
Invasive and Non-Invasive Prenatal Testing
By: Michelle Wilson Ed.D, RDMS, RDCS
nanoschematic
Advantages
Disadvantages
Lets Start from the Beginning


Why Prenatal Testing is Important
Nuchal Translucency
Maternal Serum Screening
Chorionic Villus Sampling
Lets Start from the Beginning
Amniocentesis
Chorionic villus sampling
Umbilical blood sampling
Maternal blood testing
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Maternal Serum Screening

- Used to screen for chromosomal abnml and neural tube defects
- Calculates a woman's risk based on the levels of three or more substances found in the blood or the
- Indicates whether a woman is likely to be carrying an affected fetus
- Further testing is recommended to confirm a positive result

Why Prenatal Testing is Important

- Important to both mother and unborn child
- Used by doctors to learn how the baby is growing and how the mother is doing
- Also used to detect any problems during the gestational period
- Important to have blood and urine tested
    - Look for antibodies:
        - immune to chicken pox and rubella. If, during pregnancy, a mother is infected with either for the first time, birth defects can occur

Advantages

- Sometimes unborn baby's problems can be rectified before it is born
- Gives parents the choice to terminate pregnancy if they cannot take on a child who has genetic abnormalities
- Prepares parents for a baby with genetic abnormalities

Disadvantages

- Anxiety can be caused by the results
- Various religions may be against abortion
- Test are very expensive
  - Not all insurance companies are willing to cover expenses

Remember these are screening tests:

Tests are optional
Neg result does not mean baby is healthy
Pos result does not mean baby is problamatic
Offered to everyone

Social and Ethical Implications

- Prenatal testing cannot determine how severe the mutation is
- The ethical dilemma involves the decision to continue or to end a pregnancy without having knowledge of the severity of the disorder
- Another issue that occurs is whether a newborn should be tested for disorders that we cannot treat
- Test may show false positive or false negative for some diseases which may cause parents to make irreversible decisions
- Genetic discrimination occurs when an individual or an organization treats a person differently because of real or perceived genetic difference

Why Prenatal Testing is Important

- If problems are not treated or detected, can cause the fetus to grow poorly
- Prenatal testing is important to both maternal health and the health of the fetus
- It allows the doctor to treat pregnancy related problems and to know whether to expect problems as a pregnancy progresses

Genetic Diversity

Lets Start from the Beginning

- Amniocentesis
- Chorionic villus sampling
- Umbilical blood sampling
- Maternal blood testing
- Nuchal Translucency

- MSAFP-protein produced in liver
- HCG-placental hormone (levels drop btwn 10-20 weeks)
- Estriol (E3)-placenta makes, precursors to adrenal and liver

When Do These Occur

First Trimester: Cell-free DNA test, Chorionic villus sampling, Cystic Fibrosis carrier, Nuchal Translucency, First Trimester Screen

Second Trimester: Maternal blood screen, Amniocentesis, Glucose screen and Anatomical survey

Third Trimester: GBS (groub B strep), non-stress tests or fetal heart monitoring

Cell-free DNA test: tested through maternal blood. Evals fetal DNA for genetic conditions. (around 10 wks)

Chorionic Villus sampling: another slide

Cystic fibrosis carrier: Can be done at any time. Looks for gene causing CF specifically. CF alters digestion and breathing.

First Trimester screen: another slide

Ultrasound: another slide

Second Trimester 13-28 Weeks

Maternal blood screen: blood sample from the mother is taken to check for birth defects the baby might have. This test is performed during weeks 15-20 of pregnancy and measures the alpha-feto protein, PAPP- A, human chorionic gonadotropin (hCG) and estriol

Amniocentesis: another slide

Glucose screen: this test is for the mother to see if she has developed gestational diabetes. Takes place during weeks 24-28.

Ultrasound: takes place around 20 weeks and it checks the growth, anatomy as well as if any defects are present.

Third Trimester

GBS (group B streptococcus): is a bacteria that causes infection and can be harmful to the baby as it can be passed down to the baby during labor. Most people are unaware that they carry this bacteria. 25% of pregnant women carry this bacteria. This test is done during weeks 35-37, and it is a painless test. A swab is taken from the rectum and vagina, which is then sent to the lab

Non-stress test (NST): a screen of the fetal heart, and checks the health of the baby.

