

What doctors want patients to know about chorionic villus sampling

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AT A GLANCE: Chorionic villus sampling, or CVS, is an optional prenatal diagnostic test that can identify certain genetic or chromosomal conditions early in pregnancy. Here are the key things you will learn about chorionic villus sampling from [Roopali Donepudi, MD](#), a maternal fetal medicine physician at Texas Children's Hospital, in this article:

- CVS is usually performed between 11 and 13 weeks of pregnancy using a small tissue sample from the placenta.
- The CVS prenatal test can detect chromosome abnormalities and some genetic conditions, but it does not identify every possible birth defect.
- CVS may be recommended after abnormal ultrasound findings, earlier screening results, family history or a prior pregnancy affected by a genetic condition.
- The CVS procedure usually takes about 30 minutes, with the sample collected through the abdomen or cervix under ultrasound guidance.
- Chorionic villus sampling carries some risks, including miscarriage, infection, bleeding or leaking amniotic fluid, so patients should call their physician about heavy bleeding, severe cramping or other concerns.

Prenatal testing can bring a mix of anticipation, uncertainty and difficult decisions for expectant parents. Among the diagnostic tools available during pregnancy is chorionic villus sampling (CVS). This prenatal test can help identify certain genetic or chromosomal conditions early in pregnancy, such as Down's syndrome, sickle cell disease or cystic fibrosis.

Chorionic villus sampling is different from amniocentesis, which also is a diagnostic test used to detect fetal chromosomal and genetic abnormalities. The primary differences

involve when the tests are performed, what tissues are sampled and which specific conditions they can detect.

Usually performed in the first trimester between 11 and 13 weeks of pregnancy, CVS involves taking a small sample of tissue from the placenta for laboratory testing. While the procedure can provide important answers, it also raises questions about timing, accuracy, risks and what the results may mean for a growing family.

The AMA's What Doctors Want Patients to Know™ series gives physicians a platform to share what they want patients to understand about today's healthcare headlines.

In this installment, [Roopali Donepudi, MD](#), a maternal fetal medicine physician at Texas Children's Hospital in Houston, took time to discuss what CVS can and cannot tell you, how it differs from other prenatal tests, and when it might be recommended.

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Chorionic villus sampling tests genetics and chromosomes

"Chorionic villus sampling is a procedure that is done for genetic testing or testing of the baby's chromosomes," said Dr. Donepudi, noting "this procedure is done under ultrasound guidance."



Roopali Donepudi, MD

“We’re testing specifically for chromosome abnormalities,” she said. “There can be some birth defects where the chromosomes are normal, but we do see some abnormalities with the structure.”

“For example, the structure of the heart or the kidneys or other organs. But if there’s anything abnormal with their chromosome, then it can test for that,” Dr. Donepudi said.

CVS testing is recommended for high-risk pregnancies

“Anyone who is pregnant can have genetic testing,” said Dr. Donepudi, noting that “there are two types of testing. There are screening tests, which are blood tests, or you can have the confirmation or diagnostic testing, and CVS is one of those.”

While “anyone who’s pregnant can definitely request it, we do recommend it in certain high-risk populations. That would include anyone who has had a prior pregnancy where the baby was affected by any type of birth defects,” she said, “or if they’ve had a family history of any type of genetic abnormalities.

Even in this pregnancy, if we see anything abnormal on ultrasound it would be recommended.”

Since chorionic villus sampling is an optional, diagnostic test, it is usually reserved for high-risk pregnancies rather than a routine screening. Your doctor may offer or recommend

CVS prenatal testing if you meet any of the following criteria:

- You will be 35 or older by your due date.
- You or your partner have a known family history of genetic conditions.
- You have a previous child with a genetic or chromosomal abnormality.
- Results from earlier noninvasive prenatal tests, blood work or a first-trimester ultrasound indicate possible risks.

An ultrasound is done before CVS prenatal testing

“Typically, patients first see their ob-gyn and are then referred to a maternal-fetal medicine specialist for an ultrasound,” Dr. Donepudi said. “They’re usually between 10 and 12 weeks along in their pregnancy, and the ultrasound is where we start. If any risk factors are identified during that evaluation, chorionic villus sampling may be offered.”

“The purpose of the ultrasound is to confirm they’re pregnant, confirm their due dates, things like that,” she said. “But at the same time, at that visit, we screen for who would be considered high risk and then we would recommend the testing. For the patients who are just interested in the test, we can talk about this too.”

“Patients typically see a genetic counselor to go through all the different things that we can look for, and then after they talk to the counselor, they’ll talk to the maternal fetal medicine physician as well,” Dr. Donepudi explained. “Once they decide that they want to have the procedure, then we schedule it.”

CVS prenatal testing takes about 30 minutes

A chorionic villus sampling test is “an ultrasound-guided procedure,” Dr. Donepudi said, noting “there are a couple of ways of doing chorionic villus sampling. One of which you can through the belly with what looks like a really long needle, but it’s a very thin needle.”

