

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

COGEN -IVF WORLDWIDE ONLINE CONGRESS

This is the same conference I linked to last week - it was AMAZING. It deserves all the attention for this week's webinar section.

[On Demand](#)

NEWSLETTER

The little lit review

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“It becomes your whole life”—Exploring experiences of reciprocal translocation carriers and their partners

Marta Cifuentes Ochoa, Nicola Jane Flowers, Mark Domenic Pertile, Alison Dalton Archibald



Approximately 1 in 500 individuals have a balanced, reciprocal translocation. The authors explored the experience of individuals with a reciprocal translocation and their partners. 13 semi-structured interviews were conducted.

There are few recurrent translocation but most are unique to the individual. When having a translocation, risks included infertility, recurrent miscarriage, and ongoing pregnancy with unbalanced translocation. The authors asked the following questions:

*How did you find out that you (or your partner) were a carrier?
How was it for you to find out that you (or your partner) were a carrier?
What does being (or your partner being) a carrier mean for you/your family?*

5 themes emerged:

1. experiencing emotional and reproductive impacts of carrying a reciprocal translocation
2. seeking information and support
3. contemplating and experiencing pregnancy
4. adjusting and adapting
5. contemplating carrier status in their children

“Results suggest that testing for translocations earlier in the process of reproductive investigations (i.e., rather than waiting for multiple miscarriages) may minimize the psychological impacts of pregnancy loss and provide couples with useful information that may empower them to make informed decisions about pregnancy planning and reproductive interventions.”

“while it may be useful to have an explanation for patient's reproductive difficulties, grief and loss that may accompany these results as patients realize their reproductive path may be more complex.”

“It's just been like an uphill battle, and it's been full of grief and frustration and difficulty and ... as it is we have had six pregnancies for one child and that's been really tough so, we are tired, very tired of it, yeah”

“I've been trying to have a baby and had multiple mis-carriages...I went to have a fertility discussion about having so many miscarriages and he ran some tests and they confirmed that I had a balanced translocation...I went into doing PGT, but the problem was, I just wasn't producing enough follicles to have very good cycles, you know, the problem with PGT is you need volume and I wasn't producing the volume...”

We got some conflicting information about what the odds of a successful pregnancy were, and it felt like the information is not necessarily out there or interpreted the same by different professionals

NEWSLETTER

The little lit review



Actionability of unanticipated monogenic disease risks in newborn genomic screening: Findings from the BabySeq Project

Robert C. Green, Nidhi Shah, Casie A. Genetti, Timothy Yu, Bethany Zettler, Melissa K. Uveges, Ozge Ceyhan-Birsoy, Matthew S. Lebo, Stacey Pereira, Pankaj B. Agrawal, Richard B. Parad, Amy L. McGuire, Kurt D. Christensen, Talia S. Schwartz, Heidi L. Rehm, Ingrid A. Holm, Alan H. Beggs, and The BabySeq Project Team

Infant	Presentation	Gene	Disease intervention
1	well baby	<i>ELN</i>	supravalvular aortic stenosis, AD; screening cardiac ultrasound
2	well baby	<i>BTBD</i>	biotinidase deficiency, AR; biotin supplementation
3	ICU admission for tetralogy of Fallot, pulmonic stenosis, cryptorchidism	<i>GLMN</i>	glomouvenous malformations, AD; monitoring for lesions
4	ICU admission for aortic coarctation	<i>G6PD</i>	glucose-6-phosphate dehydrogenase (G6PD) deficiency, XLR; drug safety interventions
5	well baby	<i>TTN</i>	dilated cardiomyopathy, AD; screening cardiac ultrasound
6	well baby	<i>TTN</i>	dilated cardiomyopathy, AD; screening cardiac ultrasound
7	well baby	<i>TTN</i>	dilated cardiomyopathy, AD; screening cardiac ultrasound
8	well baby	<i>TTN</i>	dilated cardiomyopathy, AD; screening cardiac ultrasound
9	ICU admission for hypoplastic left heart syndrome	<i>BRCA2</i>	hereditary breast and ovarian cancer syndrome, AD; screening mammography
10	well baby	<i>BRCA2</i>	hereditary breast and ovarian cancer syndrome, AD; screening mammography
11	ICU admission for neonatal pneumonia	<i>SLC7A9</i>	cystinuria, AD; hydration, urinary alkalization, thiol medications
12	well baby	<i>KCNQ4</i>	non-syndromic hearing loss, AD; audiologic screening, hearing aids/implants
13	well baby	<i>VCL</i>	dilated cardiomyopathy, AD; screening cardiac ultrasound
14	well baby	<i>CD46</i>	atypical hemolytic-uremic syndrome, AD; screening, plasma exchange, anti-C5 monoclonal antibody treatment
15	well baby	<i>MYBPC3</i>	hypertrophic cardiomyopathy, AD; screening cardiac ultrasound
16	ICU admission for respiratory distress	<i>MSH2</i>	Lynch syndrome, AD; screening colonoscopy
17	ICU admission for extreme prematurity	<i>CYP21A2</i>	congenital adrenal hyperplasia due to 21-hydroxylase deficiency, AR; anticipatory guidance, glucocorticoid therapy

159 babies were enrolled in the genome sequencing arm of the study, including “healthy” babies and babies in the NICU.