First Trimester: Serum and Ultrasound

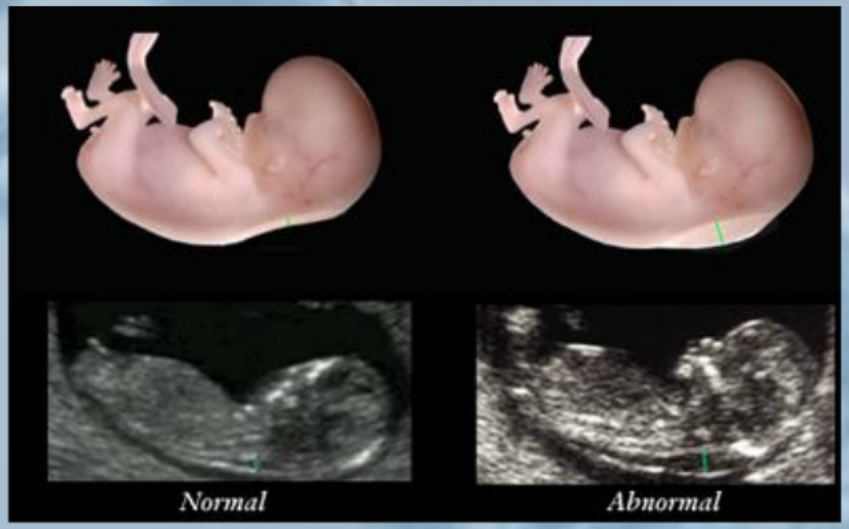
Nuchal Translucency
Anatomy Scan
Free beta hCG and pregnancy-associated plasma protein A (PAPP-A)

In the first trimester, low levels of this protein are seen in Down syndrome pregnancies.

Increased NT

Physiological nuchal fluid accumulation btw 11-14 wks

- **Turners syndrome, T21, T18, T13**
  - **Euploid-structural anomalies, skeletal defects, diaphragmatic hernia, tri regurg, adrenal hyperplasia, Noonan syndrome**
- **High risk of miscarriage or fetal death**
- **Higher risk of orofacial cleft defects**



Quad Screen or Triple Screen Tests

Performed between 15-20 weeks

- hCG, AFP, Inhibin A and Estriol

Alpha-fetoprotein is made in the part of the womb called the yolk sac and in the fetal liver, and some amount of AFP gets into the mother's blood. In neural tube defects, the skin of the fetus is not intact and so larger amounts of AFP is measured in the mother's blood. In Down syndrome, the AFP is decreased in the mother's blood, presumably because the yolk sac and fetus are smaller than usual.

Estriol is a hormone produced by the placenta, using ingredients made by the fetal liver and adrenal gland. Estriol is decreased in the Down syndrome pregnancy. This test may not be included in all screens, depending on the laboratory.

Human chorionic gonadotropin hormone is produced by the placenta, and is used to test for the presence of pregnancy. A specific smaller part of the hormone, called the beta subunit, is increased in Down syndrome pregnancies.

Inhibin A is a protein secreted by the ovary, and is designed to inhibit the production of the hormone FSH by the pituitary gland. The level of inhibin A is increased in the blood of mothers of fetuses with Down syndrome.

Once the blood test results are determined, a risk factor is calculated based on the "normal" blood tests for the testing laboratory. The average of normals is called the "population median." Test results are sometimes reported to doctors as "Multiples of the Median (MoM)." The "average" value is therefore called 1.0 MoM. Down syndrome pregnancies have lower levels of AFP and estriol, so their levels would be below the average, and therefore less than 1.0 MOM. Likewise, hCG in a Down syndrome pregnancy would be greater than 1.0 MoM. In the serum screening, the lab reports all results in either this way or as a total risk factor calculated by a software program.

Currently, pregnant women are eligible for amniocentesis or CVS if they are ³35 years, have a positive prenatal screening test, family history of genetic disease or certain ultrasound findings.

