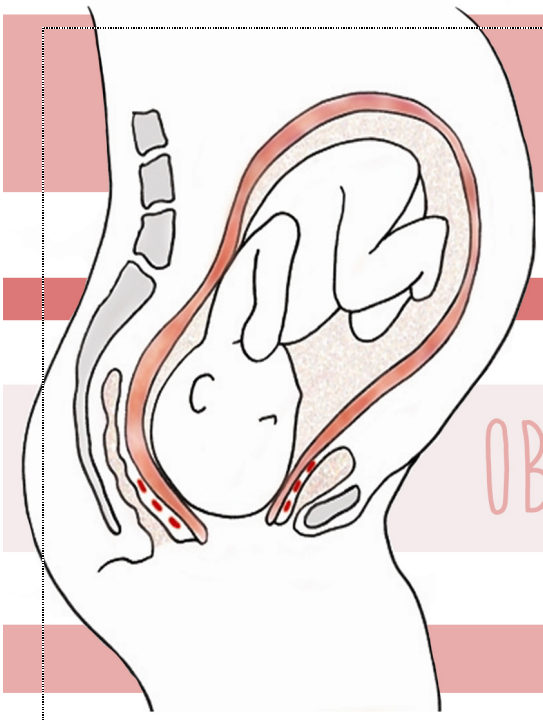

OBSTETRICS AND GYNECOLOGY

(3) Prenatal Testing

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

Objectives:

- Aims of pre-testing
- Importance
- Who is tested
- Routine testing
- US Finding
- Genetic testing

THE SLIDES WERE NOT PROVIDED BY THE DOCTOR , THIS WORK ONLY CONTAINS THE NOTES THAT WERE SAID BY THE DOCTOR DURING THE LECTURE

Routine Tests for any pregnant woman

The Aim of Prenatal-testing = insure maternal and fetal well being

- **Routine tests for all pregnant ladies include:**
- **CBC**
- **Hb**
- **WBC count**
- **Blood grouping and RhD → if the baby was RH+ve you will give Anti-D at 28 wks and after Delivery**
- **Random and fasting blood sugar**
- **Syphilis**
- **Rubella IgG→ if positive it means the mother was vaccinated at childhood**
- **Hepatitis B and C → because if the mother was a carrier there are precautions during and after delivery that need to be taken**
- **Toxoplasmosis**
- **HIV**
- **PAP Smear → in western countries its Routine because sometimes you can't detect the cervical lesions until pregnancy**
- **Routine US**
- **Urine test for asymptomatic bacteruria, and this is particularly important in sickle cell patients (should be done more than once)**
- **GBS detecting swab at >=36 to prevent GBS after Delivery**

US:

- **Fetal US is best done at 24wks**
- **US is a major tool for Dx and is important for :**

1. **Confirming viability**
2. **R/O ectopic pregnancy**
3. **Confirming the pregnancy date :**
   A. **1st trimester by the CRL (crown ramp length)**

B. 2nd trimester (at 24wks) by assessing femur length , Biparietal diameter ,Head circumference. **Week 24** is also important in discovering fetal anomalies officially and medicolegally (but you still can detect some anomalies such the frog sign of anencephaly during the tenth week)

4. Placental localization:

- by assessing its relation to the bladder .
- You want to see if its low lying→ this is important in Hx of C.S to detect any placenta previa or acreta

5. Serial US to follow up with the fetal growth especially in the suspicion of (IUGR,DM , IUFD,PED)
6. Placental function (Biophysical Profile and Doppler)

1st Trimester US:

- 1st trimester US is of extreme importance in:

1- Multipregnancies→ to detect the chorionicity mono vs. dizygotic i.e if you see two sacs this is not mono its dizygotic n.b in monochorionic there won't be thick separate sacs , usually their separating membrane is paper thin

2- Screening for chromosomal abnormalities :

- most important sign is the nuchal translucency (seen at **11-13 wks and 6 days** of pregnancy thickness behind the cervical spine) → timing is very important because if you do it after it will you won't see it
- Nuchal translucency can be in a normal karyotype in 5-10% of fetuses , that's why the next step after the US is to do karyotyping by either amniocentesis or NIVT
- Can be associated with cardiac defect , exomphalos and diaphragmatic hernia

