UNM HOSPITALS

PRENATAL TESTING FOR SOME BIRTH DEFECTS

While most babies are born healthy, any woman can have a baby with a birth defect. There are different ways to find birth defects during pregnancy. Some tests will tell you for sure if a baby has certain problems, others will not. No test can find all birth defects. It is your choice whether or not to have any testing. In deciding about these tests, you should ask yourself if you would want to know before birth if your baby has a birth defect.

SCREENING TESTS are safe, but not perfect. They will **not** tell you for sure if a baby has a birth defect. Most women with "low risk" screening test results have healthy babies. Many women with "high risk" results also have healthy babies. However, women with "high risk" results will be offered more tests.

WHAT FOR?	HOW DONE	WHEN DONE	PROS	CONS
Down syndrome Trisomy 18 Some spine and brain defects	Blood tests and Ultrasound	11 to 21 weeks Sooner is better	No risk to baby Less expensive than diagnostic tests	Some false alarms Follow-up test offered if abnormal Can miss some babies with these birth defects

DIAGNOSTIC TESTS will tell you for sure if a baby has **certain** birth defects. Diagnostic tests are not as safe as screening tests. If you think you might want a diagnostic test, you should meet with a genetic counselor to learn more.

WHAT FOR?	HOW DONE	WHEN DONE	PROS	CONS
Down syndrome and similar conditions Some spine and brain defects	Doctor takes cells from the placenta (CVS) OR some fluid from around the baby (amniocentesis).	11 weeks and later	Definite information about Down syndrome and some other birth defects	Small chance of miscarriage More expensive than screening tests. Not all types of birth defects can be found
Some other physical birth defects	Ultrasound	11 weeks and later	No known risks to baby	Not all types of birth defects can be found

PRENATAL SCREENING: WHAT ARE WE LOOKING FOR?

While most babies are born healthy, any woman can have a baby with a birth defect or serious medical problem. You may want to have tests which can tell you about your chances to have a baby with certain birth defects or certain genetic conditions. This page tells you about **some conditions** that can be found with screening. Whether you have these tests or not is up to you.

DOWN SYNDROME

- Leads to learning problems (mental retardation), health problems, and sometimes birth defects.
- Children with Down syndrome are usually able to do many things that other children can. They do need extra medical care and help with learning.
- Children do not usually die from Down syndrome
- Most children with Down syndrome are born to young, healthy parents.
- Most children with Down syndrome are born to parents with no family history of Down syndrome.

TRISOMY 18

- A serious combination of birth defects and brain damage
- Most babies with Trisomy 18 die before birth or soon after. Those that live are severely disabled.
- Most children with Trisomy 18 are born to young, healthy parents.
- Most babies with trisomy 18 are born to parents with no family history of the condition.

NEURAL TUBE DEFECTS (NTDs)

- Happen very early in pregnancy. The neural tube develops into the baby's brain and spinal cord. If the neural tube does not form normally, this leads to birth defects of the backbone, spinal cord and/or brain.
- Some NTDs can be fixed by surgery after birth. Even with surgery, many children with NTDs have physical disabilities and/or learning problems. Some NTDs are so serious that babies die from them
- Most babies with NTDs are born to healthy parents with no family history of the condition.

CYSTIC FIBROSIS (CF)

- A serious childhood disease that leads to lots of lung infections, and growth problems. CF does not affect learning ability. CF shortens a person's life.
- CF can be treated, but not cured.
- Most babies with CF are born to people with no family history of the condition.
- Both parents have to be CF carriers in order for a child to have CF.
- The chance of being a carrier depends on your family history and ethnic background
- A blood test that can be done now will find most, but not all, CF carriers
- All babies born in New Mexico are screened at birth for CF.

If you have a **close relative** with a birth defect, genetic disease or mental disability, you might want to talk to a genetic counselor about your risks and testing options. This can be done at the UNM Prenatal Diagnosis and Genetics Clinic (505-272-6611).

February, 2009

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CONSENT (PERMISSION) FOR PRENATAL TESTING

_____ I have read the information and **I want** to have a screening test.

- ❖ I understand that screening tests will not tell me for sure if my baby has a birth defect.
- ❖ I understand that a “high risk” test result does not mean my baby has a birth defect. It means I will be offered more tests.
- ❖ I understand that not all birth defects can be found with this test.

_____ I have read the information and **the only testing I want** at this time is ultrasound.

_____ I have read the information, and **I want** to talk with a genetic counselor about my testing options, including amniocentesis and CVS.

_____ _____
Patient signature Date

_____ _____
Informant signature Date

LABEL

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