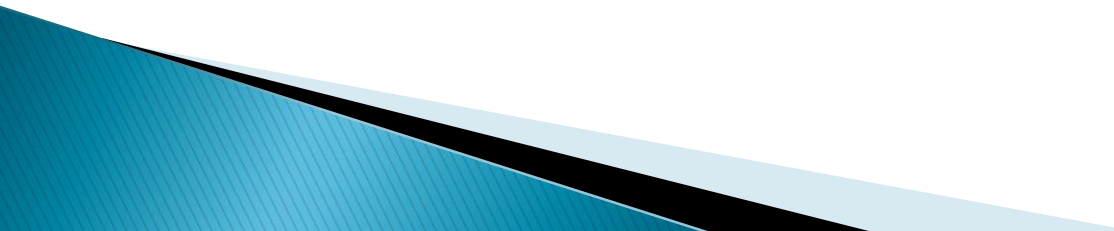
# Introduction

- ▶ Genetic abnormalities in early intrauterine life
  - ▶ Early detection before birth – pre natal diagnosis
  - ▶ Prevention and management
  - ▶ Involves analysis karyotypic, biochemical, physiological and morphological features of foetus to detect chromosomal abnormalities and anomalies, metabolic disorders and morphological malformations.
- 


# Common Techniques

- ▶ Amniocentesis
  - ▶ Chorionic villus sampling
  - ▶ Ultra sound scanning
  - ▶ Foetoscopy
  - ▶ Estimation of alpha-foetoprotein
- 

# AMNIOCENTESIS

- ▶ Transabdominal Amniocentesis – pre natal diagnosis of genetic abnormalities
  - ▶ Most widely used where there is high risk of defective child
  - ▶ More than 30 genetic abnormalities can be detected using AMNIOCENTESIS
  - ▶ Done 14<sup>th</sup> – 16<sup>th</sup> week of pregnancy
  - ▶ During this time some foetal cells will be floating in amniotic fluid
- 