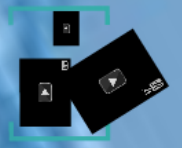
	Amniocentesis	CVS
Performed	15 -17 wks (ideal) - but available up to 22 wks[1]	11 - 13 wks[2]
Sample	Amniotic fluid	Placental villi
Results available	2 - 3 wks	2 - 3 wks
Miscarriage rate	0.01 - 0.5%[3]	1%
Advantage	- Accurate - Widely available - Tests for NTDs	- Accurate - 1st trimester test – earlier results
Disadvantage	- 2nd trimester test - later results	- Availability varies - Does not test for NTDs - ↑rate of repeat procedures due to ambiguous results

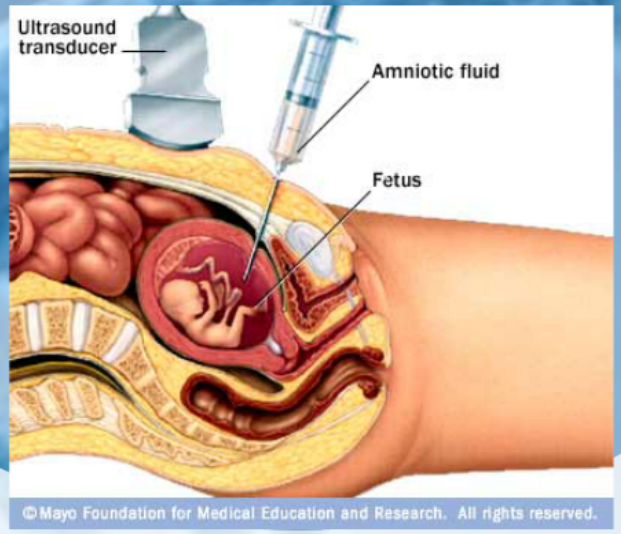
1. Amniocentesis may be available later than 22 weeks in certain circumstances.
2 The timing of CVS may vary between centres.
3 Recent studies suggest that miscarriage rate is lower than 1 in 200 (0.5%).

http://www.health.gov.on.ca/english/providers/program/child/prenatal/images/prenatal_diagnostic_testing.gif

Amniocentesis

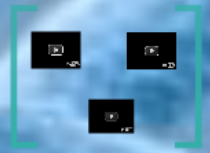
- Draws a sample of amniotic fluid
- Tested to check for fetal infections, diagnostic purposes (lung maturity) or chromosome abnormalities
    - Also used to treat polyhydramnios-

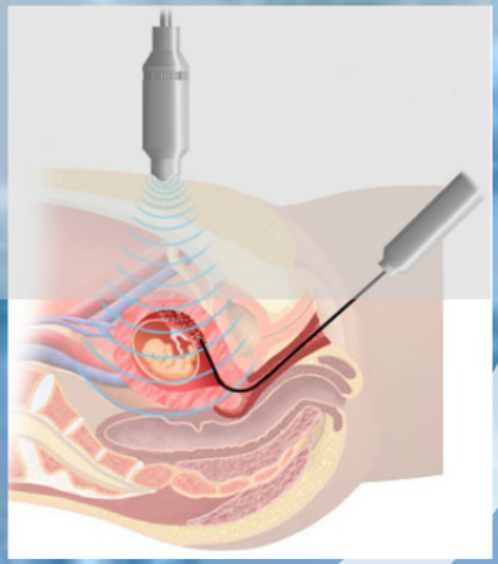




Chorionic Villus Sampling

- Tiny finger-like units that make up the placenta and provides nutrients to fetus
- Alternative to amniocentesis
- Removes some of the villi and tests for chromosome abnormalities
- Performed earlier than amniocentesis
- Risks of CVS are higher



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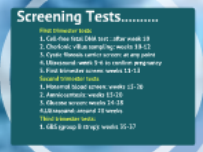
Umbilical Cord Sampling

PUBS or Chordocentesis

- Obtains fetal blood
- Check's for chromosome abnormalities or any other concerns such as a low platelet count or a thyroid condition
- The only way to confirm if a fetus is suspected to be anemic or have a platelet disorder
- Risks include possible miscarriage or infection

Fetal blood vs amniotic fluid


Screening Tests.....


YouTube

Screening Tests.....

First trimester tests

1. Cell-free fetal DNA test : after week 10
2. Chorionic villus sampling: weeks 10-12
3. Cystic fibrosis carrier screen: at any point
4. Ultrasound: week 5-6 to confirm pregnancy
5. First trimester screen: weeks 11-13

Second trimester tests

1. Maternal blood screen: weeks 15-20
2. Amniocentesis: weeks 15-20
3. Glucose screen: weeks 24-28
4. Ultrasound: around 20 weeks

Third trimester tests:

1. GBS (group B strep): weeks 35-37