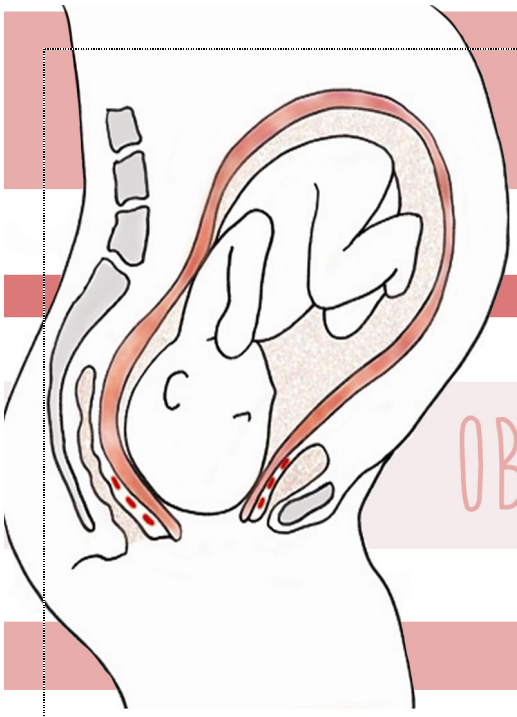

OBSTETRICS AND GYNECOLOGY

(T6) CONGENITAL FETAL ANOMALIES

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Doctor's note Team's note Not important Important 431 teamwork

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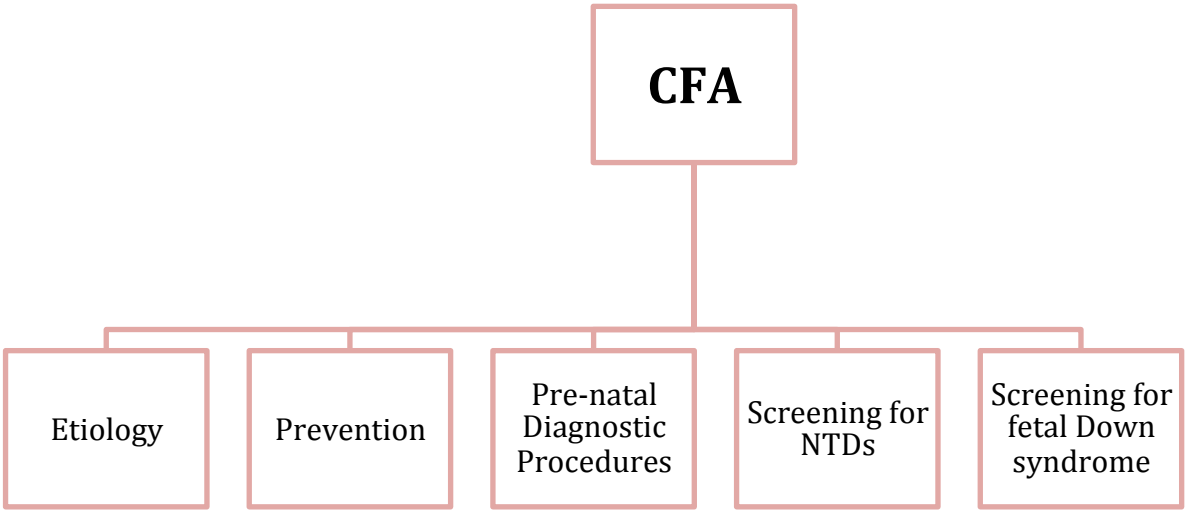
Objectives:

Not given.



Terminology

Congenital means exist since birth, whether clinical evidences are obvious or not obvious.

Anomaly means a deviation from the normal.

Malformation means faulty development of a structure.

❖ Congenital anomalies are also known as birth defects, congenital disorders or congenital malformations.

Congenital anomalies

- Most birth defects cannot be cured.
- Treatment focuses on managing the symptoms. However, there are ways to treat specific birth defects.
- Congenital abnormalities account for 20-25% of neonatal deaths.

Types of congenital anomalies:

- Physical structural defects:
  - ✘ Single structure is affected.
  - ✘ Multiple structures are affected.
- Non-structural defects
  - ✘ Inherited metabolic defects
  - ✘ Functional and behavioral deficits e.g. congenital mental retardation.

Incidence:

- Major congenital anomalies affect about 2 to 5% of all newborns.
- Minor anomalies occur in higher percentage of newborn (about 10%).
- The risk of recurrence of congenital malformations with the same patient is very important in genetic counseling.

- The most common severe congenital anomalies are heart defects, neural tube defects and Down syndrome.

Etiology of Congenital Anomalies:

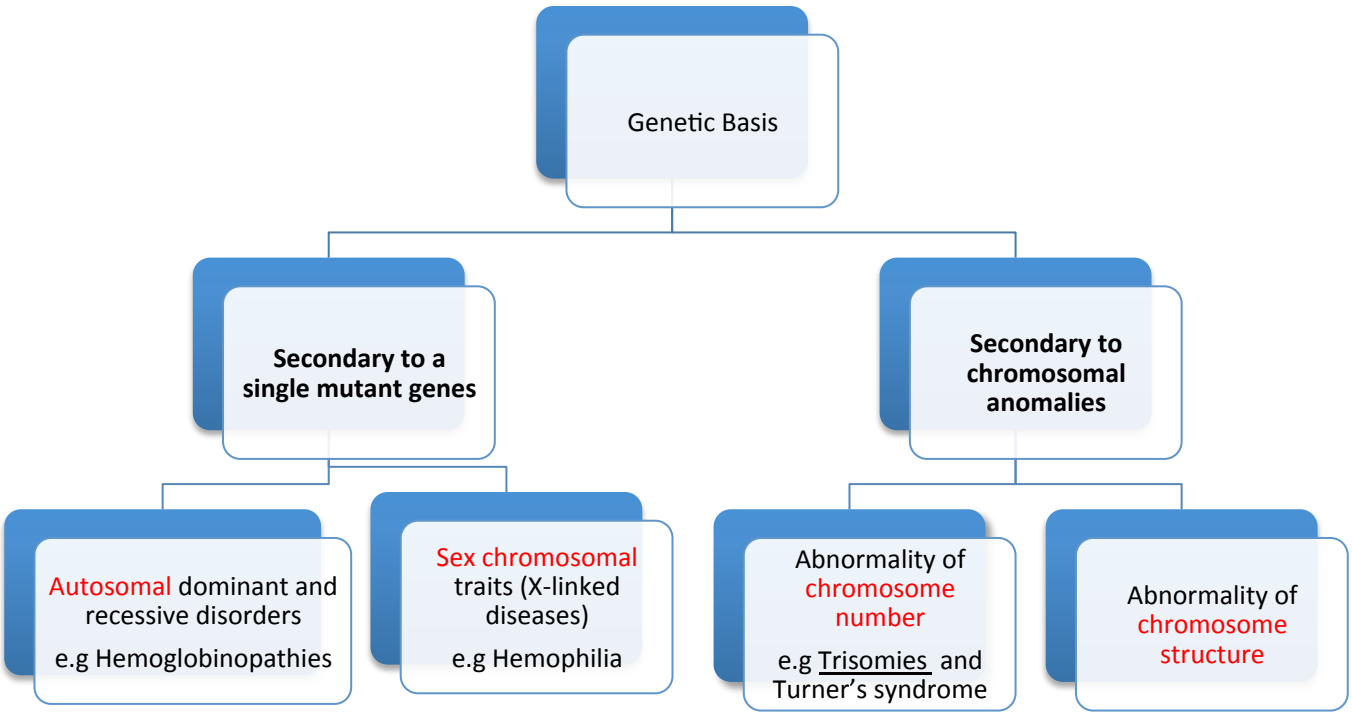
Unknown cause (60%):

