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.

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WHEN TO STOP STORAGE: IMPROVING CONVERSATIONS ABOUT UNUSED EMBRYOS

On Demand

IVIRMA ASSISTED REPRODUCTIVE SYMPOSIUM

5/8-5/11

Register

ISSUE 31

NEWSLETTER

The little lit review

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Multiplexed Serum Biomarkers to Discriminate Nonviable and Ectopic Pregnancy

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The article addresses a much needed area of research: early pregnancy. Women are learning about their pregnancies even before their missed period given app trackers and fertility treatments. However, the current methods of confirming pregnancies and location are transvaginal ultrasound and serial hCG levels. These assessments may be delayed to later gestational ages or potentially misdiagnose viability.

To address this paucity of options at this stage of pregnancy, the authors looked to biomarkers to predict viability and location of a pregnancy. Biomarkers are not novel in the space of reproduction as multiple marker screens have been utilized for chromosomal abnormality detection for years as well as potential predictors of adverse pregnancy outcomes

. The authors utilized multiple machine learning based methodologies to identify the one that used the least number of markers with the greatest test characteristics, focusing on accuracy. Models can be adjusted to maximize desired test characteristics. For the authors, accuracy was the focus and the Classification Tree Regression Analysis (CART) model won out.

It predicted viability with a 97% accuracy rate and location at 94%.

The markers that proved most predictive for viability were PSG3, which is produced by the trophoblast, CG-alpha, produced from the pituitary gland, and PAPP-A, involved with trophoblast function, collagen breakdown, and cell structure.

For location, sFLT, involved in abnormal angiogenesis, TPF12, involved in structural constitution of the cell matrix, and PSG3 were identified.

The patient population were 218 individuals that were experiencing abdominal pain, vaginal bleeding, or both from 4-10 weeks gestation by ultrasound or LMP.

The markers were identified from unbiased proteomic discovery and Olink, a high multiple immunoassay panel, for those that are involved in trophoblast cell function, angiogenesis, cell adhesion, and extracellular matrix constituents.

Ultimately, the authors are solving an important problem and have a great noninvasive solution.

NEWSLETTER

The little lit review

45,X/46,XY mosaicism: Clinical manifestations and long term follow-up

Ebba Alkhunaizi | Jenna Plamondon Albrecht | Mahmoud Aarabi | Selma F. Witchel | Diane Wherrett | Riyana Babul-Hirji | Annie Dupuis | Lyne Chiniara | Eric Chater-Diehl | Mary Shago | Cheryl Shuman | Aleksandar Rajkovic | Svetlana A. Yatsenko | David Chitayat

The article explores the phenotype of individuals with mosaicism for 45,X and 46, XY and were either postnatally or prenatally diagnosed. The authors performed a retrospective chart review of three institutions from 1984-2021. 100 patients were included: 47 raised as female and 53 raised as male. Patients with other chromosomal abnormalities, limited follow-up data, or were diagnosed subsequent to an infertility workup in adulthood were excluded. The authors recorded the various manifestations experienced by the patients.

Of note, prenatally diagnosed patients were identified either after positive maternal serum screening for Trisomy 21, advanced maternal age, increased nuchal thickening/pleural effusions, or a family history of a genetic condition. 3 prenatally diagnosed cases of full 45,X were later found to have mosaicism for 46, XY; postnatal testing can be considered for these cases.

TABLE 2 Renal and cardiac abnormalities observed in patients who underwent diagnostic testing.

Renal abnormalities		Cardiac abnormalities	
Abnormality	**No. of patients**	**Abnormality**	**No. of patients**
Duplex collecting system	4	Coarctation of the aorta	4
Horseshoe kidney	7	Bicuspid aortic valve	6
Vesicoureteral reflux	1	Hypoplastic left heart	4
Pelvic kidney	1	Dysplastic aortic valve	1
Ectopic kidney	1	Parachute and hypoplastic mitral valve	1
Malrotated right kidney with moderate hydronephrosis	1	Hypoplasia of transverse arch	2
		Mitral and aortic valve stenosis	1
		Ventricular septal defect	2
		Ventricular tachycardia	1
		Right coronary artery fistula	1

out of 78

out of 61

Note: Each cardiac abnormality is listed separately, and number of patients is more than 18 because some patients had more than one abnormality.

TABLE 1 Description of patients with gonadoblastoma.

ID	Reared sex	Age	Reason for evaluation	Ultrasound: uterus	Ultrasound: ovaries	Histology	Follow up
T-003	Female	16	Primary amenorrhea	Small	Right: normal Left: non-visualized	Right: dysgenetic with gonadoblastoma and dysgerminoma Left: dysgenetic with gonadoblastoma	Normal CT and tumor markers
T-036	Female	13	Short stature	Hypoplastic	Non-visualized	Bilateral ovarian mucinous cystadenomas and epithelial tumors	None
T-056	Female	15	Delayed puberty, short stature	Normal	Right: non-visualized Left: hypoplastic	Right: dysgenetic gonad with gonadoblastoma Left: dysgenetic gonad	None
T-057	Female	6	Behavioral problems	Normal	Unilateral ovarian mass	Bilateral dysgenetic with gonadoblastoma and dysgerminoma	None
P-084	Female	8	Short stature, behavioral issues	Normal	Non-visualized	Right: predominant ovarian elements Left: gonadoblastoma	None

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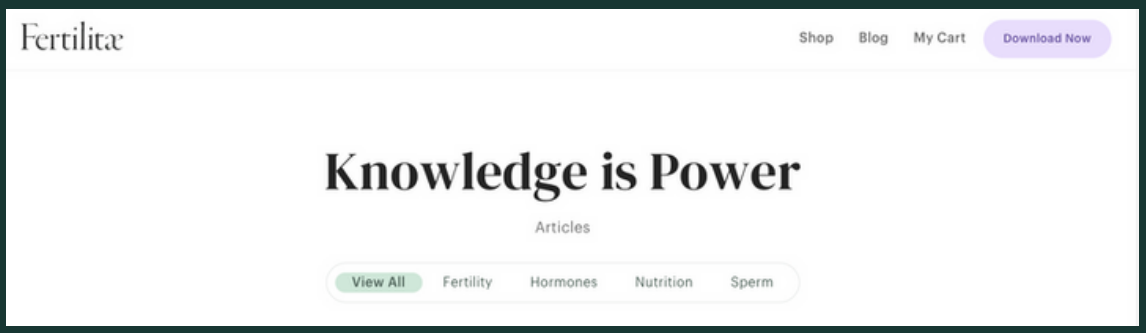
Community Content:



The Fertilitae app and website has helpful content for patients that are trying to conceive. The app is personalized for each patient's journey, and there are healthcare providers accessible through the app, including myself.

Modern Reproduction Content:

Previous newsletters are found on the site as well as LinkedIn, but why search for them when you can sign up to receive them right to your inbox :D

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