OLATHE OBSTETRICS AND GYNECOLOGY, P.A.
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PRENATAL DETECTION OF CONGENITAL MALFORMATIONS

We are now able to test for certain abnormal developments of the fetus during pregnancy. These tests are to help your pregnancy proceed as smoothly as possible, but no test is 100% perfect and will not guarantee that all babies will be normal. This information is not given to "scare" you but simply to let you understand the possible tests which are available to you in this pregnancy. If you have any questions, please ask us for information on any tests.

1. A blood test from the mother's arm for "neural tube defects" is an evaluation for abnormal development of the spinal cord. This is a group of the most common major congenital (at time of birth) malformations. This test is called "serum alphafetoprotein" and is specific for detecting spina bifida or anencephaly (incomplete development of head or spine), which occur in 2 in 1000 pregnancies. Also, unexplained high levels may be associated with inadequate fetal growth and stillbirths. Further testing would be recommended. Low levels may be indicative of Down syndrome, although this is not the primary test for Down syndrome. An abnormal result would only indicate that more testing is needed and would not by itself indicate an abnormal fetus. This test is available only between 15 and 20 weeks of your pregnancy. If your last menstrual period and due date are not certain, a sonogram may be necessary as the test result varies with the age of the fetus. This test is recommended by the American College of Obstetrics and Gynecology.

I: DO / DO NOT want the blood test to screen for neural tube defects. _____

2. Sonography (ultrasound). Recent advances in sonography allow the developing fetus to be evaluated for a variety of abnormalities including brain and spinal cord, bladder, kidney, heart and other findings. This test is frequently done in pregnancy for other reasons such as determining the due date and how well the baby is growing.

I: DO / DO NOT want sonogram to evaluate baby for abnormalities. _____

3. A blood test is available to screen for cystic fibrosis (C F) in the baby. C F frequently results in the child having complicated lung infections resulting in lifelong respiratory treatments and reduced life expectancy. The gene for C F (being a carrier of the disease) must be present in both the mother and father to be present in the baby. What are the chances of being a C F carrier or having a baby with C F?

The Chance of Being a C F Carrier Depending on Race / Ethnicity

Ethnicity / Race	Chance of Being a C F Carrier	Chance Both Partners are C F Carriers	Chance of Having a Child with C F
European, Caucasian Ashkenazi Jewish	1 in 29	1 in 841	1 in 3300
Hispanic American	1 in 46	1 in 2116	1 in 8400
African American	1 in 65	1 in 4225	1 in 16000
Asian American	1 in 90	1 in 8100	1 in 32000

If the test is positive for one parent, the other will need to be tested. If both parents are positive, referral to a specialist for further testing will be needed. The cost of this test as of April 1, 2003 is $250 but varies and may not be covered by insurance.

I: Would like a pamphlet providing more detailed information. _____

I: Would like to be tested. _____

I: Do not want to be tested. _____

4. A chromosomal test available for evaluation of the fetus' development is "genetic amniocentesis". A sonogram is done to determine the baby's position and then some of the water surrounding the baby is removed with a needle to send for testing. The tests are for chromosomal studies (such as Down syndrome) and also for neural tube defects. This is the most accurate test we have for a problem with the baby's genes or chromosomes. This test has the risk of causing miscarriage approximately 1 in 200 times. Cost for the sonogram, procedure and laboratory fees is approximately $2600. The test is limited to specific indications such as mother's age greater than 35, previous child with chromosomal abnormality, family history of genetic disease, or positive alphafetoprotein test.

I: DO / DO NOT want amniocentesis. _____

5. HIV / AIDS disease may be passed from the mother to her unborn child. We now have medicines that significantly reduce the baby's risk of developing HIV. Therefore, it is more important than ever to screen mothers for HIV. Since HIV / AIDS is now commonly passed with traditional intercourse, high risk factors such as I.V. drug use and prostitution are no longer the only risk factors. All women are recommended to be tested. No test is 100% accurate. Recent infection may be missed and repeat testing in three to six months is available.