- ✖ The causes of malformations are not identifiable in the majority of cases.

Multifactorial factors (20%):

- ✖ Multifactorial etiology denotes the presence of an interaction between genetic predisposition and non-genetic intrauterine factors.
- ✖ Common examples include neural tube defects, certain forms of hydrocephaly, facial clefts, cardiac anomalies, and imperforate anus.

Genetic basis:



Exogenous influences:

Teratogen exposures: Teratogenesis mostly occurs before the tenth week of intrauterine life (the period of embryogenesis).

Intrinsic insults:	Extrinsic insults:
Maternal infections: e.g. rubella virus.	Environmental agents e.g. pesticides
Noninfectious systemic e.g. diabetes mellitus, and seizure disorders.	Drug exposure and medications e.g. Progesterone , androgenic hormones causing masculinization of the female fetus and thyroid and antithyroid drugs.
Functional virilizing lesions of the ovary and adrenal glands.	Physical injury e.g. Exposure to high doses of ionizing radiation

The Food and Drug Administration "FDA" lists five categories of tabling for drug use in pregnancy

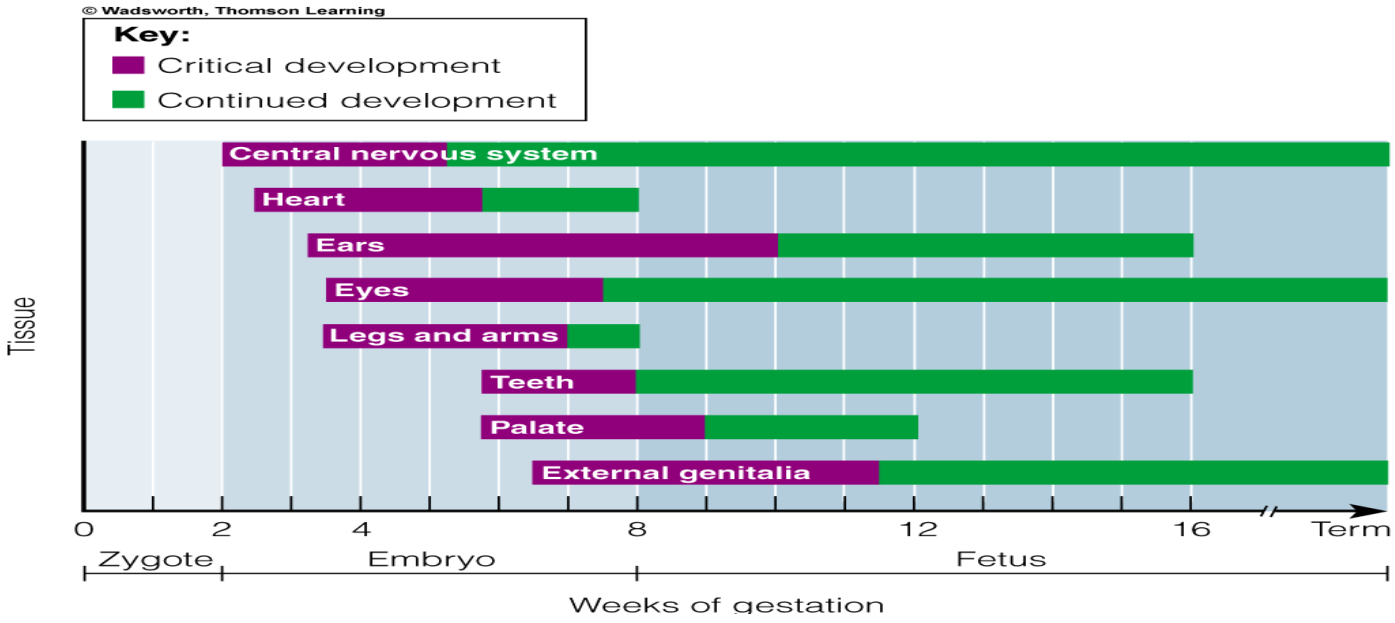
A. Controlled studies in women failed to demonstrate a risk to the fetus in the first trimester, and the possibility of fetal harm appears remote.

B. Animal studies do not indicate a risk to the fetus, there are no controlled human studies, or animal studies do show an adverse effect on the fetus, but well –controlled studies in pregnant women have failed to demonstrate a risk to the fetus.

C. Studies have shown the drug to have animal teratogenecity or embryocidal affects, but no controlled studies are available in either animals or women.

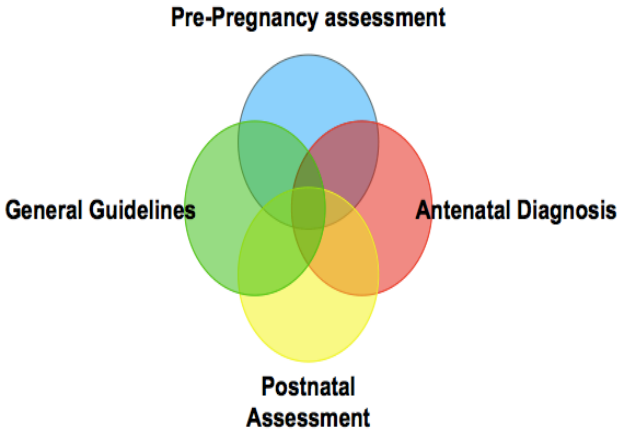
D. Positive evidence of human fetal risk exists, but benefits in certain situations (e.g., serious diseases for which safer drugs are ineffective) may make use of the drug acceptable despite its risks.

E. Studies in animals or humans have demonstrated fetal anomalies and the risk clearly outweighs any possible benefit.



Prevention

I. General guidelines:



- Control of medications during pregnancy: Minimize drug exposure.
- Detection and control of relevant maternal diseases.
- Genetic counseling.
- Prenatal diagnosis of genetic conditions and selective termination of the affected pregnancy or in-utero treatment if possible.
- Discourage consanguineous marriages when appropriate e.g. closed family, previous inheritable malformation in the family.

