NEWSLETTER

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

Webinars:

AI IN GENOMICS - PHENOTIPS PODCAST

5/22 AT 12PM EST

Register

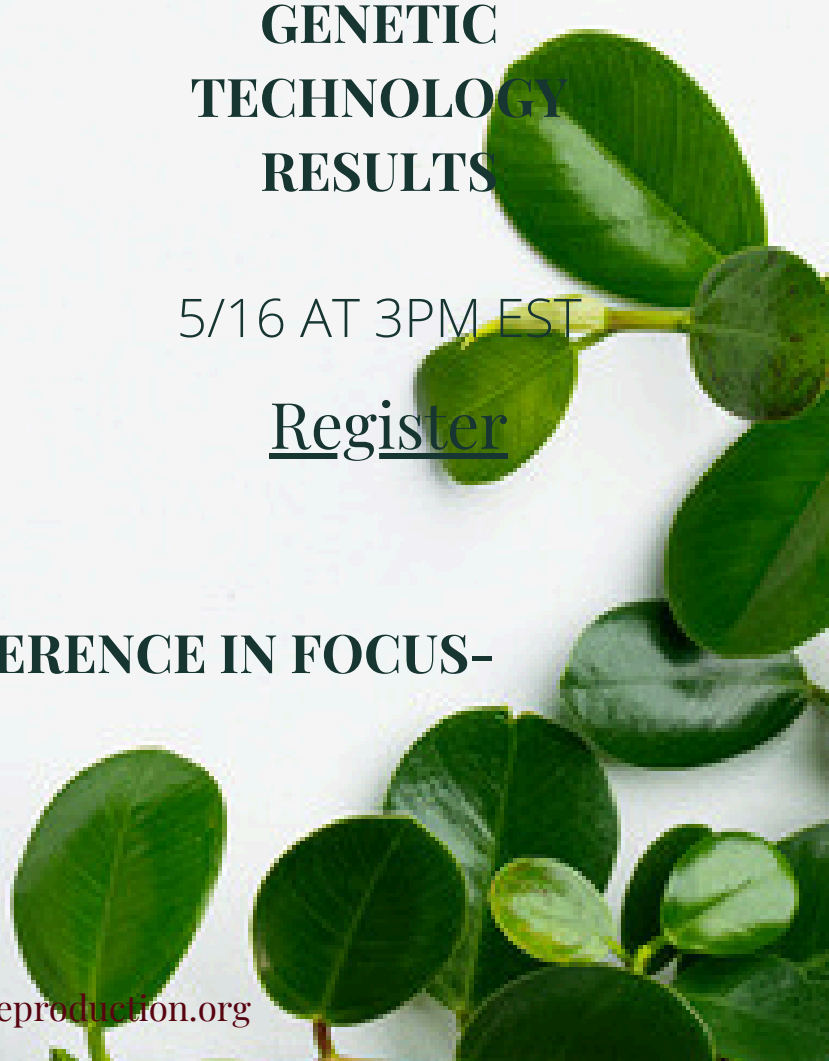
A GUIDE TO UNDERSTANDING GENETIC TECHNOLOGY RESULTS

5/16 AT 3PM EST

Register

KEEPING PATIENT PREFERENCE IN FOCUS- CGTLIVE

On Demand



ISSUE 32

NEWSLETTER

The little lit review

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Artificial intelligence and machine learning in precision and genomic medicine

Sameer Quazi

The article addresses how AI is being used in medicine. Precision medicine is a combination of human genetics, lifestyle, gene expression, and environment. To accomplish the goals of precision medicine, we need the tools to make sense of all the data. AI has the ability to use analytical skills to solve problems, prediction, dimensionality, data integration, reasoning about underlying phenomena, and turn data into clinically actionable knowledge. There are 10 algorithms most used in this setting - the article goes into detail of each and how they have been helpful in medicine. Using predictive tools is not new for our field - cancer predictive models are one of the most common such as Tyrer-Cuzick and BOADICEA. Below are ways that AI is being integrated into genomics, but the article also focuses on cardiology and oncology.

Precision Medicine and Genomics:

- Precision medicine integrates genetics, behavior, and environment for patient-specific treatment interventions, with machine learning aiding in data analysis and cost reduction.

Genome Sequencing:

- Machine learning improves sequencing accuracy by predicting DNA-binding rates and identifying sequencing errors.

Phenotyping:

- Machine learning assists in extracting and analyzing phenotypic information from electronic health records (EHR) and refining phenotype classification.

Variant Calling and Identification:

- Deep learning models enhance accuracy in variant calling, particularly in identifying somatic genetic variations and copy number variations (CNVs).

Functional Genomics:

- Machine learning methods classify and identify genetic features crucial for clinical genome analysis, such as splice sites, promoters, and enhancers.

Prediction Tools:

- In silico tools use machine learning to predict the impact of genetic variants on protein function and disease, aiding in clinical decision-making.

Improving Clinical Genomics:

- ML-driven advancements aim to improve accuracy, reliability, and clinical utility across various stages of genomic analysis, from sequencing to variant interpretation.

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The little lit review

Variation in the Incidence and Type of Full versus Mosaic Segmental Aneuploidies Identified in Blastocysts Undergoing PGT-A

Carly Cuman, Mark P. Green, Elissa Willats, Tristan Hardy, Deirdre Zander-Fox

The article characterizes the details of segmentals in the setting of PGT-A testing using Veriseq's PGS Illumina kit and sureplex amplification. The mosaic range used was 20-70% and segmental >10Mb were detected. The authors retrospectively evaluated the results of 8153 embryos from 4 clinics. The authors were interested in characterizing segmental results given studies that show less concordance of the trophectoderm (TE) biopsy to the inner cell mass (ICM and the greater reproductive potential of segmental mosaics compared to whole chromosome aneuploid mosaics.

682 of the 8153 embryos resulted as segmental. More deletions were reported than duplications. Larger chromosome were more often deleted or duplicated than small chromosomes. There were more mosaic segmental results than full segmental. Lower TE scores from embryo grading has a higher segmental rate.

The importance of these findings can be to compare the results with current hypotheses of segmental abnormalities. It's been discussed that segmentals are not only meiotic in origin as most whole chromosome aneuploids but also mitotic, thus the potential cause of the lower concordance rates between TE and ICM. There is also discussion that segmentals are the result of dividing cells or unrepaired DNA errors that may be fixed later in development. Segmentals are the hot new area of research in PGT-A.

NEWSLETTER

Community Content:

Natera has added the option to assess RhD via cell-free DNA screening. Their validation study is available upon request..

YOUR PREGNANCY haven

Your pregnancy haven is a patient resource - they are currently hosting a pregnancy after loss summit for free. They also have blog posts on pregnancy after loss among other topics.