Mosaic Embryos: What Do You Need To Know?

By Serena H. Chen, M.D. and Mark P. Trolice, M.D.

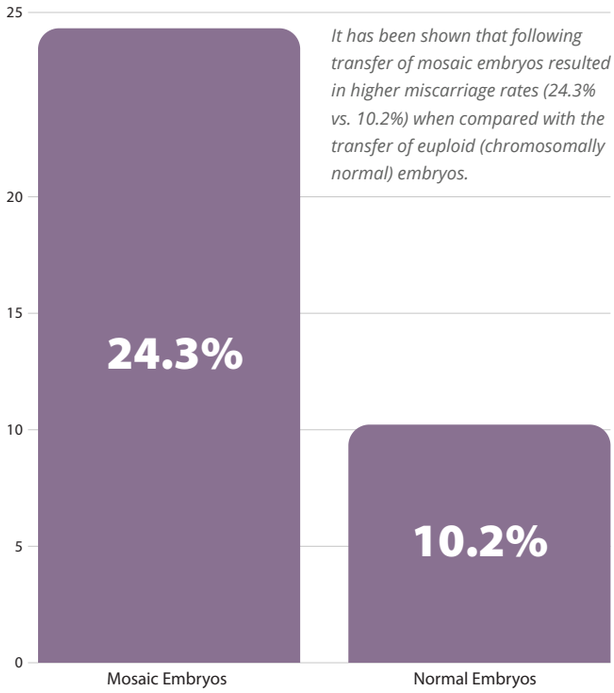
Advances in IVF allow for chromosomal testing of an embryo prior to transfer into your uterus. This exciting field of "Preimplantation" Genetic Testing (PGT) has dramatically increased in popularity and resulted in many new laboratory companies offering this embryo diagnostic procedure.

Studies demonstrate that when an embryo is found to be chromosomally normal by PGT, there appears to be a lower miscarriage rate in women above age 35.

PGT is the process of taking a cell or cells from an early-stage embryo (typically a blastocyst embryo) before it is implanted in the uterus in order to analyze the chromosomes in the embryo, or to look for specific disease-causing mutations like sickle cell disease, cystic fibrosis or muscular dystrophy.

Results of embryo testing following PGT are not simply normal and abnormal. More advanced testing of embryos shows a phenomenon called mosaicism. In a mosaic embryo, instead of one chromosomal analysis result, e.g. 46, XX (a normal female) shown in all the cells tested, a second chromosomal analysis (abnormal) also exists in some of the other cells tested, i.e. two cell lines in one embryo. The ultimate dilemma with a mosaic embryo? Scientists cannot know or agree if the embryo is 100% abnormal or if only the future cells of the placenta are affected.

Miscarriage Rates of Mosaic Embryos



It has been shown that following transfer of mosaic embryos resulted in higher miscarriage rates (24.3% vs. 10.2%) when compared with the transfer of euploid (chromosomally normal) embryos.

Currently, we have very little medical data on the implantation potential and development of mosaic embryos.

Recently it has been shown that following transfer of mosaic embryos resulted in lower ongoing pregnancy rates (39.2% vs. 63.3%) and higher miscarriage rates (24.3% vs. 10.2%) when compared with the transfer of euploid (chromosomally normal) embryos. (Reprod Biol Endocrinol 2018;16:1-6.) There is no long-term data because this phenomenon is a new result due to advanced testing technology, i.e. Next Generation Sequencing. (Fertil Steril 2017; 107:1085-1091). The few babies that have been born appear to be normal but what will happen as they get older? Does starting our life as a mosaic embryo increase your health risks in the future? Will you still be normal and healthy at age 5, 10, 20, 40 years of age?

Are all mosaic embryos alike?

It is very possible that mosaicism at the early embryo stage can be perfectly normal, but no one is currently certain. We have long known the placenta may have mosaicism, called confined placental mosaicism (CPM), in 1-2% of pregnancies tested by chorionic villus samplings (CVS) carried out for prenatal diagnosis between the 9th and 12th weeks of pregnancy. (Am J Med Genet 1997;70:179-87). So, it is very possible that you and I are mosaic and will not suffer any consequences to our future health but it is much too early to know.

Degrees of Mosaicism

High-level mosaics mean the biopsy sample we took from the embryo had more abnormal cells than normal cells – the success rate with these embryos is lower and the miscarriage rate is higher.

Low-level mosaics have more normal cells than abnormal cells in the biopsy sample and seem to have a higher success rate and a lower miscarriage rate than the high-level mosaics.

The percentage of and which abnormal chromosomes are involved may further define the mosaic embryo. Your genetic counselor can discuss what is known with your embryo(s).

Know Your Options

How we handle mosaicism results is not really agreed upon by all IVF centers. Some may not distinguish high vs. low mosaic embryos and other may not report mosaicism at all. This is a new and evolving field so there are some controversies surrounding the clinical meaning of your test results. Please make sure that you have a conversation with your doctor about your results. The significance of your test results will also depend upon your individual situation. For example a 30 year old woman struggling with multiple miscarriages may never consider transferring a mosaic embryo since she wants to minimize her risks for miscarriage as much as possible, but a 43 year old woman with no other embryos available except one mosaic embryo may feel the 30% live birth rate is her best chance at having a baby using her own eggs. Again, multiple factors can influence your decision.If you have a mosaic embryo frozen in the laboratory, it is important to know your options.

TRANSFER

It seems reasonable to consider if you do not have any normal embryos for transfer. This is a decision that should be discussed directly with your infertility specialist and genetic counselor because each person's situation is unique and very personal. While you weigh the risks and alternatives and because there is no long-term safety data, we recommend for you to undergo another egg retrieval in hopes of obtaining at least one normal embryo rather than using a mosaic embryo.

DISCARD

This option is very reasonable because of the concerns above. If you decide to discard your embryos, you will need to sign an embryo discard form. We encourage you to consider donating to research to help us in our quest to learn more about these embryos and about IVF in general so we can continue to advance the field and help other patients, now and in the future.

POSTPONE

It may also be your option to leave the embryo in storage for now. The advantage of this is that as we learn more about mosaic embryos, you may want to consider using your mosaic embryo in the future. The disadvantage of this is that you may need to pay embryo storage fees each year. If you have other normal embryos in storage this will not cost additional money, but if you do not, this will be an additional expense that may factor into your decision.

Genetic analysis of embryos is advancing at a rapid pace. We don't have all the answers but hopefully, this article will help clarify the decisions that you will need to make if you have a mosaic embryo.

For our podcast episode on this topic,please use this QR code:

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