

For most women, seeing two lines on a home pregnancy test means the excitement and anticipation of having a healthy baby in nine months' time. But for those women who suffer from recurrent miscarriage, a positive pregnancy test heralds a spark of joy that soon extinguishes, because it also brings with it a plethora of invasive testing, stress and fear.

Most miscarriages are caused by chromosomal or genetic abnormalities which occur naturally as part of the random nature of eggs and sperm joining. It's estimated that about 20% of pregnancies in women younger than 40 will end in miscarriage. It's devastating at the time but fortunately most women go on to sustain a normal pregnancy.

However, a small number of women suffer from recurrent miscarriage, which is defined as three or more miscarriages before 20 weeks gestation. It affects about two in every 100 women of reproductive age, and for these women the stress and heartbreak is monumental.

Testing times

Some Australian medical organisations, such as IVF Australia, operate miscarriage clinics, which offer medical testing to determine if there's a cause for miscarriage, plus counselling and care during the next pregnancy.

Fertility specialist Dr Gavin Sacks, who is codirector of IVF Australia's Miscarriage Care Program, says when a woman has repeated miscarriages, the suspicion is that it's not just a random event and that there's something else going on. One common cause is antiphospholipid syndrome (APS) – this is where you have antibodies in your blood that can act against the embryo. If a blood test finds you have APS it can be easily treated with blood-thinning agents or aspirin.

Another investigation into recurrent miscarriage is the karyotype test, which analyses the chromosomes of you and your partner. In a small percentage of couples one of the partners carries a chromosomal abnormality that can cause problems for a fetus if it's passed on. If this is found to be the case, it can be managed by IVF and then genetic diagnosis of the embryo before implantation.

Doctors will also test for blood clotting and metabolic disorders, as well as carry out anatomical tests to ensure there's no uterine abnormality. Your partner's sperm will be tested too, as sperm DNA fragmentation is another cause of recurrent miscarriage.

"If all these sorts of tests are carried out, we usually find an abnormality in about 50% of couples with repeated problems," says Dr Sacks. "The other 50% are left wondering 'now what?'"

Natural killer cells

A recently developed test offers hope to couples who don't know the cause of their miscarriages. In a collaborative project with Sydney's St George Hospital, Dr Sacks has been researching natural killer cells, a type of white blood cell that forms part of our immune system. Dr Sacks estimates that 25% of otherwise unexplained repeated miscarriages are due to natural killer cells. "The main purpose of these natural killer cells is as a form of surveillance," says Dr Sacks. "They go round the body looking for any form of invasion that could be a threat – it might be an infection or it could be cancer. They seek out cells that might be recognised as foreign or different and they can kill them."

Having abnormally high levels of natural killer cells in the blood and uterus are potentially very dangerous to pregnancy, as these are significant risk factors for miscarriage. Previously the only way to test for these cells was to have a uterine biopsy, but a new blood test that measures activated killer cells gives an accurate result without the need for surgery.

If doctors discover you have high levels of killer cells you can be prescribed progesterone, steroids or blood-thinning agents to suppress them. These are all safe to use in pregnancy.

The IVF Australia/St George study is the only one of its kind in this country. Specialists around Australia can do the biopsy; some women then choose to come to Sydney for the blood test. No formal trial has been done yet so there are no statistics to show how successful it is. But Dr Sacks says that over the past four years more than 50 Australian babies have been born following treatment for natural killer cells.

In fact, five of Dr Sacks's patients have had second babies following treatment to suppress their natural killer cells.

“It’s not established medicine and it is controversial,” says Dr Sacks. “Many people don’t believe in it yet but from what I can see it’s hard not to believe there’s something in it.

“There are a lot of patients who have had a terrible history and who say, ‘If it might work, why should I be denied it just because you haven’t produced a study?’ There are women who have had five, six, 10 miscarriages. People need hope and need to keep trying.”

Dealing with loss

The psychological pain following a miscarriage can be long lasting. Some women find it healing to celebrate the anniversary of when the baby was due to be born; others might buy themselves a piece of jewellery or plant a tree to remind them of the pregnancy.

IVF patients can go through as many as four cycles (and potentially four pregnancy losses) in just one year. Some couples find the grief and stress – not to mention the expense – too much and give up, while others keep trying, cycle after cycle.

“As doctors we can’t say when they should stop,” says Dr Sacks. “Ultimately the woman has to look back on her life and say, ‘I tried and I made the decision’.”

After having her first baby almost 10 years ago, the last thing 35-year-old Sarah* expected to be confronted with was multiple pregnancy loss. When she lost a pregnancy at 11 weeks it was considered fairly normal.

“It was like, ‘Oh yeah, that’s just one of those things’,” she says. But just five months later, she went through the pain and anguish of another miscarriage.

A karyotype test then found that Sarah has a chromosomal abnormality. Sarah and her husband Tom had to decide whether to keep trying naturally or opt for IVF; they decided to go for IVF and have an embryo implanted after it was checked for chromosomal anomalies. But while preparing for IVF, Sarah learnt she was pregnant again.

“We sort of thought it might be third time lucky for us,” she says. “But then I thought, ‘What if it happens again?’ I just didn’t think I could cope.”

Sadly, it wasn’t a case of third time lucky for Sarah and Tom: chorionic villus sampling showed that the fetus had an unbalanced form of Sarah’s chromosome abnormality that was incompatible with life outside the womb. The third miscarriage, this time at 14 weeks, was devastating for the family.

At the time of writing, Sarah was close to having her first embryos implanted via IVF.

“All this has made me realise how precious children are and just how hard it is to have a successful pregnancy,” she says. “It really is a miracle when babies are born.”

*Names have been changed.