“We’re watching by ultrasound in a CVS and we’re taking a tiny little sample from the placenta, so the baby is actually nowhere near that needle,” she reassured. “The other way we can do it is to go through the cervix, kind of like a pap smear in a way, so it shouldn’t be

painful.”

“It takes less than a minute for the actual procedure but getting in and doing the ultrasound can take a little bit of time,” Dr. Donepudi said.

Make sure to rest after the CVS test

“After the chorionic villus sample procedure, we recommend that night to take it really easy,” Dr. Donepudi said. “It’s not quite bedrest, but I tell everyone to be a couch potato, to just chill for the night and then you’re back to normal the next day.”

“You can have a little bit of cramping during the procedure itself. Some people will have it for maybe five to 10 minutes after, but most people are good by the time they’re ready to go home,” she said.

Only one CVS prenatal test is needed

“We try to make sure that chorionic villus sampling is only one test,” said Dr. Donepudi. “After we get the sample, we take a look at our sample to make sure it’s sufficient for testing.”

By doing that, “we wouldn’t normally have anyone come back for retesting with CVS,” she said.

That is not to say that another test is never needed. In rare cases, a physician may not have collected enough cells during the procedure and a second test is needed to get enough placental cells. Also, if you’re expecting twins and each fetus has its own placenta, you may need two CVS tests.

There are risks and side effects from CVS

“Anytime we do a needle-based procedure during pregnancy, there’s always a risk, depending on how far along you’re in the pregnancy,” Dr. Donepudi said. “For this

specifically, there's a risk for miscarriage, so we always tell patients to look out for any vaginal bleeding."

Some of the other risks of chorionic villus sampling are:

- Infection.
- Limb deformity in rare instances.
- Heavy vaginal bleeding.
- Leaking amniotic fluid.
- Rh sensitization, which is when you have Rh-negative blood, and the fetus has Rh-positive blood that mixes together.

"If a person has a little bit of spotting, that's OK. But if they have bleeding enough to soak up a pad or two in an hour or two hours, then I would definitely recommend they call their OB," she said.

The chorionic villus sampling test improves care

"The benefits are usually for the information to know what the chromosomes are," said Dr. Donepudi. "Depending on how severe the chromosomal abnormality is, it will determine how we manage their pregnancy."

"A lot of people think that we're using this test just for that immediate moment, but it actually helps determine the entire pregnancy, depending on how severe the condition is," she explained. "We can talk about how we manage mom's care, how we manage how frequently we need to do ultrasounds and how we get the pediatric subspecialists involved."

"It also affects the care of the baby," Dr. Donepudi said, adding that "a lot of the pediatric team really appreciate having this information as soon as the baby is born because then that will guide how they take care of the baby in those first few days."

Chorionic villus sampling results are ready in up to 10 days

The length of time it takes to get CVS test results “depends on what level of testing you want to do. We can do the absolute basic test, which we call a karyotype,” Dr. Donepudi said. “You just determine how many chromosomes the baby has, and we all have 23 pairs of chromosomes.

“Or you can go a little bit more into detail and do what we call a microarray. That’s actually looking at different parts of each chromosome,” she added. “Or you can do even more testing, called a whole exome sequencing, and that goes even more in depth.”

“So, depending on what our initial concern is, we recommend different levels of testing, and that result can be anywhere from seven to 10 days, but it can take a couple of weeks if we’re doing the more extensive testing,” Dr. Donepudi explained. “Our genetic counselors are monitoring the results every day. They’re reviewing to see if any of our results are back, so they will call as soon as they get any of those test results.

“We typically initially call 48 hours after the procedure just to make sure everything is OK and then whenever the results are back,” she added.

A positive CVS test requires more imaging

“When we say positive, that means that there is something abnormal with the chromosomes,” Dr. Donepudi said. “If there’s a small, tiny piece of the chromosome that’s missing or a small, tiny piece that’s extra, sometimes it may not cause any major concerns, and that’s good to know.”

“But if it’s a major abnormality, then that will lead us to talk more about additional imaging and getting all the other specialists involved,” she explained. “So, if it is a positive result, we do recommend more imaging, and we’ll have them come in much earlier than their anatomy scan.”

“Typically, the big scan is the 20-week ultrasound where we can look at all the different organs and determine the gender, but we have them come in much earlier if we do pick up any abnormality,” Dr. Donepudi said, emphasizing that “we always communicate with the OB physician as well, just so everyone’s aware of what’s going on.”

CVS also helps inform pediatric specialists

“To me and most people who do this type of testing,” said Dr. Donepudi, “we’re doing it to help not just the pregnancy, but also the pediatric subspecialist.”

“I can’t overemphasize this enough because people don’t necessarily think that far ahead,” she said. “But chorionic villus sampling is a very helpful tool for all our pediatric teams to be able to get that information well in advance.”