Pacific Obstetrics & Gynecology

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Prenatal Genetic Questionnaire

	Yes	No
1. Will you be 35 years old or older at the time of your baby's birth?	○	○
2. Have you or the baby's father had children or a close relative with any of the following?		
a. genetic/ chromosomal disorders (ex. Down syndrome)	○	○
b. muscular dystrophy	○	○
c. cystic fibrosis	○	○
d. hemophilia	○	○
e. thalassemia	○	○
f. spina bifida	○	○
g. mental retardation	○	○
h. other rare or unusual disorders	○	○
3. Are you or the baby's father Jewish?	○	○
if so, then have you/he been tested for Tay-Sachs?	○	○
4. Are you or the baby's father African American?	○	○
5. Have you or the baby's father had two or more miscarriages or stillbirths or neonatal deaths in the past?	○	○
6. Have you taken any medications, received vaccinations, had alcohol or recreational drugs or had an x-ray during this pregnancy?	○	○
if so, please explain: _____		
7. Have you had any illnesses or infections during this pregnancy?	○	○
if so, please explain: _____		
8. Have you been exposed to any chemicals/ toxins during this pregnancy?	○	○
if so, please explain: _____		
9. Are you and the baby's father blood related?	○	○
if so, please explain: _____		
10. Is there any other information regarding this pregnancy that you want to share?		
if so, please explain: _____		

Patient name: _____ Date: _____

Thank you!

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CHRISTINA J. LEE, MD JENNIFER C. LEE, MD

Carrier Screening for Cystic Fibrosis, Fragile X Syndrome, Spinal Muscular Atrophy

What is carrier testing?
Carrier testing involves a blood test from one parent (usually the mother of the baby, first) to determine if she carries a specific genetic mutation for a particular genetic disorder. If she tests positive, then the father of the baby will also be offered testing. Genetic counseling is available to further explain the results of any positive tests.

What is cystic fibrosis (CF)?
CF is the most commonly inherited disease in North America. It is a chronic disorder that primarily affects the respiratory, digestive and reproductive systems. Symptoms may include pneumonia, diarrhea, poor growth and infertility. Some people are only mildly affected and others are more severely affected that it may not be incompatible with life.
-- inheritance: autosomal recessive condition. If both parents are carriers of the same genetic mutation for CF, then there is a 1 in 4 chance that the baby may have CF.
-- carrier frequency: 1 in 30 in Caucasian Americans, 1 in 46 in Hispanics, 1 in 65 in African Americans, 1 in 90 in Asian Americans.
-- incidence of the disorder: 1 in 3,300 Caucasian, 1 in 15,000 African American and 1 in 32,000 Asian American children were born with CF in the United States in 1997.

Are you interested in carrier screening for CF? (please circle) YES NO

What is Fragile X Syndrome (FXS)?
Fragile X Syndrome is the most commonly inherited cause of mental retardation. It is a condition that manifests a spectrum of intellectual disabilities ranging from mild to severe as well as some physical characteristics (large/protruding ears, elongated face, large testes) and behavioral characteristics (hand flapping, social anxiety). If affects males more severely.
-- inheritance: has traditionally been considered an X-linked dominant condition with variable expressivity and possibly reduced penetrance
-- carrier frequency: 1 in 2000 males, 1 in 259 females
-- incidence of the disorder: 1 in 3600 males, 1 in 4000 - 6000 females

Are you interested in carrier screening for FXS? (please circle) YES NO

What is Spinal Muscular Atrophy (SMA)?
Spinal Muscular Atrophy is the most common genetic cause of infant death. SMA manifests in various degrees of severity but they all lead to progressive muscle wasting and mobility impairment.
-- inheritance: autosomal recessive condition. If both parents are carriers of this genetic mutation, then there is a 1 in 4 chance that the baby may be affected with the disorder.
-- carrier frequency: 1 in 41, across all ethnic backgrounds

Are you interested in carrier testing for SMA? (please circle) YES NO

Patient name: _____ Date: _____

Thank you!