



## What is Down syndrome?

Down syndrome (trisomy 21) is the most common genetic cause of intellectual disability, occurring in approximately 1 in 750 births. Individuals with Down syndrome typically have mild to moderate intellectual disability and may have other physical or medical problems including heart defects. This screening test identifies a pregnancy at risk for Down syndrome, but further testing is necessary to diagnose this condition.

**A Quad Screen identifies approximately 60-70% of babies with Down syndrome.**

## What is Trisomy 18?

Trisomy 18 (Edwards syndrome) is a genetic condition caused by an extra chromosome 18 in each cell of the body. Babies with this diagnosis often have multiple birth defects, such as heart defects or clubfeet, growth difficulties, and severe to profound intellectual disability. Since this condition causes so many health concerns, many babies with Trisomy 18 are miscarried, stillborn, or pass away shortly after birth, but 1 in 10 babies with Trisomy 18 live past one year. Approximately 1 in 3,000-5,000 babies are born with this diagnosis.

**A Quad Screen identifies approximately 70% of babies with Trisomy 18.**

## What is Open Spina Bifida?

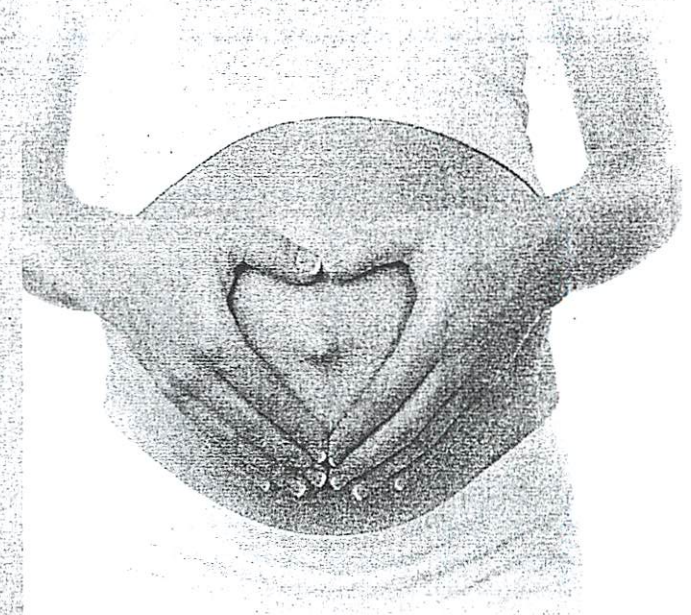
Open Spina Bifida is a birth defect of the spine resulting from a developmental difficulty very early in pregnancy. Individuals with spina bifida often have difficulty walking as well as health concerns with the bowel and bladder. Spina bifida is one of a group of birth defects that can occur along the head, neck, or spine; this group of birth defects is also called neural tube defects (NTD). One or two babies in 1,000 are born with an NTD with no prior family history. Risk factors for NTD may include diabetes, family history of NTD, or certain medication exposures.

**A Quad Screen identifies approximately 80% of babies with open spina bifida and approximately 90% of babies with anencephaly (a more severe form of NTD that occurs at the top of the head).**

## Summary

- *A Quad Screen is a blood test drawn between 14 weeks 0 days and 20 weeks 6 days from the first day of your last menstrual cycle.*
- *The Quad Screen cannot diagnose Down syndrome, Trisomy 18, or open spina bifida, but it can signal that additional testing is needed.*
- *In most cases, the Quad Screen will be "negative" and this will reassure you and your healthcare provider.*
- *Approximately 1 in 20 (5%) of patients will receive an abnormal or "positive" result. This result does not mean your baby has this diagnosis for sure; additional testing is needed to confirm or rule out a diagnosis. You and your provider may decide to obtain more information about the fetus to decide how to further manage the pregnancy.*
- *The Quad Screen does not identify all birth defects or genetic conditions. No test can identify all concerns.*
- *The Quad Screen is an optional screening test; you may choose to have or decline the screening.*

# Quad Screen



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## What is the Quad Screen?