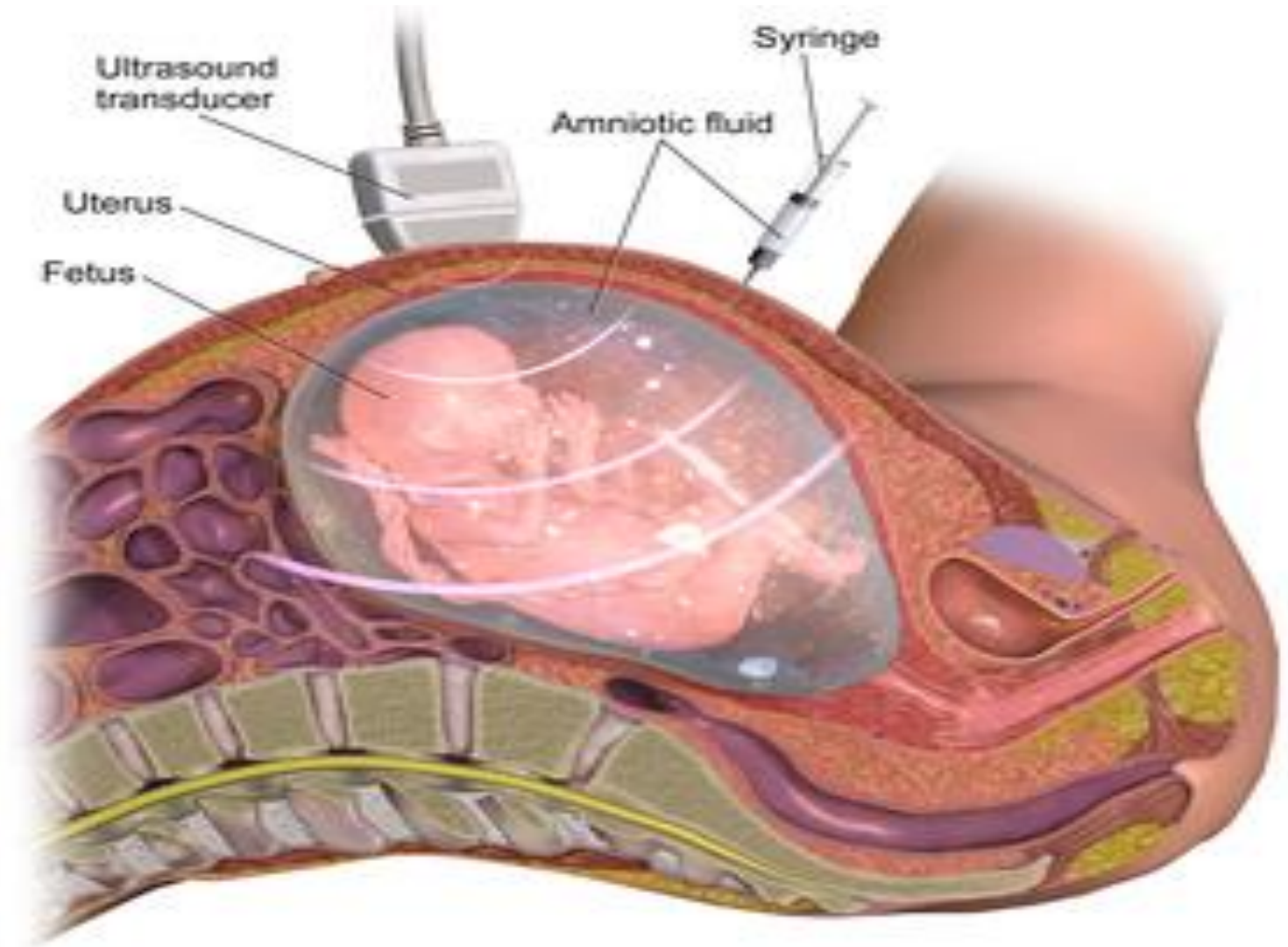
# AMNIOCENTESIS

- ▶ 10–20 ml of amniotic fluid is taken along with foetal cells using a syringe with help of a Ultra Sound Scanner Guidance
  - ▶ These cells are cultured and multiplied in suitable medium
  - ▶ Analysis of karyotype – detect the chromosomal anomalies, severe sex linked hereditary diseases, etc
  - ▶ Detect sex of child
  - ▶ Biochemical tests will enable to detect enzyme deficiencies and congenital metabolic disorders.
- 

# AMNIOCENTESIS

- ▶ AMNIOCENTESIS is the safest technique with less than 0.5% foetal mortality

# AMNIOCENTESIS



# PROCEDURE

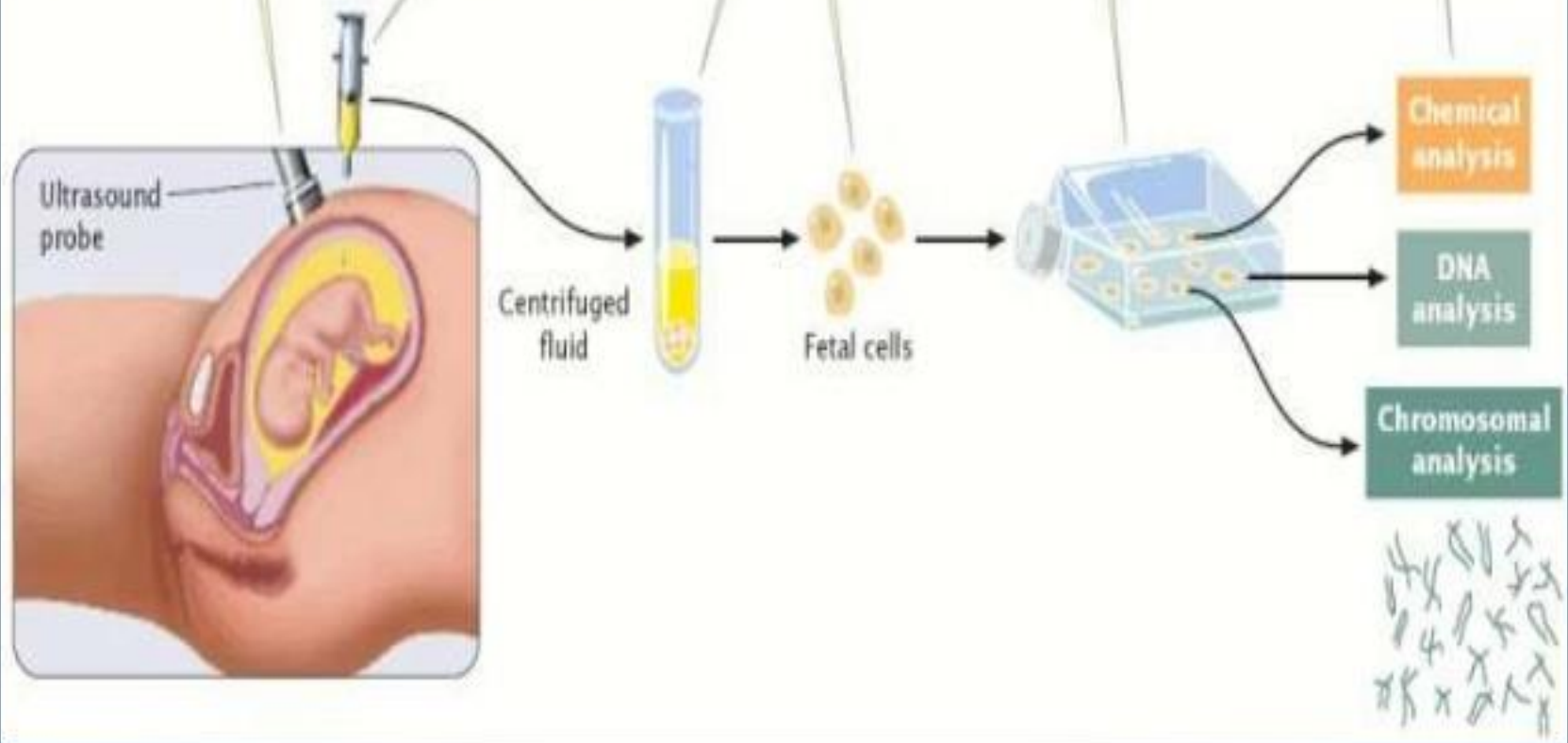
**1** Under the guidance of ultrasound, a sterile needle is inserted through the abdominal wall into the amniotic sac.

