

# AFP Tetra

## If my test result is positive, what happens next?

Follow-up options are discussed between you and your doctor. If your screening test is positive, your doctor may recommend one or more of the following:

- **Genetic counseling.** A genetic counselor can help you understand your test results and explain your options for finding out whether your baby has Down syndrome, trisomy 18, or an open neural tube defect. A genetic counseling session will include an in-depth discussion about your personal and family medical histories as well as your pregnancy history. Genetic counseling may be provided by a certified genetic counselor, a perinatologist (high-risk pregnancy doctor), or your own obstetrician.
- **Ultrasound.** This procedure uses high-frequency sound waves and a computer to create images of a developing baby. In the second trimester, a detailed ultrasound examination of a baby may be able to identify some birth defects such as open spina bifida. Babies with Down syndrome and trisomy 18 may have certain features that can be seen on ultrasound, but, in general, neither can be diagnosed by ultrasound alone. Ultrasound is also used to measure the baby and determine how far along you are in your pregnancy (your baby's gestational age). The levels of the proteins measured in the AFP Tetra test vary with each week of pregnancy, so knowing the exact gestational age is an essential part of the test. If ultrasound dating changes your baby's gestational age by 10 days or more, your physician may ask the lab to recalculate your test results. Ultrasound may reveal the presence of twins, which can also affect your AFP Tetra result.

- **Amniocentesis.** This procedure is usually performed after the 15th week of pregnancy. Ultrasound is used to guide a thin needle through the abdomen into the uterus, and a small amount of fluid (amniotic fluid) from around the baby is removed. The cells in the fluid are examined in the laboratory to find out whether a chromosome abnormality like Down syndrome or trisomy 18 is present. Amniocentesis can diagnose most chromosomal abnormalities but cannot diagnose or identify all birth defects. Alpha-fetoprotein (AFP) is also measured in the amniotic fluid, and if open spina bifida is suspected, a spinal protein called acetylcholinesterase (AChE) is measured as well. This combination of tests can diagnose most, but not all, babies with open spina bifida.<sup>5</sup>

### References

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# What is Maternal serum screening?

Maternal serum screening is a simple blood test offered during pregnancy to identify women who are at increased risk of having babies with Down syndrome, trisomy 18, or an open neural tube defect such as open spina bifida.

## What is AFP Tetra?

AFP Tetra is a maternal serum screening test that is offered between the 15th and 21st weeks (second trimester) of pregnancy. It measures the levels of 4 proteins in the mother's blood: AFP (alpha-fetoprotein), hCG (human chorionic gonadotropin), uE3 (unconjugated estriol), and dimeric inhibin A (DIA). Results of the blood test are combined with clinical information about the mother, such as her age and weight, to determine the developing baby's risk of having Down syndrome, trisomy 18, or an open neural tube defect.

## What are Down syndrome, trisomy 18, and open neural tube defects?

Down syndrome and trisomy 18 are conditions caused by chromosomal abnormalities. Chromosomes are present in every cell of the body and contain genetic information that helps determine how we look, how our bodies grow and develop, and our health. A developing baby normally receives 23 chromosomes from each parent for a total of 23 pairs of chromosomes in each cell. The chromosome pairs are numbered 1 through 23. Sometimes a baby can be born with too many or too few chromosomes. Errors in the number of chromosomes may cause a variety of birth defects, ranging from mild to severe.

In Down syndrome, also called trisomy 21, a baby has an extra copy of the number 21 chromosome. All babies with Down syndrome have some degree of mental retardation and often have physical abnormalities such as heart defects. About 1 in 800 babies is born with Down syndrome.<sup>1</sup> AFP Tetra is expected to detect 75% to 80% of pregnancies affected with Down syndrome in the early second trimester, with a false-positive rate of 5% (meaning 5% of unaffected pregnancies will have positive screening test results).<sup>2</sup> Women who have a positive screening test result will be offered further testing to find out if their babies have Down syndrome.

Trisomy 18 is also known as Edwards syndrome. Babies with this condition have an extra copy of the number 18 chromosome. Trisomy 18 causes severe mental retardation and physical abnormalities. Most babies with trisomy 18 do not survive the first year of life. Trisomy 18 is rare, occurring in 1 in every 5000 births.<sup>3</sup> AFP Tetra is expected to detect 73% of pregnancies with trisomy 18.<sup>4</sup>

Open neural tube defects, such as spina bifida and anencephaly, occur when a baby's spinal cord does not close completely during development. About 1 in 1000 babies is born with an open neural tube defect.<sup>1</sup> The effects of open spina bifida range from bladder control problems to paralysis to a buildup of fluid inside the skull (hydrocephalus). Anencephaly results in underdevelopment in parts of the brain. Babies born with anencephaly usually survive only hours or days after birth. AFP Tetra is expected to detect about 80% of cases of open spina bifida and 90% of cases of anencephaly.<sup>5</sup>

## What does it mean if my AFP Tetra screening is negative?

A negative test result indicates the likelihood that your baby has Down syndrome is reduced. However, the AFP Tetra screening test cannot completely rule out the possibility that your baby could have Down syndrome.

The measurements used in the AFP Tetra test can also be used to identify pregnancies with a relatively low risk of trisomy 18 and open spina bifida, but they cannot completely rule out the chances of having a baby with either of those conditions. Additionally, screening using the AFP Tetra test does not detect other types of chromosomal abnormalities or birth defects.

## Does a positive AFP Tetra result mean my baby has a birth defect?

No. Screening tests cannot diagnose problems with your baby or pregnancy. A positive test result can only tell you there is an increased risk that your baby may have Down syndrome, trisomy 18, or an open neural tube defect. Typically, a woman who has a positive screening result is offered additional tests to find out whether her baby has one of these conditions.