I: DO / DO NOT want the HIV / AIDS blood test. _____

Patient Signature _____ Date _____

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BIRTH DEFECTS RISK ASSESSMENT QUESTIONNAIRE

Patient Name: _____

This questionnaire is for screening only. It does not guarantee the birth of a healthy baby.

SOME MATERIAL CHARACTERISTICS CAN AFFECT YOUR PREGNANCY:

1. Will you be age 35 or over when you deliver? **YES____ NO ____**
2. Have you had, or do you now have, epilepsy or seizures? **YES ____ NO ____**
3. Are you a diabetic? **YES ____ NO ____**
4. Could you and your partner be related (first cousins, etc.)? **YES ____ NO ____**

SOME HEALTH PROBLEMS ARE MORE COMMON IN CERTAIN ETHNIC GROUPS

5. Are you or your partner?

African American / Black **YES ____ NO ____**

If yes, have you or your partner been tested for sickle cell anemia?

YES ____ NO ____ DON'T KNOW ____

have you or your partner been tested for thalassemia?

YES ____ NO ____ DON'T KNOW ____

Greek, Italian, Middle Eastern, or Asian **YES ____ NO ____**

If yes, have you or your partner been tested for thalassemia?

YES ____ NO ____ DON'T KNOW ____

Eastern European (Ashkenazi) Jewish or French Canadian **YES ____ NO ____**

If yes, have you been tested for Tay Sachs disease?

YES ____ NO ____ DON'T KNOW ____

FAMILY HISTORY CAN ALSO BE IMPORTANT

6. Have you, your partner, or anyone in either of your families had any of the following?

Down Syndrome (Mongolism / Trisomy 21) or other chromosome problem	**YES ____**	**NO ____**
Neural tube defect (Opening in the spine, spina bifida, anencephaly)	**YES ____**	**NO ____**
Mental Retardation / developmental delay	**YES ____**	**NO ____**
Fragile X syndrome	**YES ____**	**NO ____**
Huntington disease	**YES ____**	**NO ____**
Cystic Fibrosis	**YES ____**	**NO ____**
Muscular dystrophy or other muscle or nerve problems	**YES ____**	**NO ____**
Hemophilia or any other bleeding disorder	**YES ____**	**NO ____**
Other genetic condition	**YES ____**	**NO ____**

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7. Have you, your partner, or anyone in your families had a birth defect? YES____ NO ____
(such as: cleft lip, blindness, deafness, hydrocephaly (water on the brain), etc.)

If yes, please describe: _____

8. Have you, your partner or any other family member been born with a heart defect? YES ____ NO ____

9. Do you or your partner have a history of three or more miscarriages or a stillbirth? YES ____ NO ____

If yes, was there a reason found for these miscarriages? YES ____ NO ____

SOME MEDICATIONS CAN AFFECT YOUR PREGNANCY:

10. Have you taken any of these medications while pregnant?

Seizure / epilepsy medications (Dilantin, Tegretol, etc.)	**YES ____**	**NO ____**
Lithium for depression	**YES ____**	**NO ____**
Accutane or other pills for Acne	**YES ____**	**NO ____**
Any other medications of concern	**YES ____**	**NO ____**

If yes, please describe: _____

11. Have you used any of the following while pregnant?

Alcohol	**YES ____**	**NO ____**
Cocaine / crack	**YES ____**	**NO ____**
Any other drugs that concern you	**YES ____**	**NO ____**

If yes, please describe: _____

Please discuss any "YES" answers with your physician / health care provider. In some cases, further evaluation by a genetic counselor may be suggested.

I have read all of the above questions carefully, and understand that this information is important for my health care providers to determine if my baby could be at an increased risk to have an inherited disease or birth defect. I also understand that 2-3% of babies are born with a birth defect. Many birth defects cannot be detected before birth and may occur with no family history.

Patient Signature: _____ Date: _____

Provider Signature: _____