Informed Consent/Refusal for Genetic Testing

Maternal Serum Screening

1. The purpose of maternal serum screening is to identify pregnancies that may be at increased risk for open neural tube defects (ONTD), Down syndrome, or trisomy 18.
2. The screening test I am having is (circle one):
   - First*Screen*®—detects 83% of Down syndrome and 80% of trisomy 18; no information about ONTD
   - Sequential*Screen*SM—detects 80% of ONTD, 90.4% of Down syndrome, and 90% of trisomy 18
   - Integrated*Screen*SM—detects 80% of ONTD, 92% of Down syndrome, and 90% of trisomy 18
   - Serum Integrated*Screen*SM—detects 80% of ONTD, 87% of Down syndrome, and 90% of trisomy 18
   - AFP4®—detects 80% of ONTD, 81% of Down syndrome, and 80% of trisomy 18
   - MSAFP—detects 80% of ONTD, no information about Down syndrome or trisomy 18
3. Not all affected fetuses can be detected: some will be missed by any of these screening tests.
4. Some women with normal fetuses will have abnormal screening results.
5. Abnormal screening results may indicate the need for further testing, such as ultrasound and/or amniocentesis.

DNA Testing

1. The purpose of my DNA test is to determine whether I, or my fetus if fetal testing is ordered, have mutation(s) known to be associated with the following genetic condition or disease:_____.
2. This testing is done on a small sample of blood; in some cases a mouthwash sample can be used. For the fetus, testing is done on amniotic fluid, CVS or fetal blood.
3. Mutations are often different in different populations. I understand that the laboratory needs accurate information about my family history and ethnic background for the most accurate interpretation of the test results.
4. When DNA testing shows a mutation, then the person is a carrier or is affected with the condition or disease tested for. Consulting a doctor or genetic counselor is recommended to learn the full meaning of the results.
5. When the DNA testing does not show a known mutation, the chance that the person is a carrier or is affected is reduced. There is still a chance to be a carrier or to be affected because the current testing cannot find all the possible changes within a gene.
6. In some families, DNA testing might discover non-paternity (someone who is not the real father), or some other previously unknown information about family relationships, such as adoption.

Genetic Amniocentesis

1. The purpose of amniocentesis is to detect certain birth defects, including most fetal chromosome disorders and neural tube defects.
   My reason for having amniocentesis is _____.
2. Before the amniocentesis I will have an ultrasound to help locate the placenta and fetus. Ultrasound may also detect twins, incorrect dating of the pregnancy, and some, but not all, physical defects in the fetus.
3. Amniocentesis involves inserting a needle through the woman's abdomen into the fluid in her uterus. A small amount of fluid (less than 1 ounce) is taken out. There may be some discomfort when the needle is inserted.
4. There are serious complications in less than 1% of amniocentesis procedures. The most serious complication is miscarriage. Other possible, but rare, serious complications include hemorrhage, infection, or injury to the fetus. Minor complications include cramping, vaginal spotting, slight leakage of amniotic fluid, and soreness where the needle was inserted. Early amniocentesis (12-15 weeks gestation) may have a slightly higher risk than standard amniocentesis (after 15 weeks gestation) for pregnancy loss, amniotic fluid leakage, and culture failure.
5. Fewer than 1 in 100 amniocenteses need to be repeated because not enough fluid is obtained the first time. Occasionally, even though fluid is obtained, a diagnosis cannot be made, and the amniocentesis needs to be repeated.
6. The standard testing performed on an amniotic fluid sample is chromosome analysis, which can identify over 99% of chromosomal disorders, and AFP (alpha-fetoprotein) analysis, which can identify over 90% of open neural tube defects. Testing for other conditions will not be performed unless indicated in (1) above.
7. Normal test results do not guarantee the birth of a normal child. As in any laboratory test, there is a small possibility of error, and maternal cells may contaminate the sample. In addition, 3-5% of all pregnancies have birth defects which cannot be detected by testing amniotic fluid or by ultrasound examination.

Additional items of consent/refusal applicable to any of the above screening/testing

1. In the case of twins or other multiple fetuses, the results may pertain to only one of the fetuses.
2. In the case of abnormal diagnostic results, the decision to continue or to terminate the pregnancy is entirely mine.
3. The decision to consent to, or to refuse any of the above procedures/testing is entirely mine.
4. No test(s) will be performed and reported on my sample other than those authorized by my doctor; and any unused portion of my original sample will be destroyed within 2 months of receipt of the sample by the laboratory.
5. My doctor may release my pregnancy outcome or ultrasound and amniocentesis results to Genzyme Genetics to be used for statistical analysis of the laboratory's performance.
6. Genzyme Genetics will disclose the test results ONLY to the doctor named below, or to his/her agent, unless otherwise authorized by the patient or required by law.
7. My signature below indicates that I have read, or had read to me, the above information and I understand it. I have had the opportunity to discuss it, including the purposes and possible risks, with my doctor or someone my doctor has designated. I know that I may obtain professional genetic counseling if I wish, before signing this consent. I have all the information I want, and all my questions have been answered.

YES: I REQUEST that Dr./or an associate physican _____ perform amniocentesis and/or the genetic screening or testing marked above. I understand and accept the consequences of this decision.

_____	_____	_____
Patient Signature	Date	Witnessed by

NO: I DECLINE to have amniocentesis, and/or the genetic screening/testing offered to me. I understand and accept the consequences of this decision.

_____	_____	_____
Patient Signature	Date	Witnessed by

California, Georgia and New York have statutes requiring laboratories to send confidential results of certain genetic tests to state or federal health agencies for monitoring the detection of birth defects.

It is standard of care for physicians to obtain informed consent for genetic testing. This model consent form is provided by Genzyme Genetics as a courtesy to physicians and their patients. Relevant patient and/or physician educational materials are also available through Genzyme Genetics.

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