

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.

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

The One to Take Notes:

Fatal fetal anomaly: Experiences of women and their partners ✨
Peter Jackson, Stacey Power-Walsh Rebecca Dennehy, Keelin O'Donoghue

The study interviewed individuals who received a prenatal diagnosis and either decided to terminate or continue the pregnancy. The participants graciously provided insight on their feelings of shock to the initial diagnosis, their struggle with telling other people, the need for a judgement free tribe, and actions by healthcare providers that were helpful to their coping.

The One to Remind You:

Ethics in pre-ART genetics: a missed X-linked Menkes disease case ✨
A.-M. A. Gerdes, L. Birk Møller & N. Horn

This article dives into a case in which an egg donor was initially screened negative for Menkes disease, but subsequently two male infants conceived with her eggs were diagnosed with the condition. Further genetic evaluation revealed the egg donor is a carrier of the condition. The missed identification was said to be due to the variant being undescribed in the literature, thus it was not reported on the initial carrier screening panel. This case reminds us of the nuances and limitations of genetic testing, and the importance of clear and informed consent to manage expectations.

The One to Talk with Your Colleagues About:

Investigating the significance of segmental aneuploidy findings in preimplantation embryos ✨

Ludovica Picchetta, Ms.C., Christian S. Ottolini, Ph.D., Helen C. O'Neill, Ms.C., Ph.D., and Antonio Capalbo, Ms.C., Ph.D.

The segmental aneuploidy result can be one of the more trickier results to counsel patients on. This article addresses the origins of the segmental aneuploidy and reviews current literature. The article suggests one approach to better predict the ICM chromosomal constitution is to rebiopsy the embryo as studies show it is difficult to predict ICM of segmental aneuploidy trophectoderm biopsies as opposed to whole chromosome aneuploid and euploid results. An important improvement would be the development of a database, classifying the phenotypes caused by deletions and duplications, as this is a current challenge for select deletions and duplications.

Important to know:

FDA Approves First Gene Therapy for Treatment of Certain Patients with Duchenne Muscular Dystrophy: click [here](#) for the announcement

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UPCOMING

WEBINAR

National Society of Genetic Counselor's (NSGC) Member Webinar Series will have Vence Bonham, Jr.; Bernice Coleman; Erica Ramos; Katie Johansen Taber; Robert Wildin speak on **Realizing the Full Potential of Health for all Through Genomics and Precision Health: A Discussion with the National Academies Genomics Roundtable**

Sign up [here](#)

EVENT

Empower with Moxi is having a launch party for their new tool, Moxi Matching. The organization is centered on embryo donation education and support

Sign up [here](#)

PREVIOUS

WEBINAR

The American Society of Reproductive Medicine's Genetic Counseling Professional Group has a webinar series. They kicked it off with **The Business of Genetic Testing in Reproductive Medicine** with Kenny Wong as the fantastic and informative speaker. The webinar went into detail of the economics of a lab, including insurance coverage considerations. There was mention of the recent layoffs within the reproductive department of genetic testing labs. These layoffs include genetic counselors that once provided direct patient care. This dynamic pushes us to think if a lab based genetic counseling model fits in with a lab's economic position. There have been at least two articles on this point as well: [DNAExchange](#) and [ASRM](#).

PODCAST

The [Huberman Lab Podcast](#) has a few episodes on fertility, focusing on optimization. Probably every provider in reproduction has gotten the question of "what can I do to increase my fertility" from a patient. Dr. Huberman's neuroscience perspective is unique from fertility specialists and allows for a thorough discussion on hormone's neurological impacts. Environmental exposures are discussed and parallels [ASRM's recent podcast](#) with Dr. Carmen Messerlian, who shares very clear guidance on what to tell patients about diet and every day products to avoid.

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Check out the Scientific Early Life Environmental Health & Development (SEED) Program

where Dr. Messerlian's
team has developed
insanely helpful brochures
on exposures

The below is figure 4 from the article, [Lifestyle interventions to reduce endocrine-disrupting phthalate and phenol exposures among reproductive age men and women: A review and future steps](#)

Strategies	
Dietary	<ul style="list-style-type: none"> • Choose fresh or frozen foods instead of canned, processed, or packaged foods. Avoid drinking beverages from cans or plastic bottles. • Cook more meals at home, instead of eating fast food or takeout. • Buy organic produce, meat, and dairy products, when possible. • Avoid microwaving food and beverages in plastic containers. • Store food and beverages in glass, stainless steel, or ceramic containers, instead of plastic.
Behavioral	<ul style="list-style-type: none"> • Check the labels of personal care and beauty products. Choose products that do not contain phthalates, parabens, or fragrances. • Minimize use of personal care products and cosmetics. • Use mineral-based sunscreens, containing zinc oxide or titanium dioxide, instead of chemical sunscreens. • Avoid perfume, cologne, scented body sprays, and other products with fragrance. • Limit use of nail polish and nail polish remover.
Residential	<ul style="list-style-type: none"> • Reduce household dust using a wet mop or cloth, or HEPA filter vacuum. • Wash your hands regularly, avoid using antibacterial or scented hand soaps. • Choose safer cleaning products. Avoid products that contain scents or fragrances, harsh chemicals, or antibacterial/antimicrobial chemicals. • Avoid scented products including air fresheners, candles, detergents, and soaps.

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Patient Facing Content


Hertility


Hertility and companies like it ([Modern Fertility](#) and [EverlyWell](#)) have the ability to test hormone levels over the counter to better understand fertility potential and alert to possible concerns. Hertility recently provided a webinar for same sex, assigned female at birth (AFAB), couples on the reproductive options available to them. The [webinar](#) is intended for patients, and there is an [accompanying booklet](#).

Digital Health Apps

Digital health apps and memberships are killing it when it comes to producing patient content. There is a long list of the various health apps/memberships out there such as [Clue](#), [Frame Fertility](#), and [Conceive](#). Patients are able to utilize the information provided by the apps as well as, depending on the app, can chat with healthcare providers about certain topics related to reproduction.

Professional Guideline Updates

POSITION STATEMENT FROM THE INTERNATIONAL SOCIETY FOR PRENATAL DIAGNOSIS ON THE USE OF NON-INVASIVE PRENATAL TESTING FOR THE DETECTION OF FETAL CHROMOSOMAL CONDITIONS IN SINGLETON PREGNANCIES 

NONINVASIVE PRENATAL SCREENING (NIPS) FOR FETAL CHROMOSOME ABNORMALITIES IN A GENERAL-RISK POPULATION: AN EVIDENCE-BASED CLINICAL GUIDELINE OF THE AMERICAN COLLEGE OF MEDICAL GENETICS AND GENOMICS (ACMG) 

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