

## Questionnaire for new obstetrical patients

1. What was the first day of your last menstrual period? \_\_\_\_\_  
 Are your periods regular? \_\_\_\_\_ How many days apart are your periods? \_\_\_\_\_
2. Do any of your parents, siblings, or children have diabetes or hypertension? \_\_\_\_\_
3. How many times have you been pregnant? \_\_\_\_\_ How many children? \_\_\_\_\_ Please list below  
 Name            birthdate    Hospital/Doctor    birthweight    vag or C/S?        Problems?

4. Do you have any allergies-to medications, latex, or seasonal?

5. List any surgeries or hospitalizations that you have had (including gyn).

6. Have you ever had any of the following medical problems?

	<u>Yes</u>	<u>No</u>		<u>Yes</u>	<u>No</u>
Kidney disease or			phlebitis/varicose veins	___	___
Frequent UTI's	___	___	Trauma or violence	___	___
Heart disease	___	___	Epilepsy/neurologic disease	___	___
High blood pressure	___	___	blood transfusion	___	___
Diabetes	___	___	Rh (D) sensitization	___	___
Asthma or Tuberculosis	___	___	Anesthesia complications	___	___
Anxiety or depression/postpartum depress	___	___	Uterine abnormalities	___	___
Psychiatric disease	___	___	Abnormal PAP smears	___	___
Hepatitis or liver disease	___	___	Infertility	___	___
thyroid disease	___	___	IVF/Assisted reproduction	___	___
Autoimmune disorder	___	___	Breast disease	___	___

7. Have you had any of the following symptoms during this pregnancy?

	<u>Yes</u>	<u>No</u>
Vaginal bleeding	___	___
Vaginal discharge	___	___
Abdominal pain	___	___
Urinary problems	___	___

8. Are you drinking alcohol during this pregnancy?    Yes \_\_\_    No \_\_\_
9. Do you smoke cigarettes?    Yes \_\_\_    No \_\_\_
10. Do you use recreational drugs?    Yes \_\_\_    No \_\_\_
11. Do you have any cats or do you eat raw meat?    Yes \_\_\_    No \_\_\_
12. Have you had any X-rays during this pregnancy?  
 (Ultrasounds/sonos are not X-rays)    Yes \_\_\_    No \_\_\_
13. Does your partner have genital herpes?    Yes \_\_\_    No \_\_\_
14. Do you have a history of STD's (gonorrhea, Chlamydia  
 Genital herpes, syphilis)?    Yes \_\_\_    No \_\_\_
15. Have you had a rash or viral illness since your  
 Last period?    Yes \_\_\_    No \_\_\_
16. Have you lived with someone or been exposed to TB?    Yes \_\_\_    No \_\_\_

Name: \_\_\_\_\_ Date: \_\_\_\_\_

1. Will you be 35 years or older when the baby is due? ..... Yes \_\_\_ No \_\_\_

2. Have you, the baby's father, or anyone in either of your families ever had any of the following disorders?

- Down Syndrome (mongolism)..... Yes \_\_\_ No \_\_\_
- Other chromosomal abnormality ..... Yes \_\_\_ No \_\_\_
- Neural tube defect, ie, spina bifida(meningomyelocele or open spine), anencephaly Yes \_\_\_ No \_\_\_
- Hemophilia ..... Yes \_\_\_ No \_\_\_
- Muscular Dystrophy ..... Yes \_\_\_ No \_\_\_
- Cystic Fibrosis ..... Yes \_\_\_ No \_\_\_
- Huntington disease..... Yes \_\_\_ No \_\_\_
- Congenital heart defects (heart defects at birth)..... Yes \_\_\_ No \_\_\_

If yes, indicate the relationship of the affected person to you or the father of your baby: \_\_\_\_\_

3. Do you or the father of the baby have a birth defect? ..... Yes \_\_\_ No \_\_\_

If yes, who has the defect and what is it? \_\_\_\_\_

4. In any previous marriages, have you or the baby's father had a child born dead or alive, with a birth defect other than those listed in question 2 above? ..... Yes \_\_\_ No \_\_\_