❖ **Screening and Counseling before (pre-conception) and around the time of conception is mandatory.**

II. Screening can be conducted during the 3 periods listed next:

➢ **Preconception screening** can be useful to identify people at risk for specific disorders or at risk for passing a disorder onto their children.

- Screening includes obtaining family histories and carrier screening and is particularly valuable in countries where consanguineous marriage is common.

➢ **Peri-conception screening**

- Maternal characteristics may increase risk, and screening results should be used to offer appropriate care according to risk.

This may include screening for:

1. Young or advanced maternal age.
2. Screening for use of alcohol, tobacco or other psychoactive drugs.
3. Ultrasound can used to screen for Down Syndrome during the first trimester.
4. Additional tests and amniocentesis may help in the detection of neural tube defects and chromosomal abnormalities during first and second trimester.

➢ **Neonatal screening**

- Includes clinical examination and screening for disorders of blood, metabolism and hormone production.
- Screening for deafness and heart defects, as well as early detection of congenital anomalies, can facilitate life-saving treatments and prevent progression towards some physical, intellectual, visual or auditory disabilities.
- In some countries, babies are routinely screened for abnormalities of the thyroid or adrenal glands before discharge form the maternity unit

Prevention of CNS Anomalies:

a. Correction of dietary habits: Certain dietary patterns are deficient in folic acid, and need to be modified.

b. Peri-conceptional folate administration. (3 months before pregnancy)

c. Peri-conceptional control of DM. (3 months before pregnancy)

d. Special tests during pregnancy in high-risk individuals for early detection of such anomalies.

Pre-natal Diagnostic Procedures

1. History Taking
2. Abnormal findings during routine examination
3. Abnormal findings during routine investigations
4. Specific Antenatal diagnostic procedures

1. History Taking

Leading questions of special significance:

- Maternal age.
- Personal or family history of congenital anomalies e.g. familial disorders in the blood relatives.
- The ethnic origin.
- Significant maternal diseases: diabetes mellitus, infections, and acute maternal illness.

2. Suspicious Findings On Clinical Examination

A higher incidence of congenital anomalies are detected in association with:

- **Oligohydramnious: (Imp.)**
  - Renal dysplasia, renal agenesis,

- Bladder outlet obstruction and
- Intrauterine growth retardation.

✘ **Polyhydramnios:**

- Central nervous system anomalies: anencephaly, hydrocephaly.
- Gastrointestinal malformations: Tracheoesophageal fistula, duodenal atresia.

✘ Threatened abortion.

✘ Unexplained IUGR.

3. Suspicious Findings On Routine Investigations

Suspicious findings on ultrasound screening:

A. Early IUGR

B. IUGR

C. Oligohydramnios

D. Hydramnios

E. Restricted fetal movements

4. Specific Prenatal Techniques

I. Non-invasive techniques:

1. Fetal visualization includes the following:

✓ Ultrasound
✓ Fetal echocardiography.

} Each one has a seperate tutorial

✓ Magnetic resonance imaging (MRI):

Because of fetal movements, its application has been limited.

✓ Radiography:

* Congenital anomalies can be visualized by ultrasound between 16-20 weeks' gestation.

* The most sensitive marker for congenital anomalies in the 2nd trimester is Nuchal fold thickness.

The fetal skeleton can be visualized by radiography from 10 weeks' gestation onward. This technique is used for the diagnosis of inherited skeletal dysplasias, particularly osteochondrodysplasia.

2. **Screening for neural tube defects (NTDs)**

Involves measuring maternal serum alpha fetoprotein (MSAFP).

3. **Screening for fetal Down syndrome includes the following:**

- ✓ Measuring MSAFP
- ✓ Measuring maternal unconjugated estriol
- ✓ Measuring maternal serum beta-human chorionic gonadotropin (HCG)
- ✓ Measuring inhibin

4. **Other noninvasive techniques include the following:**
   - ✓ Separation of fetal cells from the mother's blood
   - ✓ Assessment of fetal-specific DNA methylation ratio

II. Invasive techniques: (You don't need to go through the details of invasive techniques)

1. **Fetal visualization techniques that are invasive include the following:**
   - ✓ Embryoscopy
   - ✓ Fetoscopy

2. **Invasive fetal tissue sampling techniques include the following:**
   - ✓ Amniocentesis
   - ✓ Chorionic villus sampling (CVS)
   - ✓ Percutaneous umbilical blood sampling (PUBS)
   - ✓ Percutaneous skin biopsy
   - ✓ Other organ biopsies, including muscle and liver biopsy

3. **Pre-implantation biopsy of blastocysts obtained by in vitro fertilization.**

4. **Cytogenetic investigations** that are invasive include the following:
   - ✓ Detection of chromosomal aberrations

- ✓ Fluorescent in situ hybridization.

1. Fetal visualization -Embryoscopy

- Embryoscopy is performed in the first trimester of pregnancy (up to 12 weeks' gestation).
- In this technique, a rigid endoscope is inserted via the cervix in the space between the amnion and the chorion, under sterile conditions and ultrasound guidance, to visualize the embryo for the diagnosis of structural malformations.

2. Fetal visualization – Fetoscopy

Is performed during the second trimester (after 16 weeks' gestation). In this technique, a fine-caliber endoscope is inserted into the amniotic cavity through a small maternal abdominal incision, under sterile conditions and ultrasound guidance, for the visualization of the embryo to detect the presence of subtle structural abnormalities. It also is used for fetal blood and tissue sampling. Fetoscopy is associated with a 3-5% risk of miscarriage; therefore, it is superseded by detailed ultrasound scanning.

3. Fetal tissue sampling:

Amniocentesis

Chorionic villus sampling(cvs)

Percutaneous umbilical blood sampling (PUBS)

Disscused in perenatal testing .lecture

4. Cytogenetic Investigations

5. Detection of chromosomal aberrations

- ▶ Chromosomal aberrations, such as deletions, duplications, translocations, and inversions diagnosed in affected parents or siblings, can be detected prenatally in a fetus by chromosomal analysis.

Screening for NTDs

Screening for NTDs (Neural tube defect) is recommended if the following are present:

- Ultrasound findings indicate NTDs.
- A child with NTDs is already in the family.
- A family history of NTDs exists, especially a mother with NTDs.
- The mother has diabetes mellitus.
- Maternal exposure to drugs, such as valproic acid, is associated with NTDs.
- Elevated level of MSAFP is present.

