



WOMEN'S HEALTH CARE GROUP OF PA
Main Line Perinatal Associates Division

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To our patients:

Congratulations on your pregnancy! We look forward to meeting you at your first office visit. Now that you are pregnant, there are many things to consider. This letter will introduce you to some of the screening and testing options available through Main Line Perinatal.

We have entered an era where parents can find out genetic information about their children prenatally. Certain genetic diseases are carried by the parents and others are only carried by the baby. Here are some available testing options that we will discuss with you further:

- **Cystic Fibrosis.** This is the most common heritable disease in Caucasians, but it can occur in all races. 1 in 30 Caucasians is a carrier. Both parents must be carriers for the child to be affected. Cystic fibrosis leads to respiratory, digestive, and fertility problems. These children have a decreased life span.
- **Spinal Muscular Atrophy.** This is the most common lethal heritable disease in infancy. The carrier rate is 1 in 40. Both parents must be carriers for the child to be affected. Spinal muscular atrophy leads to muscle weakness and respiratory failure. Most children live only 1-2 years.
- **Fragile X Syndrome.** This is the most common heritable cause of mental retardation and autism, primarily affecting boys. Only the mother must be a carrier for the child to be affected. Fragile X syndrome causes mental retardation, autism, developmental delay, and behavioral issues.
- **Chromosome Abnormalities.** This includes conditions such as Down syndrome, Trisomy 18, and Turner syndrome. These conditions do not typically run in families and the occurrence is related to the age of the mother. If parents would like to know with 100% certainty, fetal testing through amniocentesis or placental biopsy is available. Alternatively, screening tests which only require blood testing of the mother can determine if the baby is "high-risk" or "low-risk" for these disorders. The screening tests include the sequential screen and non-invasive prenatal testing.

At your first visit, we will be glad to discuss these with you further. We look forward to meeting you.

The Physicians of Main Line Perinatal