If yes, who had the defect and what was it? \_\_\_\_\_

5. Do you or the baby's father have any close relatives with mental retardation? ..... Yes \_\_\_ No \_\_\_

If yes, indicate the relationship of the affected person to you or the baby's father:

Indicate the cause, if known: \_\_\_\_\_

Have you or the baby's father ever been tested for "Fragile X " syndrome? ..... Yes \_\_\_ No \_\_\_

6. Do you, the baby's father, or a close relative in either of your families have a birth defect, any familial disorder, or a chromosomal abnormality not listed above? ..... Yes \_\_\_ No \_\_\_

If yes, indicate the condition and the relationship of the affected person to you or the baby's father: \_\_\_\_\_

7. In any previous marriages, have you or the baby's father had a stillborn child or three or more first trimester spontaneous pregnancy losses? ..... Yes \_\_\_ No \_\_\_

Have either of you had a chromosomal study? ..... Yes \_\_\_ No \_\_\_

If yes, indicate who and the results: \_\_\_\_\_

8. Are you or the baby's father of Jewish, French Canadian, or Cajun ancestry? ..... Yes \_\_\_ No \_\_\_

If so, have either of you been screened for Tay - Sachs disease? ..... Yes \_\_\_ No \_\_\_

If yes, indicate who and the results: \_\_\_\_\_

9. Has anyone in your (or the baby's father's) family ever had Canavan disease, Familial dysautonomia, Niemann-Pick disease, Fanconi anemia, Bloom syndrome, Mucopolipidosos Type IV, or Gaucher's disease..... Yes \_\_\_ No \_\_\_

10. Are you or the baby's father African American/black? ..... Yes \_\_\_ No \_\_\_

If so, have either of you been screened for sickle cell trait? ..... Yes \_\_\_ No \_\_\_

If yes, indicate who and the results: \_\_\_\_\_

Is there a family history of Sickle trait or Sickle cell disease?..... Yes \_\_\_ No \_\_\_

11. Are you or the baby's father of Italian, Greek or Mediterranean background? ..... Yes \_\_\_ No \_\_\_

If so, have either of you been tested for *b*-thalassemia (Mediterranean anemia)? ..... Yes \_\_\_ No \_\_\_

If yes, indicate who and the results: \_\_\_\_\_

12. Are you or the baby's father of Philippine or Southeast Asian ancestry? ..... Yes \_\_\_ No \_\_\_

If so, have either of you been tested for *a*-thalassemia? ..... Yes \_\_\_ No \_\_\_

If yes, indicate who and the results: \_\_\_\_\_

13. Excluding iron and vitamins, have you taken any medication or recreational drugs since being pregnant or since your last menstrual period? (including nonprescription drugs) ..... Yes \_\_\_ No \_\_\_

If yes, give name of medication/drug and time taken during pregnancy: \_\_\_\_\_

## **HIV Testing**

1. In December, 2007, New Jersey became the latest state to make HIV testing part of routine prenatal care for all pregnant patients.
2. HIV is the virus that causes aids and it is transmitted through unprotected sex, or through sharing of needles through injection drugs use.
3. A pregnant woman who has HIV can pass the virus to her baby before or during birth or by breast feeding. Women, especially, may not know they are at risk. Many women get HIV through heterosexual sex and are not aware that their partners have been at risk for HIV.
4. There are important benefits for a woman to knowing whether she has HIV or not. HIV is treatable. Treatment can prolong a woman's life and prevent transmission to her baby during pregnancy and birth.
5. Experts recommend that all pregnant women be tested for HIV regardless of whether a woman thinks she is at risk. If a woman is HIV positive, she can get treatment immediately. The CDC has found medical intervention during pregnancy can cut the mother-to-child HIV transmission from 25 percent to 2 percent.
6. All information about HIV testing and the results are kept confidential. In New Jersey, results are reported to the state Department of Health and Senior Services, where they are kept strictly confidential. Federal and state laws protect women with HIV from discrimination.
7. A woman has the right to refuse testing and she will not be denied care if she does so. If a woman refuses screening, NJ law requires that her newborn be directly tested shortly after birth.
8. If you do not refuse testing now, you will be tested around the time of your first visit and again around 28 weeks (so as to comply with New Jersey law).
9. If you refuse testing you will be asked again around 28 weeks. If you are not tested around 28 weeks, you will be asked again when admitted to the hospital. If you are still not tested, your baby will be tested (unless you document a religious objection).