Measuring maternal serum alpha-fetoprotein:

- The developing fetus has 2 major blood proteins, albumin and alpha-fetoprotein (AFP), while adults have only albumin in their blood. The MSAFP level can be used to determine the AFP levels from the fetus. AFP is produced by the yolk sac and later by the liver; it enters the amniotic fluid and then the maternal serum via fetal urine.
- In the condition of an open NTD (eg, anencephaly, spina bifida) and abdominal wall defects in the fetus, AFP diffuses rapidly from exposed fetal tissues into amniotic fluid, and the MSAFP level **rises**. However, the MSAFP levels also increase with gestational age, gestational diabetes, twins, pregnancies complicated by bleeding, and in association with intrauterine growth retardation.
- The MSAFP test has the greatest sensitivity between 16-18 weeks' gestation, but it also can be performed between 15-22 weeks' gestation.
- A combination of the MSAFP test and ultrasonography detects almost all cases of anencephaly and most cases of spina bifida.

Abnormal maternal serum alpha-fetoprotein (MSAFP).

A. MSAFP is elevated (2.5 MOM)

- Fetal anomalies such as neural tube defects, abdominal wall defects e.g. omphalocele, esophageal or intestinal obstruction, cystic hygroma, urinary obstruction, renal anomalies: polycystic kidneys, osteogenesis imperfecta, Turner's syndrome, and Rh disease.
- Obstetrical complications such as low birth weight, oligohydramnios, multifetal gestation.

B. MSAFP is abnormally low (<0.2 MOM)

A. Chromosomal trisomies of the fetus. The values are low in only one third of Down's syndrome,

B. Gestational trophoblastic disease, and fetal death.

❖ Screening for fetal Down syndrome (Imp.)

A. Measuring maternal serum alpha-fetoprotein:

In cases where a low level of MSAFP is reported, it indicates the condition of Down syndrome or other chromosomal aneuploidy and failing pregnancies.

B. Measuring maternal unconjugated estriol:

Down syndrome:

↓ AFP & Estradiol.

↑ BHCG & Inhibin.

- The amount of estriol in maternal serum depends upon viable fetus, a properly functioning placenta, and on maternal well-being.
- Fetal adrenal glands produce dehydroepiandrosterone (DHEA) that gets metabolized to estriol in the placenta. Estriol crosses to the maternal circulation and is excreted either by maternal kidney in urine or by maternal liver in the bile.
- In the third trimester, the level of estriol gives an indication for the well-being of the fetus. A low level of estriol is an indication of Down syndrome and adrenal hyperplasia with anencephaly.

The triple test:

Specified combinations of maternal serum assay of AFP, unconjugated oestriol (uE3) and hCG. Special tables are used to interpret the results. + Inhibin = Quadruple test

- If the estriol level drops to a great level, then it indicates risk to fetus.

C. Measuring maternal serum beta-human chorionic gonadotropin:

- Following conception and implantation of the developing embryo into the uterus, the trophoblasts produce enough beta-HCG, which is an indication for pregnancy.
- In the middle to late second trimester, the level of beta-HCG also can be used in conjunction with the MSAFP level to screen for chromosomal abnormalities.
- An increased beta-HCG level coupled with a decreased MSAFP level suggests Down syndrome.
- In early pregnancy, if its amount is found to be lower than expected, it indicates abortion or ectopic pregnancy. If the level of HCG is estimated to be considerably high, then it indicates the possibility of trophoblastic diseases.
- The elevated level of HCG, along with absence of the fetus on ultrasonography, indicates a hydatidiform mole.

D. Measuring maternal inhibin-A levels

- The hormone inhibin is secreted by the placenta and the corpus luteum. Inhibin-A can be measured in maternal serum. An increased level of inhibin-A is linked with an increased risk for trisomy 21. A high inhibin-A level may also be associated with a risk for preterm deliver

E. Separation of fetal cells from the mother's blood

- Fetal blood cells make access to maternal circulation through the placental villi. These cells can be collected safely from approximately 18 weeks' gestation onward, although by successful procedures, these cells can be collected at 12 weeks' gestation. The fetal cells can be sorted out and analyzed by different techniques.

- Fluorescent in situ hybridization (FISH) is one technique that can be used to diagnose aneuploid conditions, such as trisomies and monosomy X. In the condition of fetal infection with such viruses as rubella, cytomegalovirus, and toxoplasmosis, the viral immunoglobulin M (IgM) or DNA also can be identified in fetal blood.
- Fetal blood cells can be analyzed for the diagnosis of genetic disorders using molecular genetic techniques by isolating DNA and amplifying it by polymerase chain reaction (PCR).
- Fetal cells separated from a mother's blood have been successfully used in the diagnosis of cystic fibrosis, sickle cell anemia, and thalassemia in a fetus.

Benefits of Prenatal Diagnosis

- An offer of prenatal screening provides prospective mothers the option of choosing or declining to receive genetic information pertinent to their personal situation prior to conception.
- After conception, prenatal diagnosis provides various benefits. Prenatal diagnosis determines the outcome of pregnancy and identifies possible complications that can arise during birth. It can be helpful in improving the outcome of pregnancy using fetal treatment. Screening can help couples determine whether to continue the pregnancy and prepares couples for the birth of a child with an abnormality.

Fetal congenital anomalies associated with maternal diabetes

Maternal diabetes can affect different fetal organs.

1. **Cardiac anomalies:** (The most common) e.g. Ventricular septum defects and transposion of great arteries.

2. **Central nervous system anomalies**: eg. Spina befida, neural tube defect, Caudal regression syndrome (when you see a neonate with caudal regression syndrome you know his mother was diabetic during pregnancy, very specific).
3. **Renal anomalies:** Renal agenesis. Hydronephrosis.
4. **Pulmonary anomalies:** Surfactant deficiency.
5. **Gastrointestinal anomalies:** meconium plug syndrome

Summery:

- 60% of congenital anomalies are unknown cause.
- **Oligohydramnious** could be cause by Renal dysplasia, renal agenesis or Bladder outlet obstruction and Intrauterine growth retardation
- In Down syndrome: Low level of AFP and Estradiol + High level of BHCG and Inhibin.
- You check for congenital anomalies using US during week 16-20.
- In cardiac anomalies, associated extra-cardiac lesions are present in 30 % of cases.
- In gestational diabetes, cardiac anomalies are the most common, while caudal regression syndrome is the most specific.

The rest wasn't discussed by dr. Malak Alhakeem neither she put it in her slides. It's from Prof. Lulu slides.

Anomaly Scans

- Structural Assessment:
  - Systematic documentation of the essential fetal anatomy: head, neck, chest, abdomen, limbs, external genitalia,
  - Assessment of amniotic fluid volume.
  - The umbilical cord and its vessels.
- Measurements are calculated (Biometry):
  - The most significant measurements are BPD (biparietal diameter), OFD (occipto-frontal diameter), HC (head circumference), and femur length.
  - Serial measurements are used to evaluate the growth pattern of organs.