**2** A small amount of amniotic fluid is withdrawn through the needle.

**3** The amniotic fluid contains fetal cells, which are separated from the amniotic fluid...

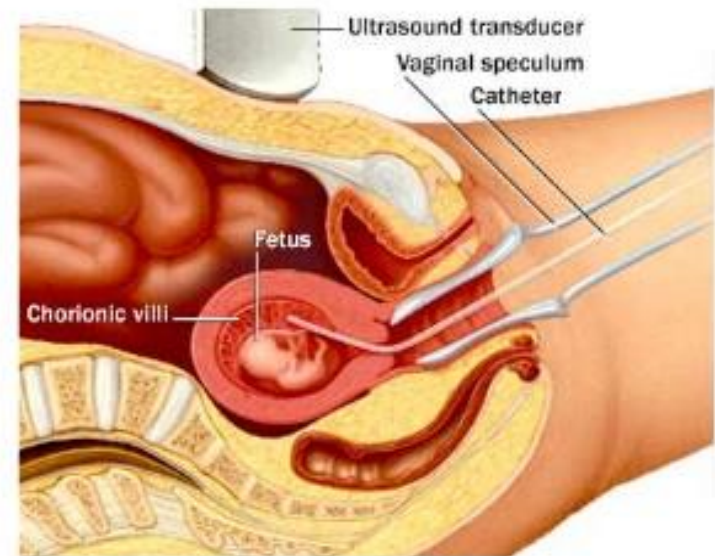
**4** ...and cultured.

