Screening Tests

Testing is offered to all pregnant women. All testing is voluntary. You may choose to do no testing.

Sequential Screen is a noninvasive testing performed in 2 parts. Part 1 can be done between 11-14 weeks of pregnancy (12 weeks is best). An ultrasound scan is performed to measure the amount of fluid at the back of the baby's neck (nuchal translucency or NT) and a maternal blood sample is drawn. Results of Part 1 are usually available within 7-10 days but will only be reported to you if abnormal.

Part 2 is second maternal blood sample done between 15-20 weeks of pregnancy (16-18 weeks best). Results are usually available after 7-10 days. The combination of the NT sono and 2 blood tests will detect about 90% of babies with Down syndrome and Trisomy 18 and about 80% of babies with open neural tube defects (ONTD). The false positive rate is 4%.

If we are unable to obtain a NT measurement, you will be offered **Serum Integrated Screen (SIS).** SIS is a noninvasive test which consists of 2 maternal blood tests; the first is done between 10-14 weeks and the second between 15-20 weeks (16-18 weeks best). There is one result only, which is usually available 7-10 days after the second blood sample. SIS will detect about 87% of babies with Down syndrome and Trisomy 18 and 80% of babies with ONTD. The false positive rate is 5%.

If you begin care after 14 weeks, you will be offered **Quad Screening, also known as AFP4 Screening.** Quad screening consists of 1 maternal blood sample drawn between 15-20 weeks of pregnancy (16-18 weeks is best). Results are available between 7-10 days. This test will pick up 80% of babies with Down syndrome and Trisomy 18 and 80% of babies with ONTD. The false positive rate is 5%.

If any of the above tests are abnormal, (including Part 1 of Sequential Screen), you will be offered the option of diagnostic testing such as amniocentesis or CVS (Chorionic Villus Sampling). In some cases, it may also be appropriate to opt for further screening such as NIPT (non-invasive prenatal testing) or a detailed (level 2) ultrasound.

If you have had normal CVS or NIPT, you will also have **Maternal Serum AFP Screening** (MSAFP). This is to screen for ONTD which CVS and NIPT do not test for. MSAFP consists of 1 blood test done between 15-20 weeks o f pregnancy (16-18 weeks best).

Remember, if you do any of the above screening tests and you get a positive result, it does not mean that there is anything wrong with your baby. All of the tests have a false positive rate.

Diagnostic Testing

You will be offered diagnostic testing if any one of the following is true:

- You are age 35 or over or will turn 35 during this pregnancy
- You have a personal or family history of certain diseases
- You have an abnormal screening test

Most insurance companies will cover these tests if you fall into one of the above categories. Diagnostic testing is voluntary. You may choose to do a screening test or no testing at all.

Amniocentesis will detect chromosomal disorders such as Down syndrome, Trisomy 18 and Trisomy 13. It can determine the presence of ONTD, such as spina bifida and hydrocephalus. It can also be used to test for other genetic diseases such as cystic fibrosis, sickle cell disease or Tay Sachs disease if your baby is at increase risk of these.

Amniocentesis is done between 15-20 weeks (16-18 weeks best). Using ultrasound, the doctor puts a thin needle in to the uterus and collects a small sample of amniotic fluid (the fluid the baby is in). This fluid and the baby's skins cells which it contains are analyzed. Results take approximately 10 days.

Amniocentesis is an invasive procedure so it has a small (1in200) risk of miscarriage or infection, which could result in the loss of your baby.

Chorionic Villus Sampling (CVS), like amniocentesis, will detect chromosomal disorders and certain genetic diseases. It does not detect ONTD, so if you opt for CVS, you will also need to have a blood test at 16-18 weeks called MSAFP.

CVS is done earlier than amniocentesis, usually between 12-14 weeks. The chorionic villi are fetal tissue present early in pregnancy which will become the placenta. Under ultrasound, a tiny piece of this tissue is collected, either by inserting a thin needle through the abdomen (similar to amniocentesis) or by inserting a thin tube through the cervix (via the vagina). This tissue is then analyzed for chromosomal abnormalities or genetic diseases as indicated.

CVS is an invasive procedure so it carries a risk of miscarriage (1in100) or infection.

CVS is not done in Ithaca, Currently it is being offered in Syracuse, Rochester or Elmira. Our office will assist in scheduling and referral.

Both amniocentesis and CVS may not be successful if not enough sample is collected, if the cells fail to grow, or if the sample is contaminated by maternal cells.

Non Invasive Prenatal Testing (NIPT) is a screening test that detects greater than 99% of pregnancies with Down syndrome, greater than 98% of pregnancies with Trisomy 18 and about 65% of pregnancies with Trisomy 13. It will also detect abnormalities of the fetal sex chromosomes such as Turner syndrome. It will also tell the sex of your baby, if you choose to know. False positive rate is 1% (0.2% for Down syndrome).

It does not detect ONTD, so MSAFP will also be needed to be performed.

NIPT consists of 1 maternal blood sample done after 10 weeks of pregnancy; the test measures the relative amount of free fetal DNA in the mother's blood. Results are available within 2 weeks after blood is drawn. It is a non invasive test; therefore, it has no risk of miscarriage.

The test is sometimes not successful when not enough free fetal DNA is detected. In this case, you can opt for repeat sample, invasive testing (amniocentesis) or screening test (depending on gestational age).

If NIPT indicates an abnormality, confirmatory testing such as amniocentesis is recommended.