Laboratory Investigative Procedures

» **Chromosome analysis:**

- Simple cytogenic techniques
- Special culture and examination for minor chromosome aberrations

» **Biochemical analysis:**

- Bilirubin in rhesus isoimmunization: normally it is excreted in fetal urine.
- Microvillar enzymes from the fetal gut for cystic fibrosis.
- Alpha-fetoprotein: nearly all of AFP is fetal in origin.

» **DNA analysis:**

- Polymerase chain reaction (PCR)
  - Chorionic villi.
  - Amniotic fluid or fetal blood.

- Molecular genetics is the study of the structure of the genes. Its value in the obstetric practice is related to the study of inherited disease.

» **Fetal Gender Determination**

- risk of a serious X-linked hereditary disorders, for which no specific prenatal diagnostic test is readily available.
  - The aim is termination of pregnancy if the fetus is of the exposed type of the X-linked disorder.

» **Fetal infection**

- Testing either the amniotic fluid or the chorionic villi or fetal blood.
- Testing for possible fetal viral infection is indicated in cases of maternal virus infection.

» **Hematological analysis:**

- Haemoglobinopathies, coagulation disorders, and fetal blood grouping

What to Do when a CFA is discovered?

Counseling

1. **Termination of pregnancy:**

   I. Cultural background and religious beliefs play a role in the decision,

   II. Indicated with malformations incompatible with life to e.g. anencephaly, and multiple malformations with poor prognosis.

2. **Prenatal therapy or surgery, available for few conditions only:**

   - Medical treatment:

   • Intrauterine heart failure and supraventricular arrhythmia's:

   - Maternal medication of digoxin and propranolol.

- Congenital adrenal hyperplasia:

  - Dexamethasone to the mother to reduces accumulation of androgenic steroids and to lessens virilization of female fetus.

✖ Surgical treatment:

3. **Planning the best circumstances for delivery.**
4. **Congenital anomalies requiring urgent surgical procedures and special care after delivery:**

   i. Gastrointestinal tract obstruction: pyloric stenosis, esophageal atresia, intestinal atresia, duodenal atresia, jejuno-ileal atresia, colonic atresia, ano-rectal malformations.

   ii. Urinary tract obstruction.

   iii. Congenital diaphragmatic hernia.

   iv. Exomphalos and extrophy (bladder cloaca).

   v. Open neural tube defects.

   vi. Congenital adrenal hyperplasia.

Some Congenital Fetal Anomalies

✖ Anomalies of the Nervous System

✖ Anomalies of the GIT

✖ Anomalies of the Genito-urinary Tract

✖ Cardiovascular Anomalies

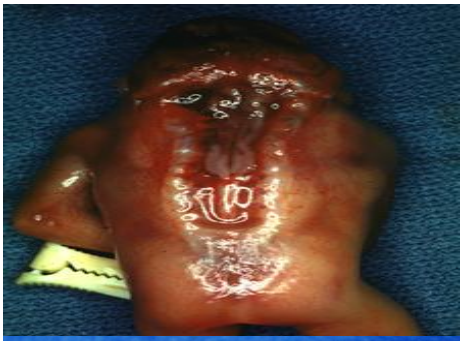
✖ Anterior Abdominal wall defects

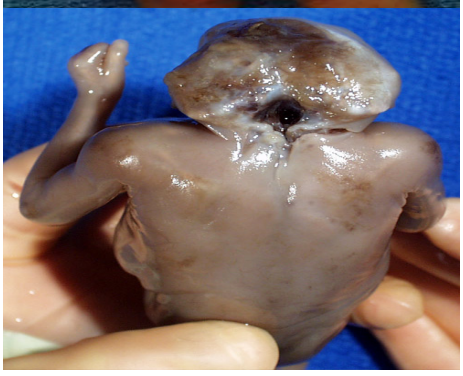
✖ Diaphragmatic hernia

✖ Down's Syndrome

✖ Non Immune Hydrops Fetalis

Anencephaly



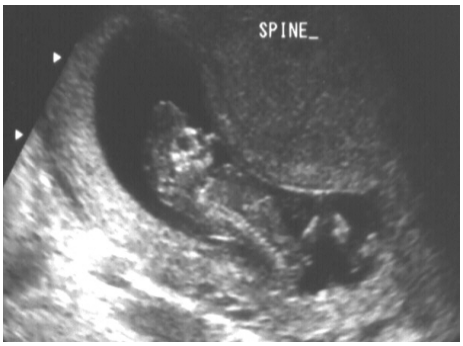


Anencephaly is a lethal anomaly due to the absence of:

- The membrane-ossifying bones of the cranial vault and consequently the skull and scalp.
- The cerebral hemispheres, underlying the above structures
- It is more common in girls.

1. Antenatal diagnosis:

- Clinical features: Polyhydramnios and abdominal palpation (absence of head).
- Investigations:
  - Raised plasma and amniotic fluid α-fetoprotein levels and
  - Ultrasound features.



2. Management of anencephalic pregnancy:

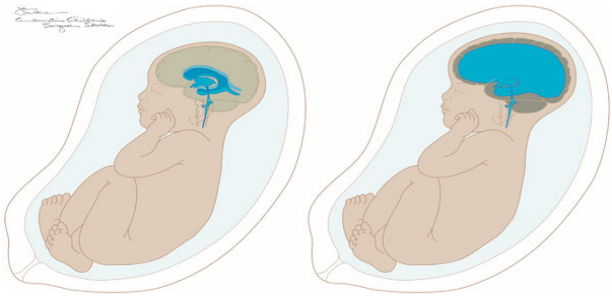
- Elective abortion.
- Vaginal delivery :there is an increased incidence of face presentation and shoulder dystocia.

Hydrocephalus

Hydrocephalus is an **excess of cerebrospinal fluid within the ventricles, and the subarachnoid space.**

- Congenital cerebral malformations: e.g. Arnold-Chiari malformation.

- Congenital fetal infections e.g. toxoplasmosis, cytomegalovirus.
- Intrauterine intracranial hemorrhage.
- Certain forms are multifactorial origin.



- Obstruction of the aqueduct of Sylvius which may be due to genetic disorders as trisomy 21, infection as toxoplasmosis and cytomegalovirus, intracranial tumors, and intracerebral hemorrhage.
- Chromosomal abnormalities: triploidy, trisomy 18, and X-linked trait.

3. Antenatal diagnosis of hydrocephalus:

- Clinical:
  - Polyhydramnios.
  - Large size head.
  - Breech presentation is common
  - During labor, vaginal examination: Wide sutures, large fontanelles and thin, soft identable cranial bones.
- Ultrasound:
  - Diagnostic value: serial ultrasound studies are important to avoid false positive diagnosis.
  - Prognostic value:
    - The type of hydrocephalus
    - The site and extent of the brain injury
    - The cerebral cortex compression is followed regularly.