I acknowledge that I have read the above.

Date \_\_\_\_\_ Signature \_\_\_\_\_

Name (please print) \_\_\_\_\_

### **For Patients who decline HIV testing, sign below**

I have decided to decline HIV testing \_\_\_\_\_

## CARRIER SCREENING FOR GENETIC DISEASES IN ASHKENAZI JEWS

Testing is available for a number of genetic disorders which are more common in people of Ashkenazi Jewish descent. After reading the information (below) on these disorders, please sign in the appropriate place below:

I acknowledge that I have read and understand the pamphlet on Jewish disorders and:

Neither the baby's father nor I are Jewish

\_\_\_\_\_  
We (or one of us) are Jewish but decline to be tested

\_\_\_\_\_  
We (or one of us) are Jewish and we wish to be tested.

**Please be aware that most patients will wish to complete all genetic testing (on both parents) as soon as possible so as to leave adequate time to test the baby for these disorders (if necessary).**

### **Genetic Disease Carrier Screening for Screening for the persons of Ashkenazi Jewish Descent**

**Canavan Disease:** Carrier Frequency 1 in 57, a demyelinating disorder affecting the central nervous system characterized by macrocephaly, developmental delay, severe hypotonia and failure to achieve independent sitting or speech. Symptoms usually occur within the first few months of life and the disease is fatal in early childhood. Currently there is no treatment.

**Familial Dysautonomia:** Carrier Frequency 1 in 30 Progressive nervous system disorder that causing vomiting, sweating, decreased pain sensitivity, and unstable blood pressure or temperature. Individuals often have normal intelligence, but may have learning disabilities. Symptoms management improves quality of life. Only 50% of affecting individuals will reach age 30.

**Tay-Sachs Disease:** Carrier Frequency 1 in 30 A lysosomal storage disorder that causes untreatable neurological degeneration. In the common infantile form, death occurs by 5 years of age. Currently there is no treatment Mutations

**Bloom Syndrome:** Carrier Frequency 1 in 100 A disorder of DNA repair, characterized by poor growth, immune deficiency, sun sensitivity and high susceptibility to cancer. Death from cancer usually occurs before 30 years of age. Intelligence is not affected

**Fanconi Anemia Group C:** Carrier Frequency 1 in 89 An inherited anemia sometimes accompanied by short stature, radial ray defects and cardiac and urogenital abnormalities. Learning disabilities or mental retardation sometimes occur. The risk of early childhood cancer, especially leukemia, is increased

**Gaucher Disease:** Carrier Frequency 1 in 15 A lysosomal storage disorder with variable severity. Children or adults may have anemia, hepatosplenomegaly, nosebleeds, and fractures. Effective treatment is available for Gaucher disease type 1. In the most severe and rare form, the brain and nervous system are involved.

**Mucopolidosis Type IV:** Carrier Frequency 1 in 122 A lysosomal storage disorder characterized by psychomotor retardation, corneal clouding, strabismus and retinal degeneration. Onset is usually within the first year of life, and affected individuals reach the developmental age of 1 to 2 years. Patients can live into adulthood. Currently there is no treatment.

**Niemann-Pick Disease Type A:** Carrier Frequency 1 in 90 A lysosomal storage disorder resulting in poor growth hepatosplenomegaly and progressive mental and physical deterioration. Death occurs by 4 years of age. Currently there is no treatment.

**Dihydroplipoamide Dehydrogenase Deficiency:** Carrier Frequency 1 in 96 Presents as persistent lactic acidosis with recurrent episodes of vomiting and abdominal pain. There is progressive neurological impairment, hepatomegaly, cortical blindness, stupor and coma. Mortality is high.

