Pacific Women's Healthcare Associates

GENETIC TESTING QUESTIONNAIRE

These questions will help in the care of your pregnancy. Your answers may indicate whether certain test would be helpful in evaluating the health of your unborn baby.

How old will you be when your baby is born? _____ Ethnicity? _____

Have you, the father of the baby, or anyone in either of your families ever had any of the following disorders? Please circle "YES" or "NO"
Please specify for each "YES" answer, the problem, and the relationship of the affected person

- Mental retardation YES NO
- Down Syndrome or any other chromosome abnormality YES NO
- Birth Defects (I.e., cleft lip or palate, limb defects) YES NO
- Spina Bifida (open spine), anencephaly YES NO
- Hydrocephalus (water on the brain) YES NO
- Congenital blindness or deafness YES NO
- Blood disorders (anemia's) YES NO
- Cystic Fibrosis YES NO
- Epilepsy or seizures YES NO
- Heart defects YES NO
- Bleeding disorders (hemophilia, blood clots, pulmonary embolism) YES NO
- Huntington's Chorea YES NO
- Kidney problems YES NO
- Mental illness YES NO
- Muscular Dystrophy YES NO
- Myotonic Dystrophy YES NO
- Neurofibromatosis YES NO

Have you had three or more miscarriages? YES NO
Are you using alcohol, tobacco, or taking any drugs? YES NO
Is there any history of twins or triplets in either of your family? YES NO
Are you concerned about any other problem your baby might have? YES NO

Other: _____

_____ _____
Patient Name Date

Choices in Cord Blood Banking

Cord blood banking is the **once in a lifetime opportunity** for parents to save the stem cells found in the blood of the newborn's umbilical cord. The preservation of these stem cells, which are different from the embryonic stem cells, allows families the benefit of having them available for **existing or future medical treatments**.

Cord blood stem cell banking is **completely safe** for both the mother and the newborn baby since cord blood stem cells are collected after the baby is born and after the umbilical cord has been clamped and cut.

Cord Blood Banking Options

When deciding what is best for you and your family, it is important to know about all of your cord blood banking options.

Family Banking allows you to store your newborn baby's cord blood stem cells **specifically for your family**, making them available immediately should your family ever need them. This service is provided by cord blood banks which charge a fee for collection, processing and storage, in which **you retain ownership of your newborn baby's stem cells**. Research has shown that transplants with related cord blood stem cells have a higher survival rate as compared with unrelated (publicly) donated cord blood stem cells.

Public Donation allows your family to offer your baby's cord blood stem cells to **the public network** at no cost if this option is available at your hospital. Your donation may then be made available to any patient requiring a cord blood stem cell transplant. Your family does not retain ownership of the cord blood once it has been donated. As a result, there is no guarantee that it will be available should it be needed by a family member. A fee is charged for cord blood stem cells released by a public bank to a patient undergoing a medical treatment. For more information about donating cord blood stem cells, please visit *www.bethematch.org/cord.*

Related Donor Cord Blood Program Through the related donor cord blood program, cord blood stem cells from your baby's umbilical cord can be collected after birth and used to **treat a biological sibling** with a diagnosed disease. A cord blood transplant may, in fact, provide their best hope for treatment. What's more there is no charge for collecting and storing the cord blood, if your family is eligible. This service is **provided free through a new program** developed by the Health Resources and Services Administration (HRSA) and administered by the National Marrow Donor Program (NMDP). Find out if your family qualifies for this program. Visit *www.marrow.org/relatedcord*

Medical Waste means that the cord blood will be **thrown out as waste**. Once discarded, these cells cannot be retrieved for future use.

I have read the information above and discussed my cord blood banking options with my healthcare provider.

_____

Patient's Signature

_____

Patient's Name

_____

Date

1- Gluckman E. Rocha V. Boyer-Chammard A., et al. Outcome of cord blood transplantation from related and unrelated donors. Eurocord Transplant Group and the European Blood and Marrow Transplantation Group. N Engl J Med. 1997:337:373-381.

Carrier Screening in Pregnancy for Common Genetic Diseases

Everyone has a risk to have a baby with problems. There are a few common disorders that can occur even without a family history and can be tested for today. You can have one simple blood test before the baby is born to determine if you *carry* the gene (DNA change) that causes the disorders shown below.

What is a carrier?
A carrier is a person who has a gene that increases the risk to have children with a genetic disease. People do not know if they are carriers until they have a blood test or an affected child. Some disorders occur only if both parents are carriers and other disorders occur only when the mother is a carrier.

What is carrier screening?
Carrier screening involves a blood test from one or both parents to determine if they carry a specific gene that increases the risk that their baby is affected. If you turn out to be at risk, prenatal testing such as amniocentesis or chorionic villus sampling (CVS) is available to determine if your unborn baby is affected. All testing is optional and you can choose which disorder(s) to be tested.

Disease	Cystic Fibrosis (CF)	Fragile X Syndrome	Spinal Muscular Atrophy (SMA)
Symptoms of Disease	*Most common inherited disease in North America.* A chronic disorder that 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 20's and 30's. CF does not affect intelligence. **CPT Code: 81220**	*The most common inherited cause of mental retardation.* Fragile X syndrome is a disorder that causes mental retardation, autism, and hyperactivity. It affects primarily boys. Women who are carriers are at risk to have a child with mental retardation. **CPT Code: 81243**	*Most common cause of inherited infant death.* SMA destroys nerve cells that affect voluntary movement. Infants with SMA have problems breathing, swallowing, controlling their head or neck, and crawling or 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. **CPT Code: 81401**
Inheritance	If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with cystic fibrosis	If a mother is a carrier, there is up to a 50% chance to have a child affected with fragile X syndrome.	If both parents are carriers, there is a 1 in 4 (25%) chance to have a child with SMA
Carrier Frequency	1 in 25 Caucasians 1 in 46 Hispanics 1 in 65 African Americans ~1 in 90 Asian Americans	1 in 260 females Occurs in all ethnic backgrounds	1 in 41 Occurs in all ethnic backgrounds
Would you like to have carrier screening? (please circle one and sign)	**YES or NO** **Sign:**	**YES or NO** **Sign:**	**YES or NO** **Sign:**