

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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Webinars

THE IMPORTANCE OF INCLUDING HEMOGLOBINOPATHIES AS A PART OF CARRIER SCREENING PANELS

Cathi R. Franklin, MS, CGC
with Christine Isaacs, MD

Listen

POPULATION GENOMICS IN CLINICAL PRACTICE

A panel discussion with Dr. Eric Green, Dr. Julian Barwell, and Dr. Fiona Brinkman, hosted by Kira Dineen.

SEPT 28, 2023
| 11am EST

Register

NIPOC: NEW NON-INVASIVE ANALYSIS OF FETAL DNA OF MISCARRIAGE

Miguel Milan Sanchez (as speaker) and Nasser Al-Asmar Piñar, MSc., Ph.D.

SEPT 26, 2023
| 7am EST

Register

NEWSLETTER

The little lit review

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Should we use CRISPR gene editing in human embryos? 

Nada Kubikova, D.Phil., David L. Keefe, M.D., Dagan Wells, Ph.D., Kutluk H. Oktay, M.D., Ph.D., Eve C. Feinberg, M.D.

I love the format of this piece, the pro and con side are side by side to invoke the sense that a real debate is going on.

PRO

Change the paradigm of prevention. Rather than termination of pregnancies via CVS or amniocentesis or embryo discarding via PGT analysis, prevent the condition via germline editing.

Germline editing is advantageous over postnatal treatment as some conditions already disrupt development and health during embryo/prenatal development as well as the treatment would proliferate to all cells during embryonic development whereas postnatally, administering to all organs may prove difficult.

Since all cells of the embryo are treated, this would include its germline cells and effectively rid of the chance to pass the variant to the next generation as well.

Cost of removing the variant is likely less than the cost of having the condition, especially for future generations. For universal healthcare systems, this could be something to consider.

CON

While editing is most useful at the earliest stages of embryo development, this is the time the embryo is most vulnerable to DNA damage, presumably because of deficiency in DNA repair present before the activation of the embryonic genome.

Several papers report large deletions and structural abnormalities affecting chromosome segments after CRISPR

Off target effects, particularly of homologous sequences

Unclear what effects it would have for subsequent generations, thus our evolution.

No consent garnered by the embryos undergoing genome editing.

Important to differentiate between enhancement and treatment; given ART is not centrally regulated, it is likely in the US, editing will extend to enhancement

Uncertainty if the gene that is edited actually has pleiotropic effects and unable to carry out alternative, intended functions

Alternative options exist including the utilization of donor egg or sperm

NEWSLETTER

The little lit review



Connecting the dots: Carrier screening and the Genetic Information Nondiscrimination Act in the United States

Stephanie M Rice, Rodney A McLaren Jr, Hiba J Mustafa, Lorraine Dugoff, Huda B Al-Kouatly

GINA is meant to prohibit genetic discrimination by insurances and employers, yet the protections are not sweeping. It does not include protections for life insurance, long-term , disability insurance, or from select employers such as military and companies with less than 15 employees. It does not protect those with pre-existing conditions or manifestations of the genetic condition (i.e someone with cystic fibrosis). If someone, identified to have an increased cancer risk given a specific genetic variant, requests life insurance, then insurer can use the genetic information in considering the patient's case. They may decline the patient's request or cost may be changed due to the information. Below is a table curated by the authors to list out the major points to include in a carrier screening consent regarding GINA specifically:

TABLE 2 Key components of the Genetic Information Nondiscrimination Act (GINA) to discuss during pretest counseling for multigene carrier screening.^a

1. Results of carrier screening are often confidential and protected under the Genetic Information Nondiscrimination Act of 2008 (GINA) from employers and health insurance companies.
2. Protections under GINA do not apply to life insurance, long-term care insurance or disability insurance companies.
3. A subset of patients screened may be identified as manifesting carriers^b or diagnosed with a disorder that can affect future coverage of these insurances.
4. Patients have the option to defer testing to make financial decisions prior to testing, if desired.

How often are people receiving positive carrier screening results?

One study in which individual had a 274 gene panel found that 64% had a positive result.

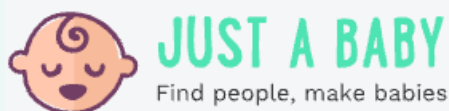
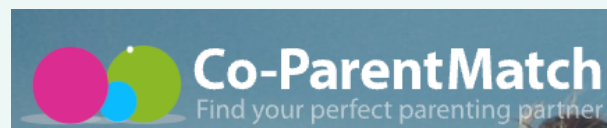
How often does the result impact the person's health?

A few studies (1, 2, 3) found that as high as 1%4 of individuals may have heath implications given the results

NEWSLETTER

Community Content:

There are multiple organizations that offer assistance in finding egg and/or sperm donor.



Modern Reproduction Content:

Modern Reproduction is now on Youtube! eek!

