



This Test May Help Find the Cause of Many Miscarriages

Experts are working on helping people who want to be parents.

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Experiencing a miscarriage can be devastating, not just for the loss of a pregnancy, but also, in part, because the cause of a miscarriage can be so inexplicable.

According to the [Mayo Clinic](#), 10 to 20 percent of confirmed pregnancies end in miscarriages.

The number is even higher in chemical pregnancies, where the miscarriage happens after a positive pregnancy test but before they're confirmed on ultrasound. Chemical pregnancies are estimated to account for about 50 to 75 percent of miscarriages.

While many assume the miscarriage happened because of something going wrong in the pregnancy, most of the time, it's because there was something wrong with the embryo itself.

Now experts want women to know there's testing that can help them get answers. This is good news because steps can then be taken to avoid the potential heartbreak later on.

What causes most miscarriages?

Chromosomal abnormalities are the main cause of miscarriages. In fact, "about 70 percent of miscarriages are due to fetal chromosome aneuploidies, which means the gain or loss of a chromosome," Mandy Katz-Jaffe, PhD, reproductive geneticist and scientific director at CCRM, reports to Healthline.

The majority of these abnormalities relate to the age of the woman. A woman is born with all the eggs she'll have in her lifetime, and, as she gets older, her eggs do too.

With the aging of the eggs, the incidence of abnormal chromosomes goes up, making the embryos nonviable. It's how nature protects us from having an unhealthy baby.

Dr. S. Zev Williams, chief of the Division of Reproductive Endocrinology and Infertility and associate professor of obstetrics and gynecology at Columbia University Medical Center, said doctors will focus very heavily on genetic factors when trying to understand the cause of a miscarriage.

"When you're thinking about a miscarriage, it's happened and we're trying to understand the cause. It's basically genetic or everything else," Williams told Healthline.

Testing for chromosomal abnormalities tells doctors what's going on with the chromosomes inside cells, both within the parents as well as in the embryos or miscarried tissue itself.

Having a miscarriage because of an abnormality doesn't mean later pregnancies will end the same way.

One exception to this is what's called a "balanced translocation," in which pieces of the chromosome have swapped spots. "[The] majority of chromosomal abnormalities are related to age of the eggs, but there's the occasional situation that we see on a regular basis where mom or dad are experiencing balanced translocation that induces preprogrammed miscarriages in 60 to 70 percent of the cases," Dr. Juergen Eisermann, founder and medical director of IVFMD, reports to Healthline.

How can testing help people concerned about miscarriages?

In very complex cases, doctors use whole exome sequencing on the parents. Embryos can be tested this way as well.

As a result, mutations that cause miscarriage may be discovered that haven't been seen before.

"We had one couple who had a very sick child who was born, and then they had two other losses all with very unusual anomalies," Williams told Healthline. "All the testing came back normal. We did a whole exome sequencing and discovered a very novel mutation that she and her partner carry, the thirteenth case in the world of it, and now we could screen their embryos to make sure they don't have another affected child."

Besides genetic testing on the parents, it's important to test embryos or products of conception for chromosomal abnormalities as well.

"If the miscarriage sample is normal, then it's unlikely that the parents have some abnormality that's causing them to have a chromosomally normal miscarriage," Williams explained.

For women who have miscarriages as a result of abnormal embryos, in vitro fertilization (IVF) may be an option.

Although it's an expensive and lengthy process, it does help try to eliminate unhealthy embryos for the best chances of a pregnancy.

The woman or the couple will go through the IVF process to produce embryos, and those embryos are screened to ensure they don't have any abnormalities, such as missing or extra chromosomes. The abnormal embryos are discarded and only the healthy unaffected ones will remain to be put back into the uterus.

"The knowledge about whether a woman carries a genetic mutation that would predispose her to recurrent miscarriages would be powerful," Katz-Jaffe says to Healthline, "and [would] allow her to make informed decisions regarding her reproductive options."

Updates to testing

While this type of testing has been expensive and takes weeks for results to come back. Williams and his team at Columbia University have been working on new technology to get test results faster.

They have helped create a handheld DNA sequencer to make genetic screening of these cells or tissue more accessible, and at a fraction of the cost, something that can be a relief to anxious parents. Instead of waiting weeks for test results to come back, they could have answers that same day.

"It allows us to do very, very rapid DNA sequencing and can be used to pick up chromosomal abnormalities and genetic mutations, and what we sequence depends on the clinical need," Williams reports, "[DNA] can come from embryos where a couple of the cells are plucked off from the embryo, and that's what's used on the sequencer, but the other area where we use it would be miscarriage samples and CCS (comprehensive chromosome screening) samples."