

Name \_\_\_\_\_ D.O.B. \_\_\_\_\_

*PLEASE READ, COMPLETE AND BRING FORM WITH YOU FOR YOUR FIRST OFFICE VISIT*

Dear EPW patient,

There are multiple tests available to you to screen for **chromosomal abnormalities** (e.g. Down syndrome), **inherited diseases** (e.g. cystic fibrosis), or **other abnormalities** (e.g. spina bifida).

Deciding on testing is a very personal decision. If you decide against testing, we routinely do a screening ultrasound in our office between 18-22 weeks. Many abnormalities can be found by ultrasound. However, even major anomalies can be missed.

You will need to check with your insurance regarding what testing is covered under your plan. We have included CPT codes next to each testing option to assist in your communication with your insurance company.

Many women base their decision to test depending on their risk of having an affected child. Older age during pregnancy, family history, or a history of having an affected infant are some of the most common reasons your risk may be increased. You will need to factor in the risk of pregnancy loss or complication from an invasive procedure and the consequences of having an affected child if testing is not done.

All women, especially those that will be age 35 or older at delivery, may want to consider amniocentesis or CVS.

The questions on page 2 can help us determine your risk. After answering those questions, on pages 3 & 4, you will find a description of some of the tests available at our office and those available in Louisville. Please let us know if you have any questions regarding your options.

Thank you for taking the time to help us assist you in making this important decision during your pregnancy,

Elizabethtown Physicians for Women

Name \_\_\_\_\_ D.O.B. \_\_\_\_\_

## GENETIC SCREENING QUESTIONS

These questions apply to *you*, the *baby's father*, and anyone in *either family*.

1. How old will you be when your baby is born? \_\_\_\_\_
  2. Are there multiple births (twins, triplets, etc)? No    Yes
  3. Is there any Jewish background? No    Yes
  4. Has a doctor told you there is a genetic/chromosomal (e.g. Down Syndrome) or inherited problem (e.g. cystic fibrosis) in either family? No    Yes
  5. If you are African American, do you have sickle cell trait or disease? No    Yes
  6. Are there any children with birth defects (e.g. spina bifida/open spine, hydrocephalus/water on the brain, heart defect)? No    Yes
  7. Are there any children on a special diet (e.g. diabetes, PKU)? No    Yes
  8. Does anyone have problems with their muscles, such as weakness, or problems walking, Duchenne's or any muscular dystrophy? No    Yes
  9. Does anyone suffer from mental retardation or mental slowing, slow to grow/develop, or slow to walk/talk? No    Yes
  10. Have any women had stillborn babies, babies that died shortly after birth, or had at least 2 miscarriages? No    Yes
  11. Is there anyone who is a free bleeder/hemophiliac or has thalassemia? No    Yes
  12. Are there any family health problems that you are worried your baby might have? No    Yes
- If you need blood products as a medical necessity, will you accept them?** **No    Yes**

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## **TESTING OPTIONS AT OUR OFFICE (for low and high risk pregnancies)**

**SECOND TRIMESTER ‘QUAD’ test (82105, 84701, 82677, 86336)**- This is a screening test which involves drawing blood from your arm at our office between **16 and 20 weeks** of your pregnancy. It detects up to 80% of open neural tube defects (e.g. spina bifida), up to 80% of Down Syndrome, and up to 60% of Trisomy 18. The test can return falsely positive in 5+% of patients.

**CYSTIC FIBROSIS or ‘CF’ (83891, 83892 x2, 83900, 83901 x14, 83909, 83912, 83914 x39)** is one of the most common inherited disorders in the United States. 1 in 3200 births is affected by CF. You could be a carrier of the gene even if there is no family history of CF. The risk of being a carrier varies by ethnicity (1 in 25 if Caucasian, 1 in 46 if Hispanic, 1 in 65 if African American, and 1 in 90 if Asian American). Both you and the father of the baby must be carriers in order for your child to have CF. Testing involves drawing blood from your arm at our office checking your DNA for the gene mutation. This can be **done at any time** before or during your pregnancy, but the earlier, the better. Once you are tested, you will never need to be tested again. If you are found to be a carrier, we then recommend the baby’s father obtain testing. Only then will be able to determine your baby’s risk.

## **TESTING OPTIONS IN LOUISVILLE (available to everyone but usually performed for higher risk pregnancies)**

**FIRST TRIMESTER SCREEN**- This is a screening ultrasound (evaluating the thickness of the fetal neck ‘nuchal translucency’) and blood work which involves pricking your finger. This is performed in Louisville by a high risk Obstetric group. The ideal time to have this performed is **12-13 weeks**. It detects up to 90% of Down Syndrome and up to 90% of Trisomy 18. It is usually combined with the second trimester ‘**AFP test**’ which involves drawing blood from your arm at our office between **16-20 weeks** to screen for open neural tube defects like spina bifida. These tests can return falsely positive in 5+% of patients. Most women who desire the First Trimester Screen also undergo the High Risk Ultrasound (see below).

**HIGH RISK ULTRASOUND (this is not a 3D or 4D ultrasound)**- Compared to the screening ultrasound done at our office, this is a more in depth screening ultrasound performed by a high risk Obstetric group in Louisville. Up to 75% of abnormalities are found by ultrasound, however even major fetal anomalies can be missed. This is performed at approximately **18 weeks**.

**AMNIOCENTESIS (59000)** - This is a diagnostic test performed by high risk Obstetricians in Louisville. It is usually offered between **15-20 weeks**. Amniotic fluid is removed with a needle through your abdomen. Loss of the fetus or other complications is usually less than 1/300-500.

**CVS (59015)** - This is a diagnostic test performed by high risk Obstetricians in Louisville. It is usually performed after **9-12 weeks**. A piece of placental tissue is removed either through your cervix or abdomen. Loss of the fetus or other complications is similar to amniocentesis (1/300-500).

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No medical test or ultrasound is perfect. A normal result does not guarantee a normal infant. False positive results commonly occur. When this happens, the test results indicate the fetus is at risk for an abnormality, when in reality the fetus is normal. This can cause significant anxiety for some patients. Please discuss any questions you may have with your physician or midwife.

I \_\_\_\_\_ (print name) verify that the above information was presented to me, explained to my satisfaction, and all my questions have been answered.

I **do/do not** (circle one) desire testing. Please assure you have checked the above tests you desire.

Patient signature \_\_\_\_\_ Date \_\_\_\_\_