

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](#)

Must Watch
click the images for the links

Fertility and Sterility | JOURNAL CLUB GLOBAL 

Join our discussion **REVISITING THE STAR TRIAL: THE FELLOWS DEBATE PGT-A** Friday, August 25 12:30PM EST

our **PANEL**       our **HOST** 

Dr. Ali Bazzi Dr. Zach Walker Dr. David Huang Dr. Megan Sax Dr. Katie Coyne Dr. Victoria Jiang Dr. Kurt Barnhart

Casual Conversations, Challenging Topics

Stem Cell Transplant as a Curative Treatment for Sickle Cell Disease

Speakers

Timothy Holston, MD PhD
Medical Director, Blood and Marrow Transplant Program, Director of Research, Comprehensive Bone Marrow Failure Center | Children's Hospital of Philadelphia

Kisha Hampton, MPH
Sickle Cell Team Lead at Indiana Hemophilia and Thrombosis Center

Andrea Matthews
Parent & Founder of The Children's Sickle Cell Foundation

Moderators

Marianna Raia, MS, CGC
Expecting Health

Karla Dawson, MPH
Be The Match

Wednesday, August 23, 2023 | 1:00 - 2:00 PM EST

The Referral with **Dr. Karan**

DR HELEN O'NEILL

NEWSLETTER

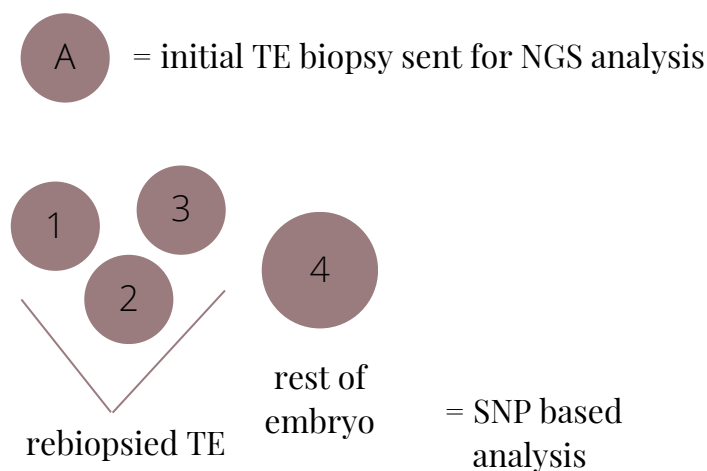
The little lit review

1

Blinded rebiopsy and analysis of non-euploid embryos with two distinct preimplantation genetic testing platforms for aneuploidy

Sarah Druckenmiller Cascante, M.D., Andria Besser, M.S., C.G.C. , Hsiao-Ling Lee, M.S. 11 , Fang Wang, Ph.D. , Caroline McCaffrey, Ph.D., James A. Grifo, M.D., Ph.D.

100 embryos had clinical TE biopsies sent to a lab performing next generation sequencing (NGS) from 2012-2021. Next, each embryo was biopsied 4 times: 3 biopsies were TE and 1 was the rest of the embryo, presumably encompassing the ICM. The rebiopsied samples were analyzed with a SNP (single nucleotide polymorphism) array. One goal was to compare the NGS result to the SNP array rebiopsied results to assess for concordance. Additionally, this approach allowed for identified of presumed biological mosaicism. SNP array did not report mosaicism, so if the rebiopsied samples revealed at least one euploid among 3 aneuploid results, then mosaicism was indicated. Read the paper for all the details, but these are the important takeaways:



1. The idea of complementary chromosome or reciprocal aneuploidy in other parts of the embryo is a reasonable discussion point in counseling as this was identified for both initially mosaic or full aneuploid NGS results and for all alterations (trisomy/monosomy/del/dups). This also supports mitotic errors underlying initial TE results.
2. This study supports the emerging data that segmental aneuploidies may be reflective of mitotic errors with other parts of the embryo being euploid. 30% of segmental aneuploidies (segA) had euploid results on all rebiopsies compared to whole chromosome mosaics which had a similar percent at 35%. Importantly, 45% of segA had aneuploidy results on all rebiopsies.

intermediate copy number = mosaic

as some of you know, there is discussion on whether it is appropriate to use the term "mosaic" to describe the intermediate copy number on NGS profiles. While biological mosaicism could be an explanation for the profile, there are other explanations as well such as "technical artifacts produced by whole genome amplification, excessive use of laser or mechanical trauma during biopsy, sampling of apoptotic TE cells or cell debris". Dr. Treff [writes at length](#) about this issue.

NEWSLETTER

The little lit review



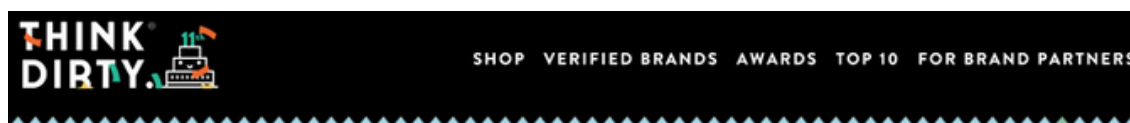
Defining Embryo Donation: An Ethics Committee Opinion (2023)

One of my first sessions reviewing PGT-A results, we ended with the moral status of the embryos as the male partner was not comfortable with the available disposition options. He felt as if his embryos were on the same moral status as a baby and was at odds with this feeling now that they had multiple embryos they were not planning to transfer. He isn't the first or last to feel this inner conflict. Patients have referred to their embryos as "embabies". ASRM's ethics committee came together to discuss embryo donation and to highlight the misleading nature of the term "embryo adoption". One major reason to avoid the use of adoption is because adoption laws may be applied to embryo donation. For example, there is a cooling off period for adoption of a child to allow for a change in mind, yet for embryo donation, the decision is final. This opinion makes salient points regarding the potential burden faced by individuals pursuing egg donation if embryo donation was treated in the same way as adoption. In reading this opinion, I was reminded of the article in a previous newsletter that discussed [individuals' egg disposition choices](#). I think there can be two sides of embryo donation that both recognizes someone's view of the embryos' status to them but also legally how to proceed with these decisions.

NEWSLETTER

Community Content:

Think dirty is an app that helps with identifying what ingredients are in certain products. The goal is to lessen the amount of harmful chemicals we expose ourselves to and allow for the ability to make swaps.



Modern Reproduction Content:

Every week, a blog entry is posted directed to the general population. This past week, a post on carrier screening and a family hx of CF was discussed. The intention of the post is to highlight the importance of obtaining the genetic test report of relatives to compare to the patient's carrier screen to ensure the familial variants were assessed at the lab. The easiest thing to do is to share the relative's report with the lab.



8/15/23

Navigating Family History: Understanding Cystic Fibrosis Carrier Screening

[Read More](#)