**Familial Hyperinsulinism:** Carrier Frequency 1 in 66 Causes hypoglycemia ranging from mild to severe. Onset ranges from newborn period to first years of life. If left untreated, may be lethal or result in irreversible neurological damage.

**Glycogen Storage Disease Type 1a:** Carrier Frequency 1 in 71 A metabolic disorder that, if untreated, results in severe hypoglycemia, hepatomegaly, growth retardation and bleeding disorders. Treatment consists of a strict diet and continuous feedings of glucose.

**Maple Syrup Urine Disease:** Carrier Frequency 1 in 81 A metabolic disorder that leads to the accumulation of branched-chain amino acids in the blood. Without treatment, classic MSUD results in mental retardation, physical disabilities, coma and death. Treatment requires dietary restriction of branched-chain amino acids through a special medical formula and intensive monitoring

**Nemaline Myopathy:** Carrier Frequency 1 in 149 A disorder characterized by weakness and poor muscle tone. Muscle weakness is usually most severe in the face, the neck flexors, and the proximal limb muscles. In the most severe form, death often occurs in the first few years of life due to respiratory failure.

**Usher Syndrome Type IF:** Carrier Frequency 1 in 141 Causes profound deafness at birth, severe balance problems, as well as vision impairment. Blindness progresses over time. Children with this disorder are slow to sit without support and typically don't walk independently before 18 months. Decline in visual acuity typically begins by age 10. Currently there is no treatment

**Usher Syndrome Type III:** Carrier Frequency 1 in 107 Hearing and vision progressively worsen, although the rate of decline varies. Individuals are usually completely blind by adulthood and hearing loss is moderate to severe. Balance is not usually affected. Currently there is no treatment.

# Testing for Cystic Fibrosis, Fragile X, and Spinal Muscular Atrophy

## Cystic Fibrosis (CF)

- *Symptoms of the disease:* **The most common inherited disease of children and young adults.** CF primarily involves the respiratory, digestive and reproductive systems. Symptoms include pneumonia, diarrhea, poor growth and infertility. Some people are only mildly affected but individuals with severe disease may die in childhood. With treatments today, people with CF can live into their 30's. CF does not affect intelligence.
- *Inheritance:* If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with cystic fibrosis.
- *General Population:* 1 in 30 average in the U.S.
- *Carrier Frequency:* varies by ethnicity
- *FOR CF:* If I am a carrier, testing my partner will help me learn more about the chance that our baby could have CF. If one parent is a carrier and the other is not, it is still possible that the baby will have CF but the chance is less than 1%. If both parents are carriers, prenatal testing is available to find out whether or not the baby has inherited the abnormal CF genes.

## Spinal Muscular Atrophy (SMA)

- *Symptoms of the disease:* **The most common inherited disease of children and young adults.** SMA destroys nerve cells that affect voluntary movement. Infants with SMA have problems breathing, swallowing, controlling their head or neck, and crawling and walking. The most common form of SMA affects infants in the first months of life and can cause death between 2-4 years of age. Less commonly the disease starts later and people can survive into adulthood. SMA does not affect intelligence. There is no cure or treatment.
- *Inheritance:* If both parents are carriers there is a 1 in 4 (25%) chance to have a child with SMA
- *General Population:* Ranges from 1 in 35 to 1 in 117 in the U.S. varies by ethnicity
- *Carrier Frequency:* varies by ethnicity
- *FOR SMA:* If I am a carrier, testing my partner will help me learn more about the chance that our baby could have SMA. If one parent is a carrier and the other is not, it is still possible that the baby will have SMA but the chance is less than 1%. If both parents are carriers, prenatal testing is available to find out whether or not the baby has inherited the abnormal SMA genes.

