

NEWSLETTER



INTRODUCTION

I don't know about you, but even though I am a genetic counselor, I can't help but want to know about all other topics in reproduction. This leaves me checking numerous emails, newsletters, LinkedIn, Instagram, and more. For my own sanity, I'm compiling what I come across in a week in one place and sharing it with you - with a genetics focus, of course :)

What to expect
when you're expecting this
newsletter

The little lit review

Upcoming webinars

Webinars of the past

Patient facing content

Professional guidelines updates

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The little lit review

①

On the reproductive capabilities of aneuploid human preimplantation embryos

Antonio Capalbo, Maurizio Poli Chaim Jalas, Eric J. Forman, and Nathan R. Treff

The article describes the differences between retrospective, randomized clinical trials, and nonselection studies. It further reviews the current nonselection studies for PGT-A mosaic and aneuploid results. PPV in the PGT-A context is difficult to measure, so the PPV in the article is defined as the likelihood of an aneuploid result to predict embryonic developmental failure.

Retrospective

Participants are divided based on their PGT-A results: euploid, mosaic, and aneuploid. An important consideration is the euploid group tends to have a better prognosis compared to the mosaic group as these participants may not have had a euploid to transfer

Randomized Clinical Trials

One group of participants is the control whereas another group undergoes an intervention, in this case euploid PGT-A results. This approach can compare the efficacy of an alternative clinical strategy but usually is underpowered to calculate PPV.

Nonselection

Experiment group is random and blinded. Results are revealed after outcomes are measured. Most helpful to calculate PPV. However, some may be semi-random if morphology of the embryo is used to select which to transfer.

②

Reproductive Risk Estimation Calculator for Balanced Translocation Carriers

Carolyn Trunca, Nancy R. Mendell, and Samantha L.P. Schilit

Since 1991, Dr. Trunca has offered the free risk assessments through [her site](#) and now, the authors offer a protocol to instruct professionals on how to calculate the translocation risk. There is a clear step by step instruction in the article. Five hypothesis were presented for what variables impact the type of segregation that occurs in meiosis of human translocation carriers.

Conclusion 1:

There is a highly significant relationship between the structural characteristics of a balanced translocation and having a liveborn child with an unbalanced translocation.

Conclusion 2:

Female carriers have a higher risk than male carriers of having a liveborn child with an unbalanced translocation.

Conclusion 3:

Ascertainment category is a strong predictor of the viability of an unbalanced translocation.

Conclusion 4:

The chromosomes involved in a translocation are predictors of risk. Translocations involving chromosomes 9, 11, 13, 18, 21, and 22 are more likely to have a liveborn child with an unbalanced translocation.

Conclusion 5:

Translocations with breaks near the terminal ends are at an increased risk of having a liveborn child with an unbalanced translocation whereas breaks more medially placed are at an increased risk of having a SAB or SB

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UPCOMING

National Society of Genetic Counselors are hosting a webinar on **A New PGT-A Horizon: Segmental Aneuploid Embryos & Their Transfer** with reproductive genetics expert, Amber Gamma.

Sign up [here](#)

The genetic testing lab, **Genomic Prediction**, is offering a webinar on their products discussed at **European Society of Human Reproduction and Embryology** (ESHRE)'s latest conference.

Sign up [here](#)

Healthcare Brew releases a newsletter a few times a week. While not usually on the topic of reproductive medicine, they provide insight on the overall health of the medical system. Their upcoming webinar covers the **state of the industry** in only 30 minutes!

Sign up [here](#)

My favorite organization to stalk, **ELSIhub.org**, continues to provide excellent content for their Friday Forum Series. This week will be **Population Descriptors in Genomic Research: Applying the NASEM Recommendations**. ELSI stands for ethical, legal, and social implications and aligns well with genetics and reproductive medicine.

Sign up [here](#)

Check out Tempus's [AI clinical assistant](#). One area of use is to reduce the need of calling the lab for basic updates.

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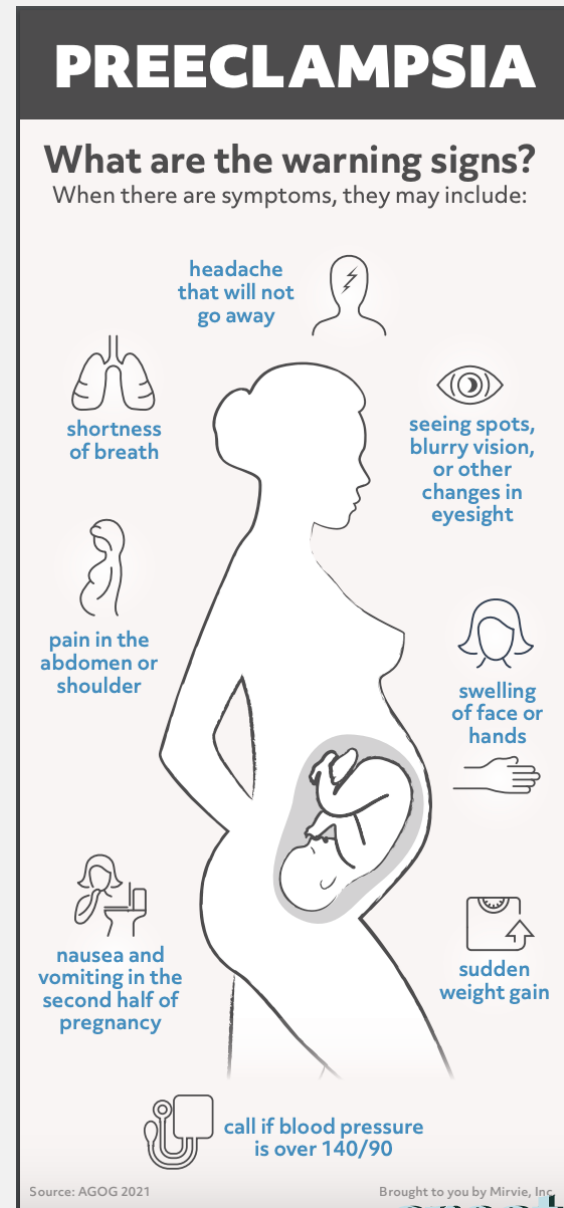
Topic of Discussion: Preeclampsia Detection

The FDA has approved Thermo Fisher's screen for preeclampsia. As shown on Mirvie's brochure, there are a list of symptoms that can indicate preeclampsia, but early identification is crucial in pregnancy health.

Thermo Fisher's webinar **today** at
1pm

Mirvie provides a care plan to prevent preeclampsia on their website in the form of checklists and a AJOG special report.

March of Dimes has a patient facing
article.



created
by Mirvie

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Patient Facing Content

Global Genes

Global Genes created a [toolkit for Family Planning](#), specifically for those who have a rare disease, are known to be carriers, or have a family history. Once clicking the link, you are taken to a page requesting some basic information like an email address as well as which toolkit you would like. The organization also provides content on: Understanding and Navigating Medicare, Caring for Yourself and Your Children, Understanding and Navigating Medicaid, and more.

Genetic Support Foundation

The [Genetic Support Foundation](#) has killer patient facing resources, including descriptions and video content, in English, Swedish, and Spanish. The Genetic Support Foundation offers genetic counseling for patients and to team up with providers for all their genetic counseling needs.

Professional Guideline Updates

ESHRE: GUIDELINE ON THE
MANAGEMENT OF RECURRENT
PREGNANCY LOSS



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