Angela Intili, M.D., Ltd.

Board Certified in Obstetrics-Gynecology

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PRENATAL GENETIC SCREEN

1. Will you be 35 years or older when the baby is due? Yes___ No___

2. Have you and/or 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 abnormalities Yes___ No___
   - ♦ Neural tube defect, i.e., spina bifida, Meningomyelocele? Yes___ No___
   - ♦ Open spine, or anencephaly? Yes___ No___
   - ♦ Cystic Fibrosis? Yes___ No___

3. Do you or the baby's father have a birth defect? Yes___ No___

4. In any previous marriages, have you or the baby's father had a child that was born deceased or alive, with a birth defect not listed above in Question 2? Yes___ No___

   If ***yes***, what was the defect and who had 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:

   _____

   If the cause of the defect is known, please indicate: _____

6. Do you, the baby's father, or a close relative in either of your families have a birth defect, familial disorder, or chromosomal abnormality not listed above? Yes__ No___

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

7. In any previous marriages, have you or the baby's father had a stillborn child or more than 3 spontaneous pregnancy losses in the first trimester? Yes___ No___

8. Have either you or the baby's father had a chromosomal study? Yes___ No___

   If ***yes***, indicate who had the study and the results: _____

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PRENATAL GENETIC SCREEN
(Cont'd)

9. Have either of you been screened for Tay-Sachs disease? Yes___ No___

   If ***yes***, indicate who was screened and the results_____

10. Are you or the baby's father African American"? Yes___ No___

    If ***yes***, have either of you been screened for sickle cell trait? Yes___ No___
    If ***yes***, indicate who was screened and the results:_____

11. Are you or the baby's father of Philippine or Southeast Asian ancestry? Yes___ No___

    If ***yes***, have either of you been tested for A-thalassemia: Yes___ No___
    If ***yes***, indicate who was tested and the results: _____

12. Excluding iron and vitamins, have you taken any prescription or non- prescription medication, or taken any illegal drugs since being pregnant or since your last menstrual period? Yes___ No___

    If ***yes***, give name of medication or drug and dates taken: _____

13. Have you had any exposure to x-rays since being pregnant or since your last menstrual period? Yes___ No___

    If ***yes***, give what type of x-ray and date: _____

14. Have you had any illnesses associated with skin rash since being pregnant or since your last menstrual cycle? Yes___ No___

    If ***yes***, what illnesses and when? _____

15. Have you or any of your sexual partners ever had any of the following disorders?
    - Herpes Yes___ No___
    - Gonorrhea Yes___ No___
    - Chlamydia Yes___ No___
    - Syphilis Yes___ No___
    - AIDS Yes___ No___

16. Do you smoke cigarettes or consume alcohol regularly? Yes___ No___

    If ***yes***, indicate daily amount: _____

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CONSENT FOR PRENATAL SCREENING BLOOD TEST FOR NEURAL TUBE DEFECTS AND DOWN SYNDROME

Most children in the United States are born healthy; however, 2-3% of babies are born with some type of major birth defect. Most of these birth defects are difficult to detect before the baby is born. Recently, a test to detect one type of these defects, ***Neural Tube Defects***, has been made available. ***Neural Tube Defects*** occur when the brain and the spinal cord do not form properly, resulting in anencephaly (a severe defect of the head and brain which is not compatible with life) or spina bifida (an open spine). This occurs in 1-2 pregnancies per 1000.

If an infant has one of these problems, an increased amount of protein called alpha-fetaprotein (AFP) is secreted into the amniotic fluid around the baby and into the mother's blood. By checking the level of AFP, 90% of these defects can be detected. ***This blood test is done at 15 to 19 weeks of gestation.***

Of 1000 women tested, 50 women will have an elevated level of AFP. These women will have a second blood sample drawn and 30 women will still have the elevated levels. These women will have an ultrasound to detect twins or other benign conditions which may have caused the elevated levels. Approximately 17 women will have no explanation for the elevated levels and will need to have amniocentesis (placing a needle in the sac around the baby to collect amniotic fluid) to measure AFP. This will show 1-2 women with infants having a Neural Tube Defect. In addition, low AFP may be predictive of Down Syndrome. Please know that this test is optional.

Please indicate below whether you desire to have this testing done.

_____ I choose to have this screening done.

_____
(Signature of Patient) (Date)

_____ I choose to decline this screening.

_____
(Signature of Patient) (Date)

_____
(Signature of Physician) (Date)

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CONSENT FOR CYSTIC FIBROSIS TESTING

Cystic fibrosis is an inherited genetic condition. It is inherited in a recessive manner. This means that both parents must be carriers to have an affected child. Being a carrier does not affect an individual person's health.

Cystic Fibrosis is a common disorder for Caucasians, but is found in all ethnic groups. The American College of Obstetricians and Gynecologists has recommended that carrier screening be offered to all couples when pregnant or considering pregnancy. If testing determines that the mother has the recessive gene, then the father will also need to be tested to determine if the child could be affected with CF.

CF testing is not required; it is an option. The decision to have this screening is your personal choice.

_____ ***I choose to have*** this screening done.

_____
(Signature of Patient) (Date)

_____ ***I choose to decline*** this screening.

_____
(Signature of Patient) (Date)

_____
(Signature of Physician) (Date)

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SCREENING FOR ASHKENAZI JEWISH GENETIC DISEASE PANEL

TAY-SACHS DISEASE

Tay–Sachs disease (TSD) is a severe progressive disorder of the central nervous system that causes death within the first few years of life. Infants with TSD appear normal at birth but by age 5-6 months develop poor muscle tone, delayed development, loss of developmental milestones, and mental retardation. Children with TSD lose their eyesight at age 12-18 months. This condition usually is fatal by age 6 years. Tay-Sachs disease is caused by a deficiency of the hexosaminidase A enzyme. No effective treatment is currently available.

Carrier screening should be offered *before* pregnancy to individuals and couples at high risk, including those of ***Ashkenazi Jewish, French–Canadian***, or ***Cajun*** descent and those with a family history consistent with TSD.

The TSD carrier rate in Jewish individuals of Eastern European descent (Ashkenazi) is approximately 1 in 30; the carrier rate for non-Jewish individuals is estimated to be 1 in 300. It has been determined that individuals of French–Canadian and Cajun descent also have a greater carrier frequency than the general population. Most individuals of Jewish ancestry in North America are descended from *Ashkenazi Jewish* communities and, thus, are at an increased risk for having offspring with this condition. The basis for these recommendations seems to be the high detection rate. The Committee on Genetics reaffirms support for screening for Tay-Sachs disease.

This test is optional. Please indicate below whether you desire to have this testing done.

_____ ***I choose to have*** this screening done.

_____
(Signature of Patient) (Date)

_____ ***I choose to decline*** this screening.

_____
(Signature of Patient) (Date)

_____
(Signature of Physician) (Date)