2nd Trimester US:

- In the 2nd trimester you use the fetal biometry → HC, BPD,AC,FL → for fetal growth
- Amniotic fluid volume is also done to assess the placenta and exclude Fetal anomalies → make sure the needle is inserted in a space away from the fetus and the cord
- Imp for OSCE → they may get US picture for the 4 biometry measures

Tests for placental Function:

1- Biophysical profile → US (breathing movement by assessing the chest rise, tone (flexion/extension), movement (body/limbs), amniotic fluid volume) + CGT

- Each is scored a 0 or 2 not 1 and a maximum score of 10
- When low go ahead and do the Doppler

2- Doppler: important for (OSCE)

- Done high risk pregnancy DM , IUGR , PET ..
- They usually measure a free loop of the umbilical cord to assess the flow in it, Results either: normal , reduced EDF , absent EDF, Reversed (this would kill the fetus in 2-4hours)

Chromosomal anomalies:

1- In the past they used to rely on the maternal age the older the higher the risk

2- The current internationally medico-legally method used to detect fetal abnormality = **1st trimester screen by biochemical (PAP-A and B-HCG) screening test at 10 weeks** not before .. Why ?

- To council for Early abortion
- Less abortion complications at later stages
- Insurance and religious issue → they don't abort after 14 weeks

- The two most important tests are:

1- **PAPA → low levels** usually 0.5 multiple of the median (MOM) between 8 to 14 wks and usually in IUGR, Abortion, pre-eclampsia

2- **B-HCG** increases by **two folds**

- →Triple and quadruple tests of the second trimester are no longer done

3- US :

- **Nuchal translucency → the most important sign to look for**
- Nasal bone flatness → in down syndrome which will be absent in first trimester along with facio-maxillary angle which will be flat
- Ductus venous blood flow
- Tricuspid regurgitation→ when seen in first trimester HUGE sign of chromosomal anomaly → why do they get this and the ductus venosis problem ? due to the low tone seen in down syndrome

Important: International figures in down syndrome:

- **General risk of down syndrome is 1:1000**
- **Risk at 35y/o mother 1:365**
- **Risk at 40 y/o mother 1:109**
- **Risk at 45 y/o mother 1:32**
- **Risk of recurrence is 1% which is 0.75 higher than the maternal age related risk**
- **In parental aneuploidy the risk is 30%**

Genetic testing:

A- **Amniocentesis:**

- When an US shows abnormal translucency the next step is amniocentesis
- It could be diagnostic → **16-20 wks** → why not before 16 wks? High risk of club foot
- Indication :
  - genetic and chromosomal analysis
  - NTD by assessing the AFP levels
  - DNA and assessing inherited disorders like muscular dystrophy
  - RH iso-immunization patients by assessing bilirubin level
  - L/S to assess the fetal lung maturation
- It could be **therapeutic**→ in **twin to twin transfusion** at any time to reduce the fluid →because they have severe polyhydramnios states and maternal distress
- Its US guided
- Must be away from the placenta and fetus to avoid complications such as → bleeding , fetal head injury
- **Anti-d is only given if the mother is RH-ve**
- Risks of the procedure include → leaking of the amniotic fluid, abdominal pain, vaginal bleeding, infection, umbilical and fetal head injury, **clubfoot (if done before 16 wks)**, when used therapeutically sudden decompression may cause abruption placenta
- Risk of **abortion 1:200**

B- **Chordocentesis (Percutaneous Umbilical Cord Blood Sampling) :**

- Withdraw blood from the umbilical cord at the placental insertion
- Indications
  - Fetal DNA
  - Fetal HB
  - Fetal Intrauterine blood transfusion→ in RH iso-immunization
  - If you suspect infections Such as CMV
  - Not usually used for chromosomal assessment
- Risk

- IUFD
- Fetal Bradycardia → touching any vessel in the cord → umbilical artery spasm can lead to immediate death and so an emergency C.S is needed to save the fetus

C- Chorionic Villous Sampling (CVS) :

