

North Carolina Women's Hospital
Cardiac Echogenic Focus
(kard-e-ak ek-o-jen-ik fo-kes)

What is an echogenic focus?

An echogenic focus is a “bright spot” in the heart that can be seen on ultrasound. It is most commonly seen in the left side of the heart.

How common is it?

An echogenic focus is seen in about 20% (1 in 5) of ultrasounds done during the second or third trimester.

What does having a baby with an echogenic focus mean?

An echogenic focus is not a heart defect and it does not keep the heart from working correctly. Most babies with an echogenic focus seen on ultrasound will be healthy. If there are additional findings seen on ultrasound, or if you will be 35 years or older at your due date, it may be a sign that the baby has a chromosome problem like Down syndrome. An echogenic focus seen on ultrasound is a clue to look at other parts of the body for problems and to consider more testing to look at the baby's chromosomes.

Chromosomes are structures that hold all of our genetic information. This genetic information acts as our body's instruction manual. If there is a change in these instructions, like extra or lost information, a baby may not develop properly. Most people have 46 chromosomes in every cell of their body. These chromosomes come in pairs, with one coming from the mother, and one from the father. This makes 23 pairs of chromosomes. If there is an extra or missing chromosome, then there are too many or too few instructions. Extra or missing instructions can cause different kinds of problems like birth defects or mental retardation.

What is Down syndrome?

Down syndrome (also known as trisomy 21) is a medical condition with signs including different facial features, mental retardation, and specific health concerns, including heart problems. Babies with Down syndrome may have a flat face, a small nose, small ears and extra skin at the back of the neck. People with Down syndrome need extra medical care throughout their lives, but in most cases, the quality of life for someone born with Down syndrome today is much better than it was in the past.

What causes Down syndrome?

Down syndrome is caused by an extra copy of chromosome 21. About 1 in every 800 babies has an extra chromosome 21. This happens when the egg or the sperm that came

together to make the baby had an extra chromosome. Extra chromosomes occur by chance and can happen in any pregnancy. It is not caused by anything that the parents did or did not do. We do know that as women get older, there is an increased chance of having a child with Down syndrome, but that is not something we can control.

How can I find out more about echogenic focus and Down syndrome?

You can talk to a genetic counselor about echogenic focus and Down syndrome. She or he will try to answer any questions you have. A genetic counselor can also talk about testing to find out if your baby has Down syndrome or any other chromosome problem.

There are two types of testing the genetic counselor will talk about. One is a screening test, sometimes called a quad screen. This is a blood test. It looks at chemicals the pregnancy has produced in your blood. Different levels of the chemicals are clues about whether the baby has a chromosome problem, but it will not tell you for sure if the baby has any extra or missing chromosomes.

The second test is called an amniocentesis. It is diagnostic and it *can* tell you for sure if the baby has a chromosome problem like Down syndrome. An amniocentesis is performed by taking a sample of the fluid that is around the baby. Because a needle is used to obtain the fluid, there is a risk for miscarriage or loss of the pregnancy. About 0.2-0.3% (or 1 in 300-500) of people who have this test may have a miscarriage.

Most babies with an echogenic focus do NOT have Down syndrome. Talk to the genetic counselor about the chances of your baby having a chromosome problem like Down syndrome.

You can schedule an appointment with a genetic counselor at UNC by calling (919) 843-6095.