## Fragile X Syndrome

- *Symptoms of the disease:* **The most common inherited cause of mental retardation.** Fragile X syndrome involves developmental delay, mental retardation, autism and hyperactivity. It primarily affects boys. Women who are carriers are at risk to have a child with mental retardation.
- *Inheritance:* If a mother is a carrier, there is a 50% chance to have a child with fragile x syndrome
- *General Population:* ~1 in 260 women
- *Carrier Frequency:* occurs in all ethnic backgrounds
- *FOR FRAGILE X:* If I am a carrier, prenatal testing is available to find out whether or not the baby has inherited the abnormal fragile x gene.

### **You should be certain you understand the following points:**

- The purpose of these tests is to determine whether I am a carrier of one of the common genetic abnormalities that cause CF, SMA, and/or fragile x syndrome
- The tests do not detect all carriers of these disease
- The laboratory needs accurate information about my family history for the most accurate interpretation of the test results
- The decision to have carrier testing is completely mine.
- No other test will be performed and reported on my sample unless authorized by my doctor, and any unused portion of my original sample will be destroyed within two months of receipt of the sample by the laboratory
- I have read, or had read to me, the information in this brochure and I understand it. Before signing this form, I have had the opportunity to discuss carrier testing further with my doctor, someone my doctor has designated, or with a genetics professional. I have all the information I want and all my questions have been answered. I have decided that:

I want CF Carrier Testing

I do not want CF carrier testing.

I want SMA carrier Testing

I do not want SMA carrier testing.

I want Fragile X testing

I do not want Fragile X testing

Patient Signature: \_\_\_\_\_

Date: \_\_\_\_\_

## What Is A Genetic Counselor?

Traditionally, a genetic counselor has a masters degree in genetic counseling and has studied genetic diseases and how those diseases run in families. The genetic counselor can help a person or family understand their risk for genetic conditions (such as cystic fibrosis, cancer, or Down syndrome), educate the person or family about that disease, and assess the risk of passing those diseases on to children.

**We strongly recommend genetic counseling for all pregnant women who may be interested in pursuing testing for chromosomal or genetic (hereditary) problems. The options available for testing can be confusing, and genetic counseling can help decide which choice is right for you.**

## Testing for Chromosomal Abnormalities

Amniocentesis (done at about 16 weeks) or CVS (done at about 11 weeks) are the only methods that find almost all chromosomal problems. Both of these tests, however, are invasive and can cause a miscarriage. Please note that they do not test for all birth defects or genetic problems.

Non-invasive tests for chromosomal problems are “screening tests” that provide you with your risk of having a baby with certain chromosomal problems. None of these tests, however, give you a clear “yes or no” answer. They simply tell you what your chance is for having a baby with certain problems (like Down syndrome). For example, a non-invasive test may tell a woman that her risk of having a baby with Down syndrome is one in 200 or one in 3000. Most patients who are told that they are at increased risk of Down syndrome do not really have a problem (they have a “false positive”).

Screening tests are only able to screen for two of the most frequent problems (Down syndrome and Trisomy 18) and will not be positive in all patients who have a fetus with these disorders. Amniocentesis and CVS are diagnostic tests and can detect all types of chromosomal problems.

Screening tests are done at three different stages of your pregnancy:

1. At the end of the first trimester (approx. 11 to 13 ½ weeks), blood test results are combined with an ultrasound (nuchal translucency). Many (or most) women will choose to start with this test.

2. Maternal serum AFP4 (quad screen) done at 16 to 18 weeks. This blood test can be drawn in our office. You will receive a pamphlet explaining this test in more detail at the visit prior to 16 weeks. This test can be combined with the first trimester test (the modified sequential test) to give one result which detects more cases of chromosomal problems than either test alone.

3. A “genetic” ultrasound done by a perinatologist (high-risk obstetrician) at about 20 weeks.

You can read about testing for Down’s syndrome (and Trisomy 18), genetic disorders, amniocentesis, and CVS by selecting the links on our website in the [Patient Education](#) section under: [Testing for chromosomal or genetic disorders](#). Brochures are also available in our office.

Please sign below in the appropriate place

I have read and understand the above information and:

I wish to have testing for chromosomal problems (such as Down’s syndrome) \_\_\_\_\_

I do not wish to have testing for chromosomal problems (such as Down’s syndrome)\_\_\_\_\_