4. Congenital hydrocephalus is commonly associated with other neurological or general congenital malformations e.g. spina bifida, harelip, clubfoot, or imperforate anus.

Management Options:

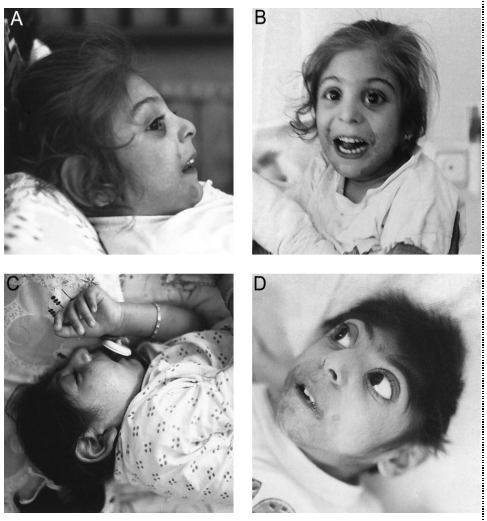
5. Termination of pregnancy could be offered, if diagnosis is definite in the early second trimester.
    - Search for associated anomalies.
    - Establishment of the etiology if possible.
    - Amniocentesis to determine fetal karyotype.
6. Intrauterine therapy (under ultrasound guide): Attempts at intrauterine ventriculo-amniotic shunts (with a one way valve may be done to drain CSF from cerebral ventricles into the amniotic sac to prevent compression and atrophy of brain tissues) are being tested.
7. **Effects of hydrocephaly on vaginal delivery:**

- ✖ Breech presentation is common.
- ✖ Feto-pelvic disproportion: Non-engagement of the presenting large head and obstructed labor. Some infants cannot be delivered without destructive procedure or cesarean section.

- Conduct of delivery:
    - ✖ Destructive procedures to facilitate vaginal delivery.
    - ✖ The head is perforated and cerebrospinal fluid is drawn off.
    - ✖ In breech presentation, the aftercoming head is either perforated or spinal tapping is carried out. A metal canula is introduced through the spinal canal (or through spina bifida is present).

8. Delivery of a live newborn, with possible cesarean section, when there are favorable signs.
    - ✖ Absence of associated anomalies.
    - ✖ Stable hydrocephalus.
    - ✖ Cerebral mantel remains more than 10 mm (thickness of cerebral cortex) and the newborn will have surgical procedures after delivery i.e. shunting operations (ventriculo-peritoneal shunt).

Microcephaly



Microcephaly is an **abnormally small head.**

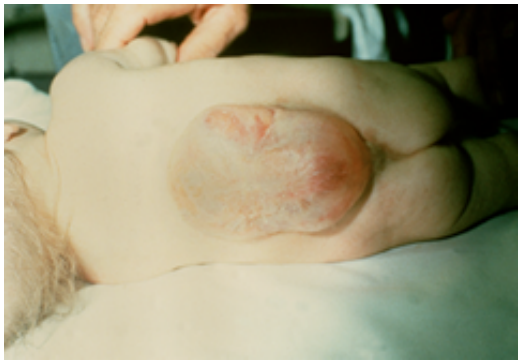
9. Diagnosis depends on biometry: Occipto-frontal diameter (OFD) and BPD are reduced.

10. Complications of microcephalus:

- Mental retardation: the smaller the head the worse the prognosis.
- The presence of associated anomalies

Spina bifida and Meningomyelocele

Spina bifida is a defect in the spine resulting from failure of the two halves of the vertebral arch to fuse.



- Ultrasound features of spina bifida:
  - The features appear by 18-20 weeks gestation in about 90 per cent of cases.
  - The posterior ossification centers of the spine, at the level of the defect are widely spaced. The vertebral segment appears in U-shape. The defect may b e visualized on longitudinal scanning.
  - There is restricted motility of the lower limbs

11. The prognosis is related to the following:

- Presence and severity of neurological involvement
- The presence of associated abnormalities: e.g.

» Arnold-Chiari malformation (coexisting hydrocephalus due to prolapse of the cerebellar hemispheres (obstructing the flow of CSF). It is to be noted that 90 per cent of cases of spina bifida have or develops hydrocephalus later on.

» Orthopedic malformations: congenital dislocation of hips, foot deformity e.g. talipes, and kyphoscoliosis.

» Chromosomal defects: e.g. trisomy 18.

12. Types:

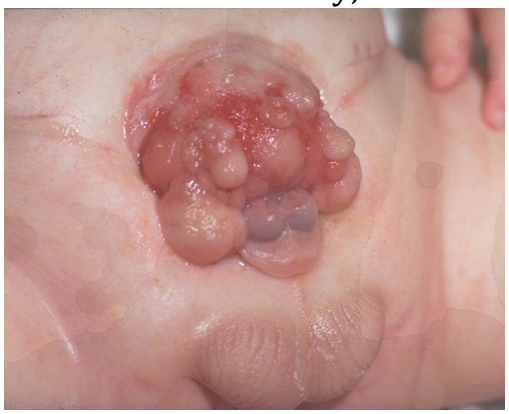
- Spina bifida occulta: The bone only is affected, while the spinal cord and the membranes are intact. There may be a patch of hairy skin or a dimple over the affected area. It has a good prognosis. No treatment is required.
- Spina bifida cystica ' overta'which includes:
  - Meningocele. It is protrusion and herniation of the meninges, through a bony deficit to the skin.
  - Meningomyelocele: It is a protrusion of heterotopic neural tissue with the meninges. The defect is in the midline and affects the skin of the back, muscles, bones of the vertebral arches and neural tube. The membrane is easily ruptured
  - Myelocele: No skin or meninges to cover the lesion. It is usually incompatible with life.

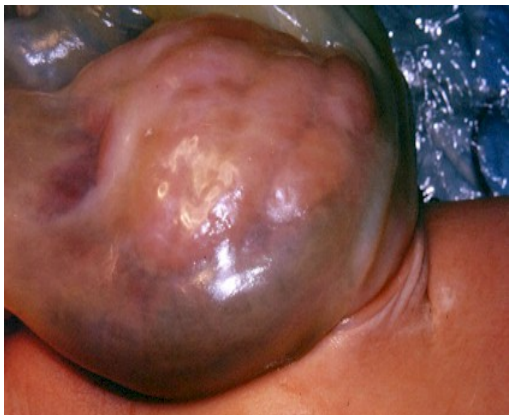
13. Immediate care after delivery:

- Cover the lesion with a sterile non-adhesive dressing to minimize trauma and infection.
- Search for associated malformations
- Consult a neurosurgeon.