The Quad Screen is an optional screening test available to pregnant women. Using a small sample of the mother's blood this test measures the following four proteins:

- alpha-fetoprotein (AFP)
- estriol (uE<sub>3</sub>)
- human chorionic gonadotropin (hCG)
- dimeric inhibin A (DIA)

The Quad Screen uses the patterns of the four proteins along with the woman's age, race, weight, and other pregnancy information to identify pregnancies that may have certain birth defects or genetic conditions such as *Down syndrome*, *Trisomy 18*, and *open spina bifida*.

## Why have the Quad Screen?

Patients who choose the Quad Screen want more information about Down syndrome, Trisomy 18, and open spina bifida during their pregnancy. In every pregnancy there is a possibility that a baby may have one of these diagnoses. Most babies born with these concerns have no family history of these conditions. This screening test may provide information important for making decisions regarding the management of a pregnancy. It can also prepare a family for the birth of a child with healthcare needs.

## Why might someone decline or decide not to have a Quad Screen?

There are many reasons an individual might not want the Quad Screen. Some individuals find the process causes anxiety. Others do not want to know the chance for Down syndrome, Trisomy 18, or open spina bifida. Some individuals decide instead to have a diagnostic test, like amniocentesis, while others find their insurance will not cover the cost. Also, if First Trimester Screening has been completed in the pregnancy, it is not recommended that a patient have a Quad Screen. If First Trimester Screening has been completed, "AFP only" is the recommended blood test for open spina bifida only.

## How long does it take to get results?

Results are typically available in less than one week.

## Potential Results and Implications:

### Positive (High Risk)

- A positive result can be seen for many reasons. Babies can be healthy, there may be a birth defect or genetic condition causing the unusual pattern of proteins, or there may be a small risk for other pregnancy complications.
- A screening test cannot diagnose a birth defect or genetic condition.
- Additional testing, including a detailed ultrasound, optional amniocentesis, and genetic counseling is offered to provide additional information.

### If the result is positive, can the Quad Screen be repeated?

- The Quad Screen can be repeated if the initial blood was drawn before 14 weeks or with some results that indicate an increased risk for open spina bifida.
- The Quad Screen should not be repeated if the result indicates a risk for Down syndrome or Trisomy 18.

### Negative (Low Risk)

- In most cases, the result will be negative and no additional diagnostic tests will be recommended for Down syndrome, Trisomy 18, or open spina bifida. Although this is reassuring, it does not rule out all birth defects.
- Even when the Quad Screen is negative an ultrasound is often recommended around 20 weeks to evaluate for general birth defects, fluid levels, and baby's growth.

## What is Ultrasound?

*An ultrasound exam uses high-frequency sound waves to create images on a video screen. The ultrasound is helpful in determining gestational age (age of the pregnancy starting at the onset of the last normal menstrual period), approximate fetal growth, number of babies, and certain abnormalities of the fetus. An ultrasound also cannot identify all birth defects or genetic conditions.*

## What is Amniocentesis?

*Amniocentesis is a procedure performed by a physician to obtain a small amount of fluid (amniotic fluid) surrounding the baby as it develops. This fluid is cultured and analyzed in the laboratory for appropriate tests including chromosome analysis and level of AFP. The procedure is done with ultrasound guidance. Most mothers and babies experience no complications beyond a mild cramping sensation. In a few instances, however, additional concerns may arise. The probability that an amniocentesis will lead to serious complications is less than 0.15% (1 in 600), when done by a high-risk pregnancy physician.*

## When can the Quad Screen be drawn?

Each laboratory varies, but the woman's blood is typically drawn between 14 and 21 weeks after the first day of the woman's last menstrual period.

## Can a Quad Screen be completed with a twin pregnancy?

A result is given when there are two babies but cannot be calculated when there are more than two babies. A Quad Screen is unable to calculate a risk for Trisomy 18 if there is more than one baby.