**5** Tests are then performed on the cultured cells.



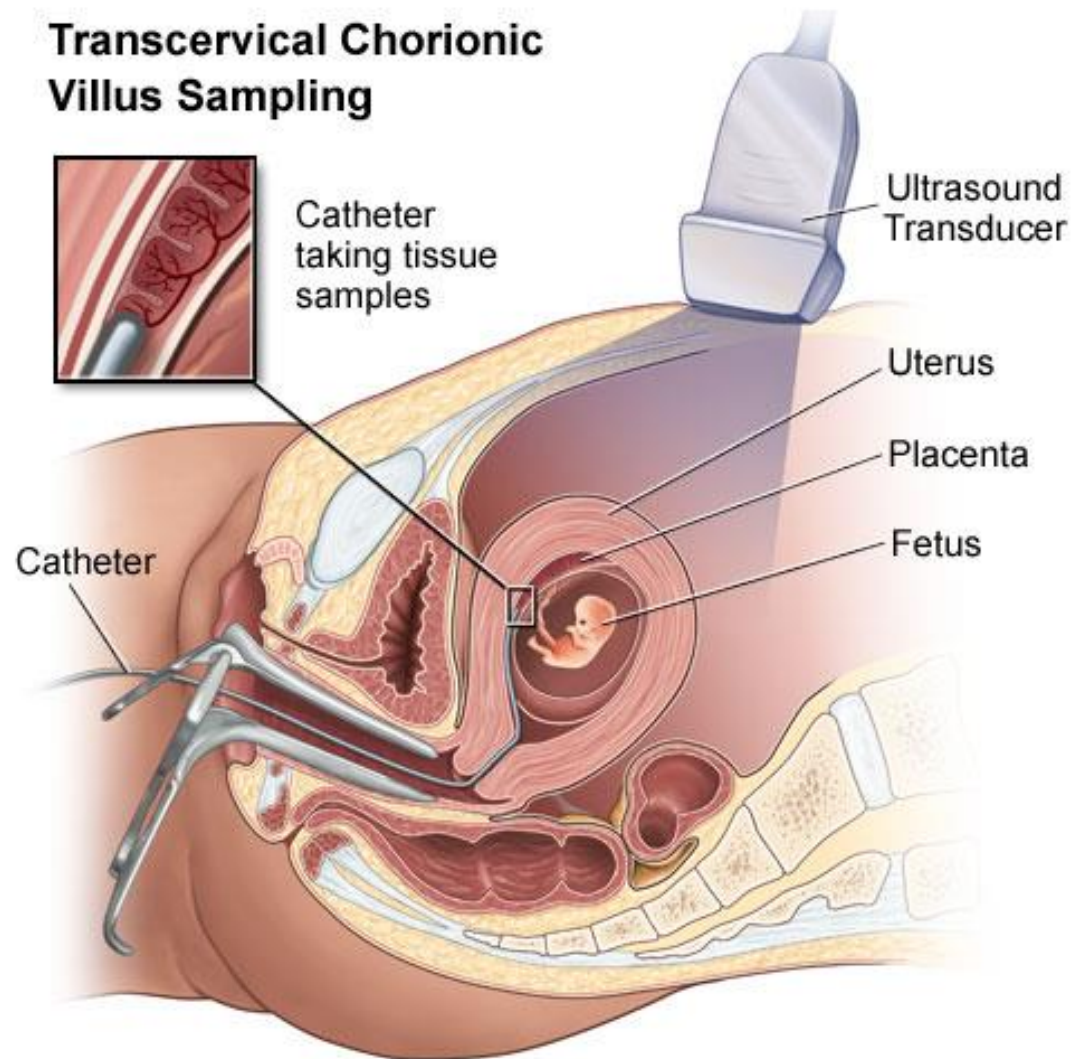
# Chorionic villus sampling (CVS)

- An alternative method to amniocentesis which is more modern but more risky.
- **Definition:** Procedure used for prenatal genetic testing in which a small piece of the chorion is removed from a pregnant woman. A catheter is inserted through the vagina and cervix into the uterus. Suction is then applied to remove the sample.
- Performed between 10 - 12 weeks of conception.
- **Testing for abnormalities in Fetal DNA:**
  - Can test for: the child's sex, down syndrome...etc.
  - Fetal DNA can also be used for karyotyping

