NEWSLETTER

This newsletter centers on reproductive topics with a genetics focus. If there is an organization or upcoming webinar you'd like me to include in a future newsletter, please feel free to reach out at genetics@modernreproduction.org.

Sign up for the newsletter here

upcoming webinar

Southern Genetic Counselor Webinar Series

"*Genomics and Precision Medicine Policy: Why Genetic Counselors Should Engage in Advocacy*"

August 8th, 3pm EST

Sign Up

NEWSLETTER

The little lit review

1 Opportunities for artificial intelligence in healthcare and in vitro fertilization

Brian Miloski

Per ChatGPT: The article discusses the role of artificial intelligence (AI) as a foundational technology with transformative potential in various industries, including healthcare. It highlights the importance of proper planning, transparency, and accountability in AI implementation, citing examples of failed healthcare AI projects like Palantir's system in the UK and IBM Watson for Oncology. The challenges of data privacy, bias, and human touch in healthcare AI are addressed. The article then explores AI's applications in in vitro fertilization (IVF), focusing on areas such as image analysis, IVF optimization, and sperm analysis. ALQIMI's work in developing AI models for IVF protocol optimization and dosage prediction is discussed, along with challenges faced due to imbalanced data. The article provides recommendations for successful AI implementation, emphasizing clear problem definition, high-quality data, multidisciplinary teams, and iterative refinement. It concludes by highlighting the growing role of narrow AI in improving decision-making and overall quality of life, while distinguishing it from broader discussions around general or autonomous AI.

Link to ALQIMI

AI in IVF

- image analysis
- IVF optimization/predictive analysis
- sperm analysis
- educational content, pre-test counseling

2 Perinatal and postnatal outcomes up to the third year of life after the transfer of mosaic embryos compared with euploid embryos

R. Morales Sabater, B. Lledo , J.A. Ortiz, L. Arenas, A. Bernabeu , J.C. Castillo, R. Bernabeu

Per ChatGPT: The study aimed to compare the perinatal and postnatal outcomes of children conceived after transferring low-grade mosaic embryos to those conceived with euploid embryos in in vitro fertilization (IVF) cycles. The study included 172 children born between October 2017 and August 2022, divided into two groups: euploid (115 children) and mosaic embryo (57 children). Mosaic embryos had a *mosaicism level below 50%*. The study found no significant differences in perinatal outcomes and physical health between the two groups. Maternal age was higher in the mosaic group, but other parameters such as gestational age, birth weight, and Apgar scores showed no significant differences. The incidence of congenital anomalies was similar in both groups and were all minor anomalies. No health problems or chronic diseases were recorded in either group during early childhood. The study suggests that transferring low-grade mosaic embryos can result in healthy children and provides reassurance about their perinatal and postnatal clinical outcomes. However, the study acknowledges the limitation of its sample size and suggests the need for further larger studies to validate these findings.

Are you more or less interested in reading a summary if it is written by chatGPT?

Poll

NEWSLETTER

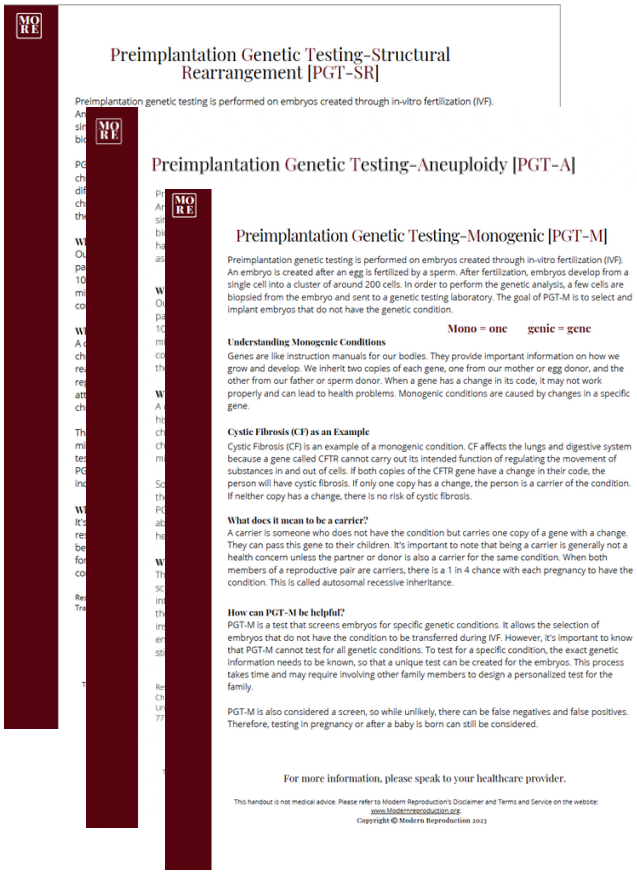
Patient Facing Content

US Fertility Network

More on youtube - the US Fertility Network and Fertility Network UK both have Youtube channels in order to provide free patient video content. Fertility Network UK has more recently published videos and while not US based can still be a helpful resource for patients.

MR's content update:

PGT handouts



Preimplantation Genetic Testing-Structural Rearrangement [PGT-SR]

Preimplantation genetic testing is performed on embryos created through in-vitro fertilization (IVF).

Preimplantation Genetic Testing-Aneuploidy [PGT-A]

Preimplantation Genetic Testing-Monogenic [PGT-M]

Preimplantation genetic testing is performed on embryos created through in-vitro fertilization (IVF). An embryo is created after an egg is fertilized by a sperm. After fertilization, embryos develop from a single cell into a cluster of around 200 cells. In order to perform the genetic analysis, a few cells are biopsied from the embryo and sent to a genetic testing laboratory. The goal of PGT-M is to select and implant embryos that do not have the genetic condition.

Mono = one genic = gene

Understanding Monogenic Conditions

Genes are like instruction manuals for our bodies. They provide important information on how we grow and develop. We inherit two copies of each gene, one from our mother or egg donor, and the other from our father or sperm donor. When a gene has a change in its code, it may not work properly and can lead to health problems. Monogenic conditions are caused by changes in a specific gene.

Cystic Fibrosis (CF) as an Example

Cystic Fibrosis (CF) is an example of a monogenic condition. CF affects the lungs and digestive system because a gene called CFTR cannot carry out its intended function of regulating the movement of substances in and out of cells. If both copies of the CFTR gene have a change in their code, the person will have cystic fibrosis. If only one copy has a change, the person is a carrier of the condition. If neither copy has a change, there is no risk of cystic fibrosis.

What does it mean to be a carrier?

A carrier is someone who does not have the condition but carries one copy of a gene with a change. They can pass this gene to their children. It's important to note that being a carrier is generally not a health concern unless the partner or donor is also a carrier for the same condition. When both members of a reproductive pair are carriers, there is a 1 in 4 chance with each pregnancy to have the condition. This is called autosomal recessive inheritance.

How can PGT-M be helpful?

PGT-M is a test that screens embryos for specific genetic conditions. It allows the selection of embryos that do not have the condition to be transferred during IVF. However, it's important to know that PGT-M cannot test for all genetic conditions. To test for a specific condition, the exact genetic information needs to be known, so that a unique test can be created for the embryos. This process takes time and may require involving other family members to design a personalized test for the family.

PGT-M is also considered a screen, so while unlikely, there can be false negatives and false positives. Therefore, testing in pregnancy or after a baby is born can still be considered.

For more information, please speak to your healthcare provider.

Fertility Skills is an online, free, self paced course on fertility for HCPs