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Prenatal Carrier Screening Acceptance Rates in Adopted Individuals

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I. Introduction and Purpose

Patients who are referred for prenatal genetic counseling at Integrated Genetics are routinely offered general population carrier screening for Cystic Fibrosis (CF), Spinal Muscular Atrophy (SMA), and Fragile X syndrome. The offering of these tests is based on the physician's direction along with ACOG and ACMG guidelines regarding CF and SMA.

The purpose of this analysis was to determine if the acceptance rates for these same general population carrier screening tests are different in patients who identify themselves as being adopted. We limited the data collection and analysis to the female partner who was referred as the primary patient for prenatal or preconception comprehensive genetic counseling.

II. Study Design

We looked at all patients referred for comprehensive prenatal or pre-pregnancy genetic counseling in 2014 and 2015 and identified 643 adopted patients during the 2 year time period. When analyzing the acceptance rates for the testing, we excluded any patients for whom the testing was previously done prior to the genetic counseling session. All patients were given an informational sheet discussing the disorders for which they were going to be offered carrier testing which included descriptions of the disorder, symptoms, inheritance pattern, and carrier frequency. During the genetic counseling appointment, the patients were offered additional information about the option of carrier testing and prompted for a decision about whether they wanted to pursue that testing.

Patient Information Sheet:

A portion of the 1 page information sheet provided to patients prior to genetic counseling

Carrier Screening in Pregnancy for Common Genetic Diseases

Although most people have healthy babies, with every pregnancy there is a 3-4% chance to have a baby born with problems. The following are a few common, serious disorders that can occur even without a family history. You can have carrier screening (a simple blood test) before the baby is born to determine if you *carry* the genes that cause 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 specific 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.

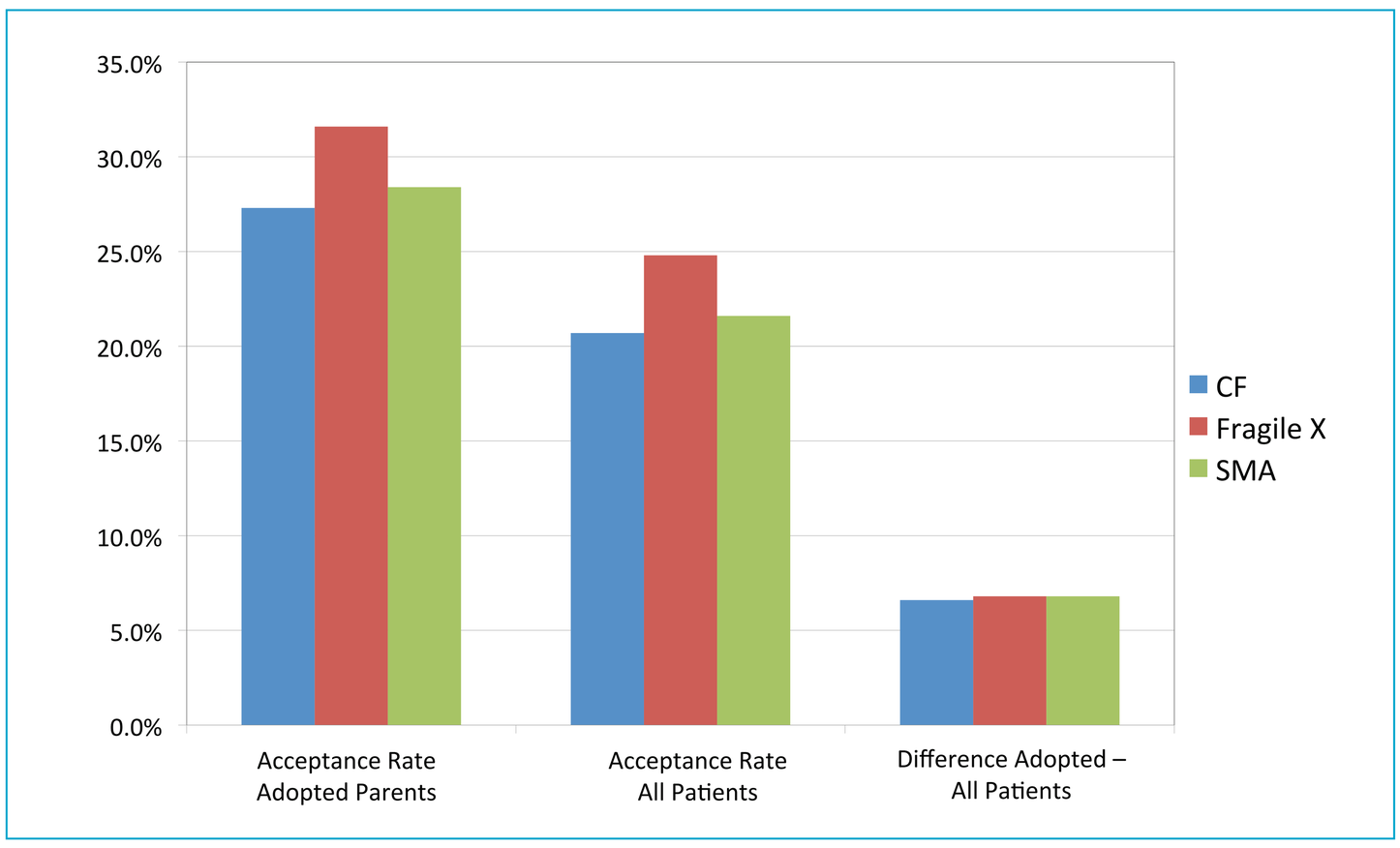
What is carrier screening?
Carrier screening involves a blood test from one or both parents to determine if they carry a specific gene that increases the risk for that disorder. If you turn out to be a carrier, prenatal testing such as amniocentesis or chorionic villus sampling (CVS) is available to determine if your unborn baby is affected. All testing is optional and you can choose which disorder(s) for which you want 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	*The most common inherited cause of mental retardation.* Fragile X syndrome is a disorder that causes mental retardation, autism, and hyperactivity. It affects both boys and girls, although boys are usually more severely affected than girls. Women who are carriers are at risk to have a	*Most common inherited cause of 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 and 4 years of age. Less

III. Findings

For each of the three tests, we found that a significantly higher percentage of patients who were adopted accepted genetic testing as compared to the entire patient population.

- The acceptance rates for CF were 6.6% higher in the adopted patient group (27.30% compared to 20.68%) with p-value of 0.0018 by chi-square test.
- The acceptance rates for SMA were 6.8% higher in the adopted patient group (28.38% compared to 21.64%) with p-value of 0.0007 by chi-square test.
- The acceptance rates for Fragile X were 6.8% higher in the adopted patient group (31.57% compared to 24.79%) with p-value of 0.0007 by chi-square test.



IV. Discussion

The higher acceptance rates, in this analysis, for carrier screening of CF, SMA, and Fragile X, in patients who were adopted, are suggestive of a patient group that is more motivated to obtain information about inherited genetic risk factors. Patients were not asked for their reasons for pursuing carrier testing, therefore motivation for testing was not assessed in this study. However, one might propose that a lack of knowledge may lead to more anxiety about family history, which would be an underlying factor in the increased interest in testing for inherited genetic disorders. Currently, there are no recommendations specifically aimed at genetic testing in a patient who is adopted. Given the increased acceptance rates for these 3 tests, there may also be an increased level of interest in carrier screening for additional inherited disorders. The adopted patient population may be better served by being offered the option of expanded carrier screening to cover a broader spectrum of inherited genetic disorders.

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