

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.

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when you're expecting this
newsletter

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The little lit review

①

Obstetrical, perinatal, and genetic outcomes associated with nonreportable prenatal cell-free DNA screening results ✨

Norton et al. (Study funded by Natera)

Nonreportable prenatal cell-free screening results with the SNP based analysis were shown in this study to be associated with preeclampsia, preterm birth, and aneuploidy. 17,851 patient results were included in the study with 602 receiving nonreportable results. Of 10 confirmed aneuploidy cases that initially received a nonreportable result, upon a redraw, 1 resulted as high risk, 2 were again nonreportable, and 1 resulted as low risk. Given the findings, diagnostic testing should be offered and surveillance of possible adverse pregnancy outcomes.

②

How does cell-based non-invasive prenatal test (NIPT) perform against chorionic villus sampling and cell-free NIPT in detecting trisomies and copy number variations? A clinical study from Denmark ✨

A.-M. A. Gerdes, L. Birk Møller & N. Horn

Two studies are discussed in this article - the first compared cbNIPT results to CVS results of women who received high risk combined first trimester screening. The other compared cell free NIPT (cfNIPT) and cell based NIPT (cbNIPT) results. cbNIPT showed promising results when compared to CVS results: 35/40 aberrations were detected. The 5 undetected were mosaic. When compared to cfDNA screening, the failure rate for cbNIPT was 7.8% versus cfDNA's 2.8%. 30mL of blood was obtained for cbNIPT compared to 20mL for cfDNA analysis. cbNIPT was able to differentiate between maternal and fetal cells and had lower CNV size detection compared to cfDNA. There is a commercial offering of this test in Denmark and possibly one lab in the States, yet it is not yet a widely utilized test given the need for more studies.

③

'I know it's not normal but it's normal to me, and that's all that matters': experiences of young adults conceived through egg donation, sperm donation, and surrogacy ✨

V. Jadv, C. Jones, P. Hall, S. Imrie and S. Golombok

The study focused on a cohort of young adults, who were enrolled in the UK Longitudinal Study of Assisted Reproduction Families, and either born via sperm or egg donation, gestational surrogacy, or traditional surrogacy (where the surrogate also provided the egg cell). The adults in the cohort were mostly informed of their conception around the age of 4. Of the 35 individuals, 16 searched for information on conception and 10 had contact with the surrogate or donor. Contact may have been a relationship or social media friends with limited to no contact. 21 participants were unconcerned with their conception whereas 14 felt special or unique. Of note, 2 participants, one born through traditional surrogacy and the other through egg donation, felt their relationship to their mother was affected by the lack of genetics. No participants felt this way for their relationship to their father. Overall, this study provides important insight to counseling and discussion points for families.

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UPCOMING

Repro Facts will have an Expert Debrief of the publications discussed at ESHRE 2023. It is a FREE event on Saturday July 8th (5am EST). The organization has [slide decks](#) of previous conferences with Practice Points on each slide and videos!

Sign up
[here](#)

Of Note:

ACMG has put out a [naming recommendation](#): **Prenatal cell-free DNA screening** is to be the term used as opposed to the other names for the test.

PREVIOUS

A few months ago, Lettercase National Center for Prenatal and Postnatal Resources at the University of Kentucky's Human Development Institute released a course for [Understanding Disability and Best Practices for Communicating a Diagnosis](#). Within the thoughtful curriculum is the introduction to the history of disability rights, basic phenotype and psychosocial aspects of genetic conditions, prenatal testing history, best practices for communicating a prenatal/postnatal diagnosis, and more. The organization provides CEUs for Genetic Counselors.

As most labs offer webinar series and CEUs in order to market their products and educate, Natera also has a [collection of webinars](#). One webinar of interest was [Integrating Single-Gene NIPT into Prenatal Screening](#). It was published last June and highlighted some cases of positive results. This webinar coincides nicely with the latest publication on single gene NIPT: [Single gene non-invasive prenatal screening for autosomal dominant conditions in a high-risk cohort](#). Of note, the webinar stated single gene NIPT could be an adjunct to diagnostic testing and, depending on the results, determine what additional testing to consider. The possibility of a false negative result was not addressed as a concern for this approach when it should be given the screening nature of single gene NIPT. The recent publication states in the conclusion that single gene NIPT should not replace clinically indicated diagnostic tests. The article demonstrates cases in which the screen lead to earlier diagnosis and enhanced surveillance.

[Vistara Gene List](#)

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Hot Topic of Discussion: **Embryo Models/Synthetic Embryos**

Without trying, this topic has come up for me multiple times this past week from various sources. This page is the lowdown.

Short Story:

Manipulating skin stem cells into gamete cells has been on the horizon for the last few years, yet more recently, there has been the creation of stem cell based embryos models. The intention of these models are not initially to replace IVF rather to be a way to study early embryo development, miscarriage, and placental function. This [handout](#) is a helpful visual for the process.

The actual stories on the topic:

- ✦ Heartbeat and blood reportedly observed in human stem-cell-based embryo model
- ✦ The ISSCR Statement on New Research with Embryo Models
- ✦ New York Times: Scientists Debut Lab Models of Human Embryos
- 🔊 Synthetic human embryos: can the law keep pace with the science? – podcast
- ✦ The New Yorker: The Future of Fertility
- ✦ Lab-Made Human Eggs and Sperm: Dishing Up a Eugenic Future?

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Patient Facing Content

Pregnancy Loss


Finding support after experiencing pregnancy loss is important. One way to find resources in your area for your patients is by searching for "pregnancy loss support in your area". Many hospitals offer support groups that focus on specific types of losses. There are also websites like [Miscarriage Hope Desk](#), [Pockets of Light](#), and [Poppy Seed Health](#) that provide support groups and information for families going through pregnancy loss.

Embryoman

Embryoman is not new - he's been around for a few years compiling and interpreting research in the IVF space on his website, [Remembryo](#), and posts on social media for those updates. As a healthcare provider, it's helpful to see what articles he pulls, and I'm sure tremendously helpful for patients with a desire for all the scientific information.

Professional Guideline Updates

UPDATED RECOMMENDATIONS FOR CFTR CARRIER SCREENING: A POSITION STATEMENT OF THE AMERICAN COLLEGE OF MEDICAL GENETICS AND GENOMICS (ACMG) 

EXPANDED CARRIER SCREENING FOR REPRODUCTIVE RISK ASSESSMENT: AN EVIDENCE-BASED PRACTICE GUIDELINE FROM THE NATIONAL SOCIETY OF GENETIC COUNSELORS 

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