- Sampling of the **cytotrophoblasts**
- Done between **10 to 14 wks** → not before because no cells are detected and not after because it's too risky
- It can be done transvaginally
- You aspirate at the chorionic side at the chorionic villous
- Indications :
- Karyotyping
- For advanced age when first trimester biochemical screening was bad or US abnormal
- Family Hx of chromosomal anomaly
- Abnormal parental karyotyping
- Can be done in other genetic anomalies like :
- Single gene disorders
- Cystic Fibrosis
- Duchenne Dystrophy
- Osteogenisis imperfecta
- Not used anymore because its invasive and is substituted with Free Fetal DNA in Maternal Blood
- Risk
  - abortion 1:100
  - Risk of contamination with maternal residual cells
  - Mosiacism →needs amniocenteses
  - **Limb reduction when done before 9 wks**
  - Bleeding
  - Ruptured membranes
  - Spotting
  - Rh -ve give anti-D

D- Free Fetal DNA and RNA in Maternal Blood (non-invasive testing)

- Venous maternal sample is withdrawn
- The current method Internationally used
- They can determine
  - Fetal DNA type and age
  - Gestational age
  - Number of pregnancies

- Number of abortions
- Chromosomes of the fetus
- Gender of the fetus
- DNA age
- RH and blood type
- Fetal age from the DNA age
- Genetic disease Dx by separating the fetal cells found in the mothers sample

- Done at **10 wks** by doing direct PCR
- You only require **10 cc of maternal blood to do it a 3-6% of the maternal blood cell is fetal**
- Results are out within 10 day
- Not recommended in infections as we usually assess them by following the IgG/ IgM levels in the blood
- Expensive
- > 300 genetic disease that can be detected examples include Cystic Fibrosis, inborn errors of metabolism, acondroplasia, myotonic dystrophy, huntingtons, early onset dystonia, congenital adrenal hyperplasia

prenatal laboratory testing:

- Asymptomatic bacteriuria screening and treating → significantly reduces the risk for pyelonephritis and preterm delivery.
- Rh negative women should receive RhO(D) immune globulin (RhO-GAM) at 28 weeks of gestation and postpartum and at any time when sensitization may occur (e.g., threatened abortion or invasive procedures such as amniocentesis and chorionic villus sampling).
- Rubella vaccination is contraindicated during pregnancy, and pregnant women who are found to be seronegative should be vaccinated immediately postpartum.
- Syphilis testing is mandatory→ Early diagnosis and treatment of syphilis reduce perinatal morbidity.
- Hepatitis B → Women who test negative for surface antigen and are at high risk for hepatitis B infection (e.g., health-care workers) should receive vaccination before and during pregnancy.
- Infants born to women who test positive for hepatitis B surface antigen should receive both hepatitis B immune

globulin (HBIG) and hepatitis B vaccine within 12 hours of birth, followed by two more injections of hepatitis B vaccine in the first 6 months of life.

- ➢ HIV Voluntary and confidential counseling and testing should be offered and documented in the medical record.
- Diagnosis and treatment significantly reduce the risk for vertical transmission.
- ➢ Other tests, such as screening for sexually transmitted infections like gonorrhea and chlamydia, are generally considered routine.
- ➢ All pregnant women at high risk for tuberculosis should be screened with a purified protein derivative (PPD) skin test when they begin prenatal care.
- ➢ Information about nutrition, behavioral changes to expect, and the benefits of breastfeeding should be provided as prenatal care progresses.

Laboratory tests	Check complete blood count, urinalysis, type and screen, rubella, syphilis, hepatitis B, HIV, cervical cytology; screen for gonorrhea, chlamydia, and diabetes in selected populations. Consider thyroid-stimulating hormone.

MCQ'S

A 38-year-old woman attends for antenatal booking at 15 weeks gestation. This is her first pregnancy. She requests the most reliable prenatal test for Down's syndrome. Which is the SINGLE MOST appropriate test? Select ONE option only.

A. Amniocentesis & chromosome analysis
B. Chorionic villous biopsy
C. Serum alpha fetoprotein
D. Ultrasound for nuchal fold thickness
E. Urine ß human chorionic gonadotrophin (ß HCG)

For mistakes or feedback

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You can find more notes on this topic in the (Management of Labor) teamwork.