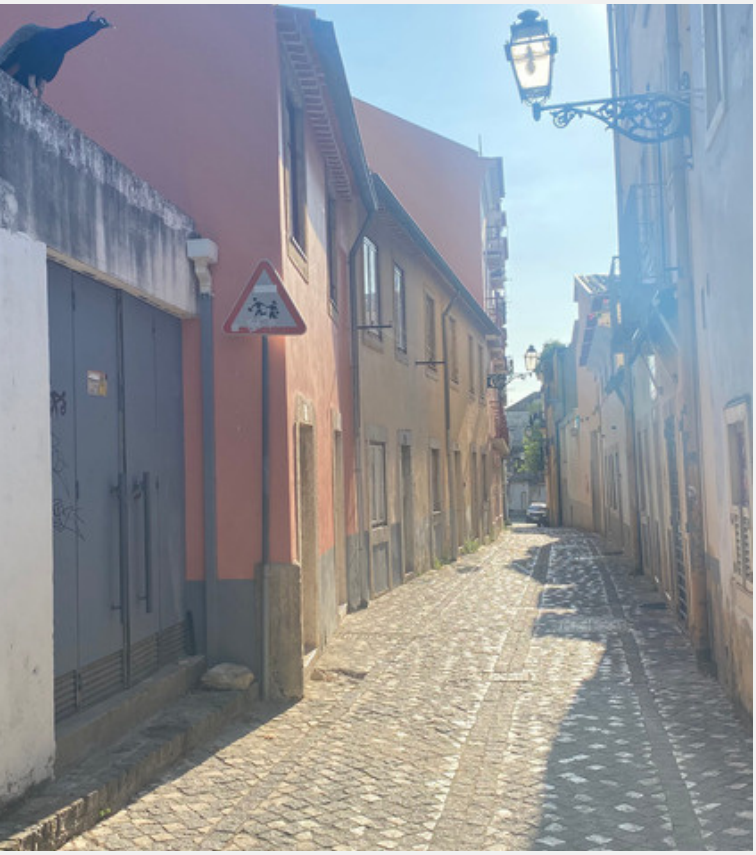
NEWSLETTER



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

NEWSLETTER

The little lit review

1 Prenatal testing for imprinting disorders: A laboratory perspective

Jasmin Beygo, Silvia Russo, Pierpaola Tannorella, Gijs W E Santen, Andreas Dufke, Elia Schlaich, Thomas Eggermann

The article reviews the mechanisms of imprinting disorders and testing to perform in pregnancy. There are helpful visuals, including a chart of the various imprinting disorders and the mechanisms.

Mechanisms

Uniparental Disomy:

- Often the result of trisomy rescue - maternal age and familial structural chromosome variants are risk factors.
- Robertsonian translocation carriers involv chr 14 and 15 have a 0.6-0.8% or less risk for UPD in offspring
- Clinically relevant for chr 6, 7, 11, 14, 15, and 20 as well as autosomal recessive pathogenic variants that would be in homozygote state

Single nucleotide variants or copy number variants in the expressed allele; can be de novo or inherited

Imprinting Defects:

- ART has a correlation to aberrant methylation markers but the cause is not yet clear, particularly for Beckwith Weidemann syndrome.
- Pathogenic variants in the subcortical maternal complex are being identified to cause reproductive failures, aneuploidy, and imprinting disorders.

When to Consider

-Parental Robertsonian translocation carriers, specifically invol 14/15
-Positive NIPT/CVS for aneuploidy and subsequent euploid amnio results
-Omphalocele (Beckwith Weidemann syndrome, neonatal diabetes mellitus, and KOS14 suspicion)
-Placental mesenchymal dysplasia - BWS and KOS14
-Visceromegaly, macroglossia, and placentomegaly, IUGR, oligo or polyhydramnios also consider imprinting disorders
-Family history or predisposing genetic constitution for imprinting disorders

2 Elective egg freezers' disposition decisions: a qualitative study

Lucy E. Caughey, B.Beh.Sc.,Katherine M. White, Ph.D, Sarah Lensen, Ph.D., Michelle Peate, Ph.D.

The article discusses individuals who underwent elective egg freezing and are either deciding on future, current, or past disposition. The article interviewed 31 individuals and provided immense detail and quotes on the individuals' thought processes. Often, an individual's first disposition preference changed from their final disposition preference and a "trigger" event occurred when they made their final decision such as storage limits. Select individuals decided for reclaiming of their oocytes which allowed them to take the oocytes home and have a ceremony. The authors suggest disposition counseling with a focus on grief at the beginning and end of the process.

NEWSLETTER

UPCOMING

While this newsletter is intended for an American audience, I find other countries' updates and approaches an important part of education. Their approaches may be in the context of their insurance coverage but may invoke innovative ideas for us. **The Progress Educational Trust** (PET) organization is a great resource and have an upcoming webinar on, their one year update on Women's Health Strategy.

Sign up here

The Clinical Training Center for Sexual and Reproductive Health offers multiple educational resources, including a podcast on coding. They've included worksheets to help keep track of the various coding options. The latest was on **early pregnancy loss billing and coding**.

Check it out here

The Genetic Counseling Experience Initiative offers journal club meetings, primarily intended for prospective genetic counselors, yet is open to everyone. Their upcoming journal club will be **Chromosomal Abnormalities in Couples with Primary and Secondary Infertility** on Sunday July 23rd at 3pm EST

Check it out here

As you've probably seen, the FDA has approved an over the counter birth control pill.

NEWSLETTER

Topic of Discussion: PGT-A result **Segmental Aneuploidy**

What is a segmental aneuploid result?

Labs have their own set of guidelines of how to report out PGT-A findings. They may utilize different categories of results and cut offs for each. For example, Cooper Genomics uses the range of 20-80% for mosaic reporting whereas Igenomix uses 30-70%.

Segmental Aneuploid is intended to mean a result in which majority of the DNA tested has a specific segmental abnormality present. There is a separate category of result called segmental mosaic which has its own interpretation, different from segmental aneuploidy.

Recent and previous studies have found the concordance rate of the inner cell mass and the trophectoderm are lower for segmental abnormalities results compared to whole chromosomes abnormalities. When a result reveals a whole chromosome abnormality such as Trisomy 1 in the trophectoderm, then it is likely that the inner cell mass also has Trisomy 1. However, if a 15p duplication is present in the trophectoderm biopsy, it is less certain that the inner cell mass also has the duplication. It is possible the inner cell mass is euploid. One explanation for the reduced concordance may be the origin of these abnormalities. Whole chromosome abnormalities are believed to be meiotic in origin whereas segmental abnormalities may be mitotic.

Given the emerging data, it may be that the segmental aneuploid result will have different transfer recommendations than a whole chromosome abnormality. More studies are needed, particularly focusing on reproductive outcomes.

OG Segmental Study

MORE RECENT:

The concordance rates of an initial trophectoderm biopsy with the rest of the embryo using PGTseq, a targeted next-generation sequencing platform for preimplantation genetic testing-aneuploidy

Investigating the significance of segmental aneuploidy findings in preimplantation embryos

NEWSLETTER

Patient Facing Content

Rescripted

Rescripted has a newsletter, blog posts, support groups, "Ask Me Anything" (AMAs) with healthcare professionals, two podcasts, and what I find most notable, a poll section with responses. One of the best ways to improve patient care skills is by hearing directly from patients what does and does not work for them.

Professional Guideline Updates

ESHRE: GUIDELINE ON UNEXPLAINED INFERTILITY

Sign up for updates here

Comprehensive prenatal genetics resources for healthcare providers -The Jackson Laboratory