Anterior Abdominal Wall Defects

- The defect in closure may involve the lower part of abdominal wall only, or bladder, urethra and penis, and/or clitoris and labia.
- These are associated with increased MSAFP.
- Unfortunately high percentage of cases have an associated cardiac and chromosome abnormalities.





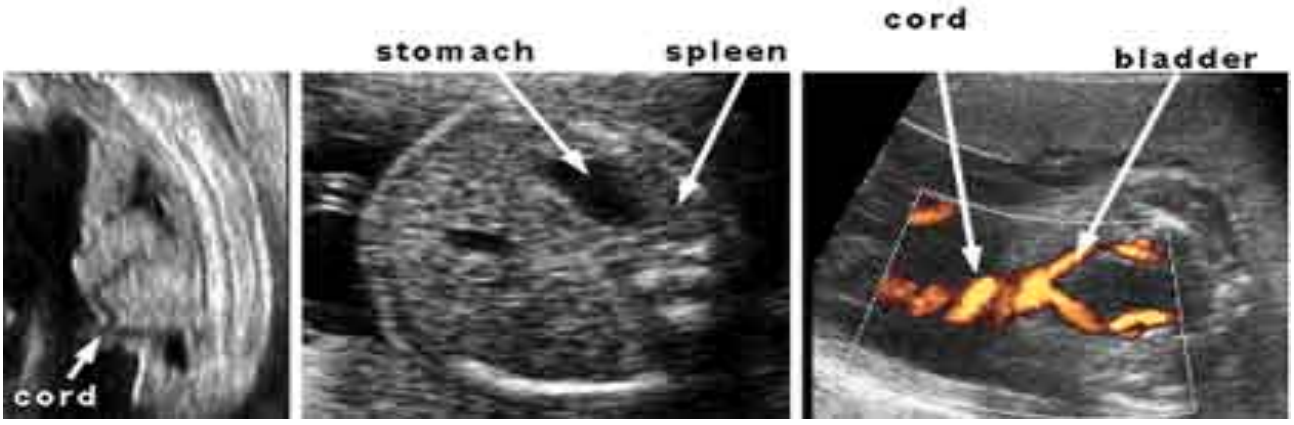
Types:

- Omphalocele (exomphalos): Congenital herniation of some of the intra-abdominal contents through the umbilical ring.

DD. Gastroschisis – Hernia of the umbilical cord

- Ectopia vesicae:

the defect involves the bladder. [Exstrophy of bladder: The trigone and urethral orifices are exposed]



Management of defects of the anterior abdominal wall:

- Immediate care:
  - Do not clamp protruding mass. Clamp the umbilical cord few centimeters distal to the swelling.

- Keep the hernial sac moist and warm, using pads soaked in a normal saline solution.
- Protect from irritation, traumatic injury of covering membrane or organs and from infection.
- Empty the stomach of air with a nasogastric tube.

✖ Surgical corrective repair.

Gastrointestinal Tract Anomalies

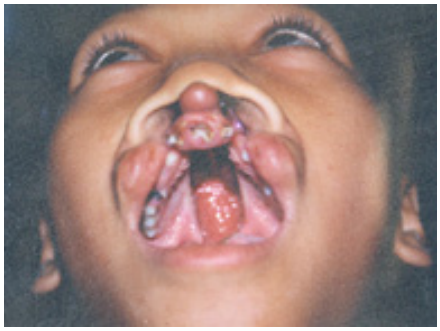
The prognosis is generally good after surgical correction [provided NO other anomalies co-exist]

Malformations presenting with intestinal obstruction:

- Bowel obstruction above the ileum.
  - All usually results in polyhydramnios due to failure of absorption of swallowed amniotic fluid.
  - After delivery there is vomiting and abdominal distension.
  - Surgery at early neonatal life is successful in duodenal atresia, esophageal atresia, pyloric stenosis, jejeunal and ileal atresia.
- Bowel obstruction below the ileum.
  - Generalized distention of bowel loops on ultrasound
  - The causes are:
    - Dysfunctional: the distention may be transient and resolve spontaneously.
    - Meconium ileus.
    - Anal atresia and imperforate anus.
    - Hirschsprung's disease

Cleft Lip and Cleft Palate

- Several teratogens may cause either of the two conditions. Generally both are not associated with other gastrointestinal malformations.



- **Cleft Lip:** It is cleft in the upper lip.
  - It may be unilateral or bilateral.
  - a small notch in the vermilion to a complete separation extending into the of the nose.
  - There may be feeding problems.
  - It is often associated with floor cleft palate.
  - Surgical repair can be done in the first few days of life.
- **Cleft Palate:**
  - Bifid uvula. A cleft on midline uvula.
  - Cleft soft palate.
  - Cleft bony palate.
  - Gap in the alveolar arch.
    - Feeding problems may develop e.g. aspiration and infection.
    - Corrective surgery: best results if performed around one year of age.
    - Postoperative complications: are not rare.
      - Recurrent otitis media.
      - Speech and hearing problems.

Urogenital System Abnormalities

- Renal Agenesis:
  - It is a rare abnormality. It is fatal when bilateral.

- Potter's syndrome:
  - Renal agenesis,
  - pulmonary hypoplasia,
  - oligohydramnios,
  - IUGR, characteristic compressed facial features, flattened nose, small chin, prominent epicanthal folds and with a low-set ears.
  - At birth there is severe respiratory problems. Ultrasonic confirmation is difficult because it is based on the documentation of the absence of the renal echoes, in severe oligohydramnios. Also perirenal fat or adrenal glands may mimic the renal shadow.

Obstructive uropathy:

- Various causes of obstruction to urinary flow. Ultrasound diagnosis: Enlarged bladder and/or hydronephrosis. The condition may be unilateral or bilateral.
- Features after delivery: abdominal mass because of enlarged bladder and/or hydronephrosis.

Types:

- Pelviureteric junction obstruction is considered as acute rather than chronic obstruction. The prognosis is favorable.
- Posterior urethral valves: occur in male. They are responsible to varying degrees of dilatation of the renal tract.
- Complete urethral stenosis: complete absence of amniotic fluid and gross dilatation of the renal tract. Kidneys may be subjected to severe dysplasia, and appears small with increased echogenicity

Anomalies of the external genitalia

- Chromosomal anomalies.

- Adrenal cortical hyperplasia.
- Maternal intake of adrenogenic substances.