- 1/159 had a likely pathogenic or pathogenic variant that explained their presentation.
- 17/159 had unanticipated monogenic disease risks (uMDRs).
 - 3/17 upon further investigation did have subclinical findings.

The authors tracked immediate and long-term medical outcomes for these infants and their families over 3–5 year to understand the impact of the recommended intervention given the results.

“The concept of actionability is difficult to discuss because for some the ability to provide enhanced surveillance or even knowledgeable anticipation justify a broader definition, while for others this concept only includes conditions where a treatment can slow or stop progression or unequivocally improve the individual’s prognosis”


In the previous review, patients felt better able to adapt when learning about their translocation carrier status earlier. This sentiment has been expressed for x-linked carriers as well. Does this constitute actionability?

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Community Content:



Learn Explore About Connect Talk To CPN En Español For Clinicians

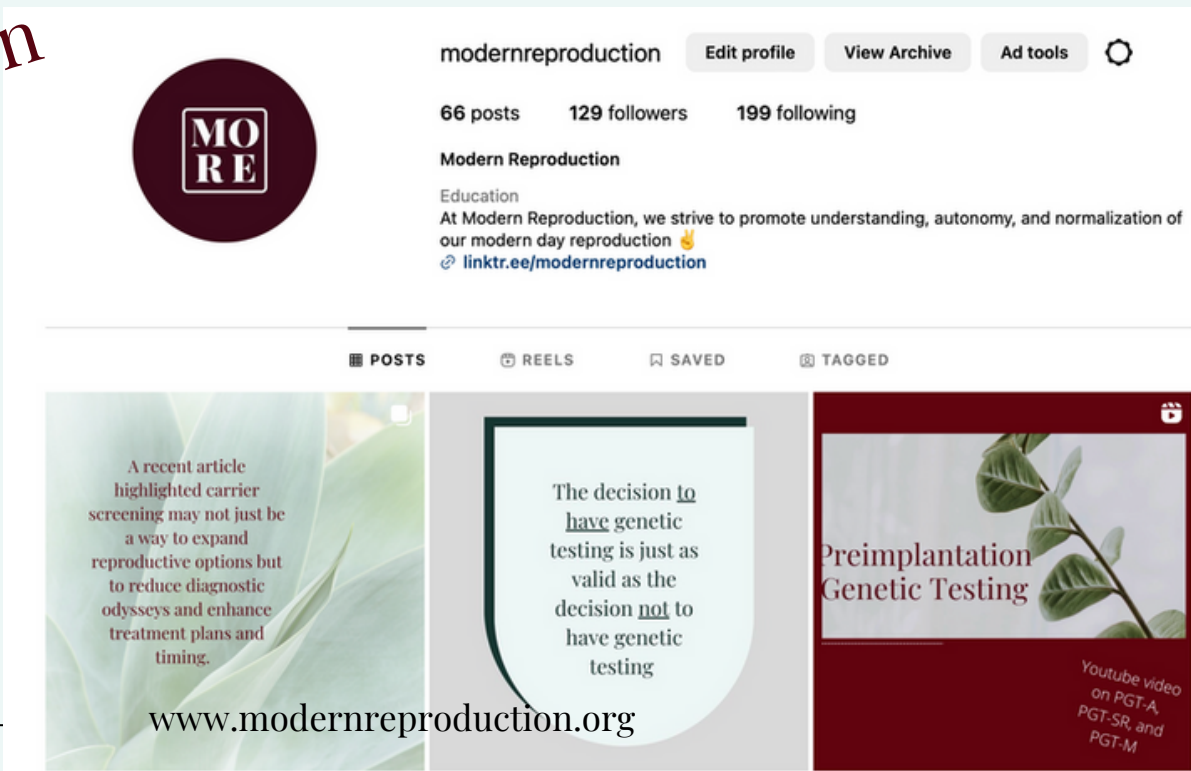


WELCOME.

Courageous Parents Network is a non-profit organization that orients and empowers parents and others caring for children with serious medical conditions, by providing resources and tools that reflect the experience and perspective of other families and clinicians. Here—in videos, podcasts, printable guides, Guided Pathways, and blog—you will find wisdom from families and pediatric clinicians to help you feel you are doing the very best you can for your child and family.

[INTRO VIDEO](#) [TOUR CPN](#) [TALK TO US](#)

Modern Reproduction Content:



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Education
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POSTS REELS SAVED TAGGED

A recent article highlighted carrier screening may not just be a way to expand reproductive options but to reduce diagnostic odysseys and enhance treatment plans and timing.

www.modernreproduction.org

The decision to have genetic testing is just as valid as the decision not to have genetic testing

Preimplantation Genetic Testing

Youtube video on PGT-A, PGT-SR, and PGT-M