Carrier Screening For Genetic Diseases Preconception Consent

(Female and/or Male Partner)

The goal of our practice at ARMS is to make sure that you receive optimal care to improve your chances of having a healthy pregnancy and a healthy child. An important consideration in your pre-pregnancy planning for a healthy family is genetic carrier screening of you as prospective parents. Genetic carrier screening can help you understand your risk of having a child with a serious genetic disease.

Each of us carries some genetic mutations or alterations in the DNA of our genes. For the great majority of us, these DNA gene mutations remain silent and do not cause disease. However, some of us are carriers of mutations in our genes that have the potential to cause genetic diseases in our children. Typically carriers for recessive genetic diseases are healthy individuals who carry a genetic mutation in one of a pair of genes that are necessary for our health. Carriers of recessive disease mutations are generally not affected by the abnormal gene mutation because the normal gene in the pair of genes compensates for the abnormal gene's deficiency. However, when two parents are carriers for the same genetic disease, they can transmit their abnormal gene mutations through their sperm and egg into the genes of the fertilized egg. The abnormal mutations in both genes of the fertilized egg ultimately result in the future child being affected with the genetic disease caused by the abnormal genes. Sadly, most people do not know they are carriers for a genetic disease until they have a child born with the disease.

Genetic screening is available to detect if future parents are carriers for recessive genetic diseases prior to their conceiving a pregnancy. Genetic screening is usually performed on one parent first, and if the first parent tests positive as a carrier for a disease, then the other parent is tested; however, both parents may elect to be tested at the same time.

The American College of Medical Genetics (ACMG) recommends that every adult planning a pregnancy be offered pre-pregnancy genetic screening for the incurable and commonly fatal genetic diseases of cystic fibrosis (CF) and spinal muscular atrophy (SMA). The frequency of carriers in the general population for CF is 1 in 25-30 Caucasians; 1 in 46 Hispanics; 1 in 65 Africans, and 1 in 90 Asians. The frequency of carriers in the general population for SMA ranges from 1 in 40 to 1 in 60 regardless of ethnicity. In addition, The American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) recommend screening for certain other incurable genetic diseases as indicated due to family history or ethnicity. These diseases are Sickle Cell Anemia in African ethnicity; Thalassemia in Southeast Asian and Mediterranean ethnicity; and in individuals of Ashkenazi Jewish ethnicity: Tay Sachs Disease, Canavan Disease, Bloom Syndrome, Familial Dysautonomia, Fanconi anemia group C, Gaucher's Disease, Mucolipidosis IV, and Niemann-Pick Disease type A. Genetic screening for the most common inherited form of mental retardation, Fragile X Syndrome, is available for all women and is recommended for prospective parents with a family history of mental retardation, autism, attention deficit disorder, adult onset tremor and ataxia (poor motor coordination), and premature ovarian menopause. The premutation carrier frequency for Fragile X Syndrome in women is 1/100-1/250 (men are not premutation carriers)

Prior to initiating fertility treatment at Arizona Reproductive Medicine Specialists (ARMS) we would like for you to make an informed choice in selecting preconception genetic carrier screening or deciding against having preconception genetic carrier screening.

ACOG-ACMG Preconception Screening: The Family Prep Screen by Counsyl Laboratory

The Family Prep Screen is an expanded test panel that screens for more than one hundred incurable genetic recessive diseases and includes all of the preconception genetic carrier screen tests recommended by The American College of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics (ACMG) for individuals of all ethnicities. Please visit www.counsyl.com/diseases for the full list of the diseases screened in this expanded screening test

Genetic carrier screening is often covered by insurance. The laboratory we recommend, Counsyl, will handle the billing process. Counsyl treats all patients as if this test was covered as an in-network benefit under their plan. As with any medical bill, patients will be responsible for co-pays, co-insurances, or deductibles according to their insurance plans. It is patient's responsibility to contact Counsyl for pricing for testing. All payments or billing are solely between Counsyl and the patient. ARMS does not guarantee coverage or price for this testing. Patient acknowledges no price by an ARMS employee was provided.

I acknowledge that I elect to NOT be screened by The Family Prep Screen by Counsyl Laboratory, including waiving Fragile X testing (For Female Patients)

Patient Name: PRINT	
Signature:	
Date:	
Partner Signature: (If Applicable)	
Date:	

I acknowledge that I elect to be screened by The Family Prep Screen by Counsyl Laboratory

Patient Name: PRINT	
Signature:	
Date:	
Partner Signature: (If Applicable)	
Date:	

_____We understand and agree that if spouse tests positive for any condition, the partner will need to be tested, prior to moving forward with treatment.