Undescended Testicles

- Preterm
- When to correct

Epispadius/Hypospadius

- Associated with XXY, Trisomy 18

Cardiac Anomalies

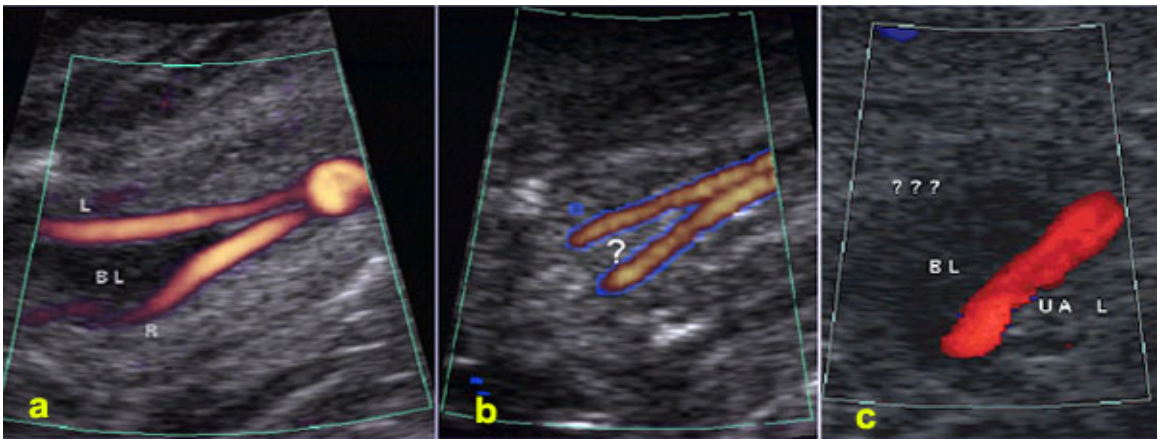
- Some are minor self-limiting or easily correctable defects, while some are serious and can be lethal.
- The common lesions are ventricular septal defects, patent ductus arteriosus, atrial septal defect, pulmonary stenosis, fallot's tetralogy.
- Associated extra-cardiac lesions are present in 30 % of cases.
- Ultrasound examination of the fetal chest:
  - Four-chamber view of the heart: View at right angles to the longitudinal aspect of the fetal spine. That view demonstrates arrhythmias.
  - M-mode tracings of different cardiac chambers or structures.
  - Doppler color-flow mapping. To define the pattern of the blood flow.

- Fetal Echocardiography

Single umbilical artery

- May be a single anomaly
- Possible associated malformations

- ✖ Esophageal atresia,
- ✖ Imperforate anus,
- ✖ Trisomy 18 syndrome.

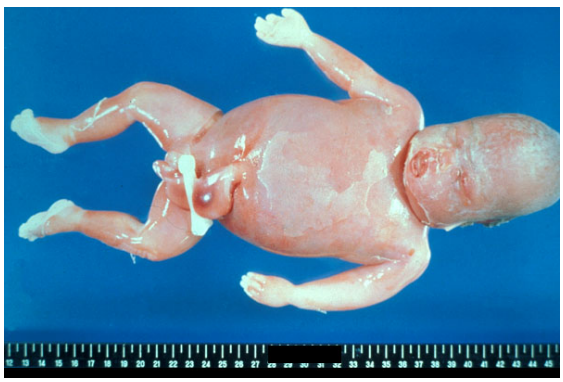


Diaphragmatic Hernia

- Pulmonary hypoplasia is a common serious associated anomaly.
  - ✖ Chromosomal anomalies are commonly encountered
- Antenatal diagnosis by ultrasound (cystic spaces within the chest).
- Presentation at birth:
  - ✓ Respiratory distress, scaphoid abdomen, displaced apex beat.
  - ✓ Radiological examination: intrathoracic bowel shadows

Hydrops Fetalis

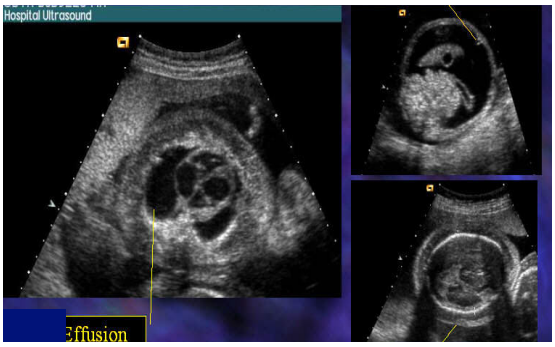
- Hydrops fetalis is excessive fluid accumulation within the fetal soft tissues (tissue edema) and body cavities (effusions).
- Ultrasound features of full blown hydrops:
  - ✓ Increased skin thickening: > 5 mm.
  - ✓ Placental thickening: > 4 cm.



- ✓ Body cavities: Significant pleural and pericardial effusions and ascites.

Causes of fetal hydrops:

- ➢ Immune hydrops fetalis.
  - ✓ Due to chronic intrauterine anemia. The well-known example is Rh isoimmunization.
- ➢ Non-immune hydrops fetalis: Generally it has a high incidence of mortality.
  - ✓ Fetal cardiac arrhythmias e.g. supraventricular tachycardia. Due to heart failure.
  - ✓ Fetal structural cardiac anomalies e.g. hypoplastic left heart, due to heart failure.
  - ✓ Pulmonary hypoplasia
  - ✓ Renal dysplasia.
  - ✓ Hypoproteinaemia
  - ✓ Congenital nephrosis.



  - ✓ Intrauterine infections: due to chronic intrauterine anemia e.g. toxoplasmosis, rubella, cytomegalovirus infections, congenital hepatitis, parvovirus infection.
  - ✓ Chromosomal abnormalities: e.g. Turner's syndrome, trisomy 18 or 21.
  - ✓ Congenital hematological disorders: e.g. thalaaemia.
  - ✓ Twin-to-twin transfusion.

Down's syndrome

- It is Trisomy 21 syndrome.
  - Incidence: general incidence is 1:600. The incidence rises with increase of maternal age.
    - 1:365 at 36 years and 1:40 at the age of 40 years
- Neonatal features:

- Head: Flat face and flat occiput, third fontanelle, upward slanted palpebral fissure, inner epicanthal folds and simply formed ears, nose: small, flat nasal bridge, mouth: small and the tongue protrudes.
- Neck: short, broad. Loose folds in posterior neck.
- Hands: simian single palmar crease, short metacarpals and phalanges. Hypotonia.
- Short humerus and femur
- Heart: High incidence of cardiac defects e.g. artrioventricular canal defect.
- Increased incidence of leukemias
- Gastrointestinal: Duodenal atresia, Hirschsprung's disease.

Antenatal Diagnosis of Down's syndrome:

- **First trimester**
  - ✓ Increased Nuchal Thickness
  - ✓ PAPP-A
  - ✓ Failure to detect the nasal bone

When the nasal bone line appears as a thin line, less echogenic than the overlying skin, it suggests that the nasal bone is not yet ossified, and it is therefore classified as being absent [11-13^{+6} weeks]

- **Second Trimester Screening**
  - ✓ The Triple Test
  - ✓ The Quad Test:
    - Triple Test + Dimeric Inhibin A (DIA)
- Integrated first and Second Trimester Screening
- Diagnostic procedures MUST involve genetic testing of samples obtained from the baby