California Center for Reproductive Health

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CARRIER SCREENING FOR GENETIC DISEASES AND CHROMOSOMAL TESTING OF EMBRYOS

The goal of our practice is to make sure that you receive optimal care and attention to improve your chances of having a healthy pregnancy, and of course, a healthy child. Below are several testing options available at out Center, which can allow you to make educated decisions about your reproductive care.

CARRIER SCREENING FOR GENETIC DISEASES

Carrier screening can help you understand your risk of having a child with a genetic disease. Typically, carriers are healthy individuals; but when two parents are the carriers of the same genetic mutation they can have a child affected with the actual disease. Most people do not know they are carriers until they have a child born with the disease.

The Expanded Carrier Screening (ECS) test screens for diseases recommended by the American Congress of Obstetricians and Gynecologists and the American College of Medical Genetics. Such disorders include Cystic Fibrosis, Spinal Muscular Atrophy, Tay-Sachs disease, and Sickle Cell disease, among others. Some genetic diseases can significantly impair a child's normal development. For some of these conditions, early treatment can improve pregnancy outcomes. If both you and your partner are carriers for the same genetic mutation, your child has 1 in 4 (25%) chance of having that disease. However, through assisted reproductive techniques (ART) such as in vitro fertilization (IVF) and preimplantation Genetic Testing-Monogenic disease (PGT-M), embryos may be screened for your specific mutation and only embryos which are unaffected by the disease can be transferred into the uterus, significantly reducing the risk that a child will inherit the genetic disease. Alternatively, you may opt not to pursue PGT-M and instead undergo testing during your pregnancy to make informed reproductive decisions. Some individuals consider adoption or opt to not have children. Even if you would not choose any of these options, you can use the information to prepare for the birth of a child with a genetic disorder. You will have the opportunity to speak with your physician and a genetic counselor about the medical options available to you.

Like any carrier screening test, not all genetic diseases are screened for and some carriers of genetic mutations will not be detected. Therefore, carrier screening can reduce, but not eliminate, the chances for a genetic disease in the fetus.

The genetic screening test is covered by most insurance policies. The test results will be ready in about two weeks and you will be notified shortly thereafter. *For patients covered by insurance, the patient responsibility will be determined by the processing laboratory. For cash patients, the cost is $599.*

I acknowledge being offered carrier screening for genetic diseases, and

			_____ **Today's date**
☐	**I decline testing for myself and my partner at this time**	_____ **Patient**	_____ **Partner**
☐	**I wish for myself and/or my partner to be tested**	_____ **Patient**	_____ **Partner**

SCREENING FOR CHROMOSOMAL ABNORMALITIES

Preimplantation Genetic Testing-Aneuploidy (PGT-A) pertains to testing of embryos for chromosomal abnormalities before they are transferred into the uterus. Specifically, each embryo may be tested to determine if it has the full complement of 46 chromosomes. If an embryo is screened as having a chromosomal abnormality, such as an extra chromosome (e.g. Down syndrome), or a missing chromosome (e.g. Turner syndrome), couples have the choice of avoiding transfer of such embryos into the woman's uterus. Studies have shown improved live birth rates and reduced miscarriage rates in women who undergo IVF and PGT-A as compared with IVF alone. This test is a voluntary test only available for couples undergoing IVF, and serves as an adjunct to the IVF treatment.

Like any screening test, not all chromosomal abnormalities can be screened for and some chromosomal abnormalities will not be detected with PGT-A. Therefor PGT-A can reduce, but not eliminate, the chances for a chromosomal abnormality in the fetus.

Preimplantation genetic testing is *not* covered by most insurance plans. In the majority of cases PGT-A necessitates freezing of embryos. Test results will be ready in about two weeks and you will be notified shortly thereafter. *For cash patients, the cost is $5200.*

I acknowledge being offered PGT-A for chromosomal abnormalities, and

			_____ **Today's date**
☐	**I decline testing of embryos with PGT-A at this time**	_____ **Patient**	_____ **Partner**
☐	**I wish to have more information before deciding on PGT-A**	_____ **Patient**	_____ **Partner**
☐	**I wish to have testing of embryos with PGT-A done**	_____ **Patient**	_____ **Partner**