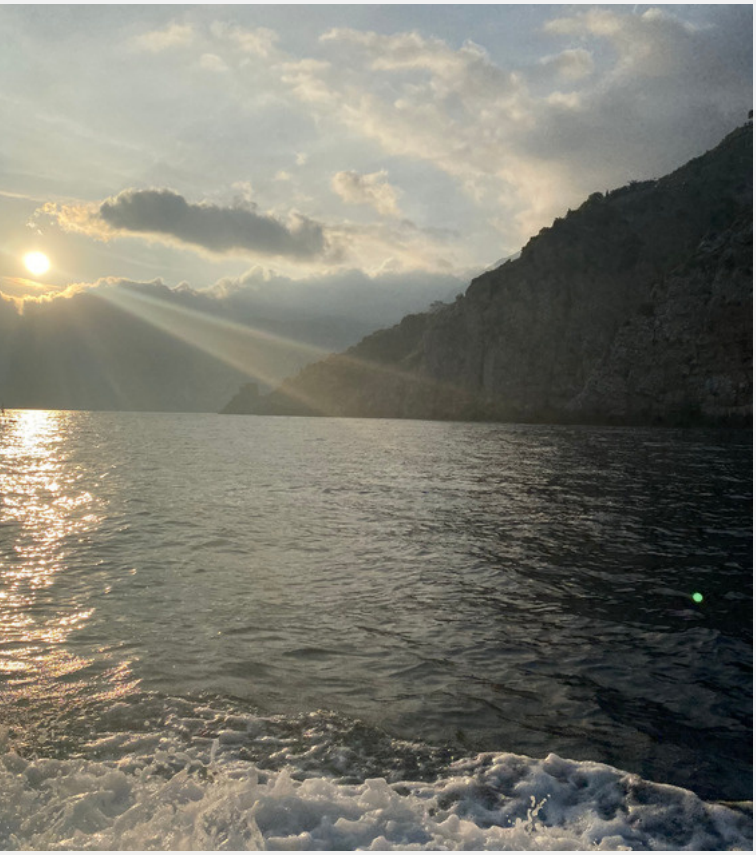
NEWSLETTER



INTRODUCTION

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

What to expect

when you're expecting this newsletter

The little lit review

Upcoming webinars

Patient facing content

Modern Reproduction Updates

NEWSLETTER

The little lit review

1 Pericentric inversion (Inv) 9 variant—reproductive risk factor or benign finding?

Katrina Merrion & Melissa Maisenbacher

52 trophectoderm biopsies from seven couples, of which one member had an inversion, were received for concurrent 24 chromosome PGT-A. 5 females and 1 male had a inv(9)(p11q13) and 1 male had a inv(9)(p12q13). Mean maternal age was 33.3 years. Euploid rate for couples with inversion was 61.5% whereas an age matched control group was 59.3%. Aneuploid rate was 38.5% and 40.7%, respectively. Of the aneuploid results for the inversion group, none involved chromosome 9 and given there was no increase in aneuploidy rate for the inversion group, no interchromosomal effect was observed.

The summary we needed

2 Preconception carrier screening yield: effect of variants of unknown significance in partners of carriers with clinically significant variants

Hila Fridman, MSc, Doron M. Behar, MD, PhD, Shai Carmi, PhD, and Ephrat Levy-Lahad, MD

Expanded carrier screening results do NOT report out variants of uncertain significance as other genetic tests typically do. The impact of a variant of uncertain significance is not clear, so, typically, medical management does not change. In the prenatal setting, since medical management is not expected to change due to this result, it is not routinely revealed on carrier screening reports. VUSs are usually downgraded to benign or likely benign variants.

This study investigated further if this practice inadvertently misses at risk couples (ARC). The study concludes: "expanded carrier screening detects one member of the couple with a pathogenic/likely pathogenic and the other with a coding VUS in ≈7% of couples. Even if only 10% of coding VUS are ultimately reclassified as Pathogenic/Likely Pathogenic Variants, ARC detection rates would increase by ≈20%".

What I found most significant is the idea for reanalysis of expanded carrier screening results prior to each pregnancy to capture if any of the VUSs, if identified, have been reclassified to P/LP. Labs might update reports as they update the variant, but this practice requires the clinician to recontact a patient that may no longer be in their care or have changed their contact information. Reanalysis prior to pregnancy may be one way to navigate ECS and possible VUS reclassification.

NEWSLETTER

Patient Facing Content

Miscarriage Hope Desk

Miscarriage Hope Desk is an online resource for individuals who have experienced recurrent pregnancy loss. The organization provides stories from individuals with similar histories as well as support groups and programs.

PRIVATE FACEBOOK GROUP

JOIN PREGNANCY AFTER LOSS

UPCOMING WEBINAR

In a previous newsletter, I mentioned the new digital health apps that have popped up throughout the recent years. **Femtech Lab** is an organization that helps these apps and groups to be successful, focusing on the business side and networking. They have various events, including webinars.

Sign up here

Blog Post

What should I expect with PGT-M?

Modern Reproduction Updates

What to Expect: PGT-M

Handout