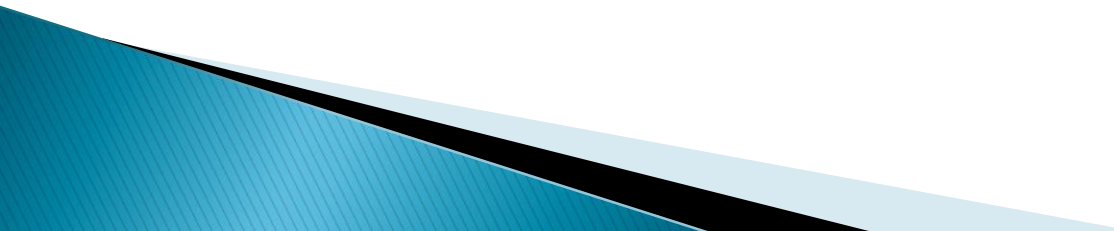


# CHORIONIC VILLUS SAMPLING

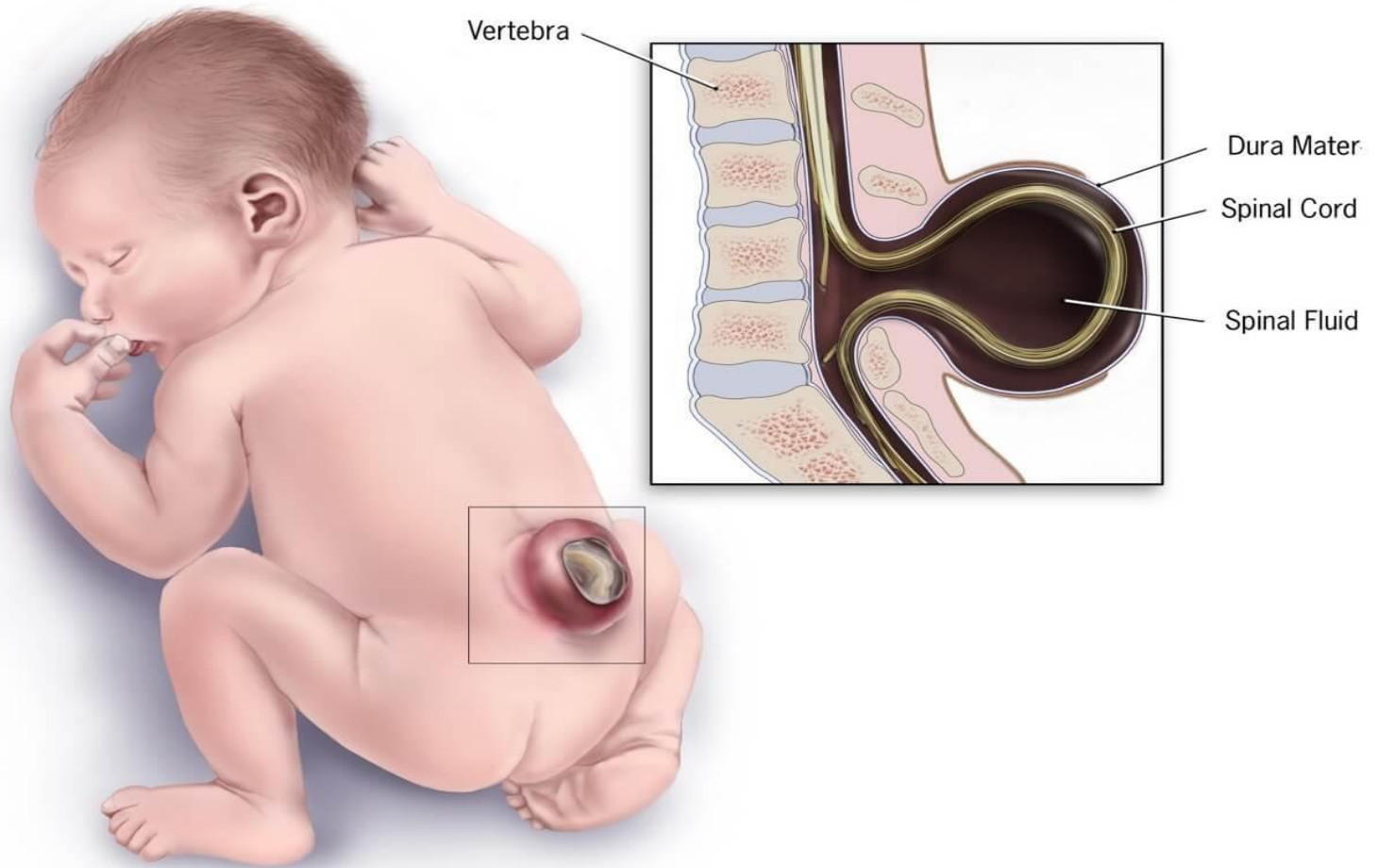
## Transcervical Chorionic Villus Sampling



# ALPHA-FOETOPROTEIN (AFP) ESTIMATION

- ▶ AFP – protein of foetal origin found in amniotic fluid
  - ▶ High levels indicate – foetal malformation or abnormality
  - ▶ Diagnose neural tube defects, spina bifida, anencephalopathy, etc.
- 

## Spina Bifida (Open Defect)



# ULTRASOUND SCANNING



*Thank you*