What's missing from current methods for genetic screening of sperm donors?

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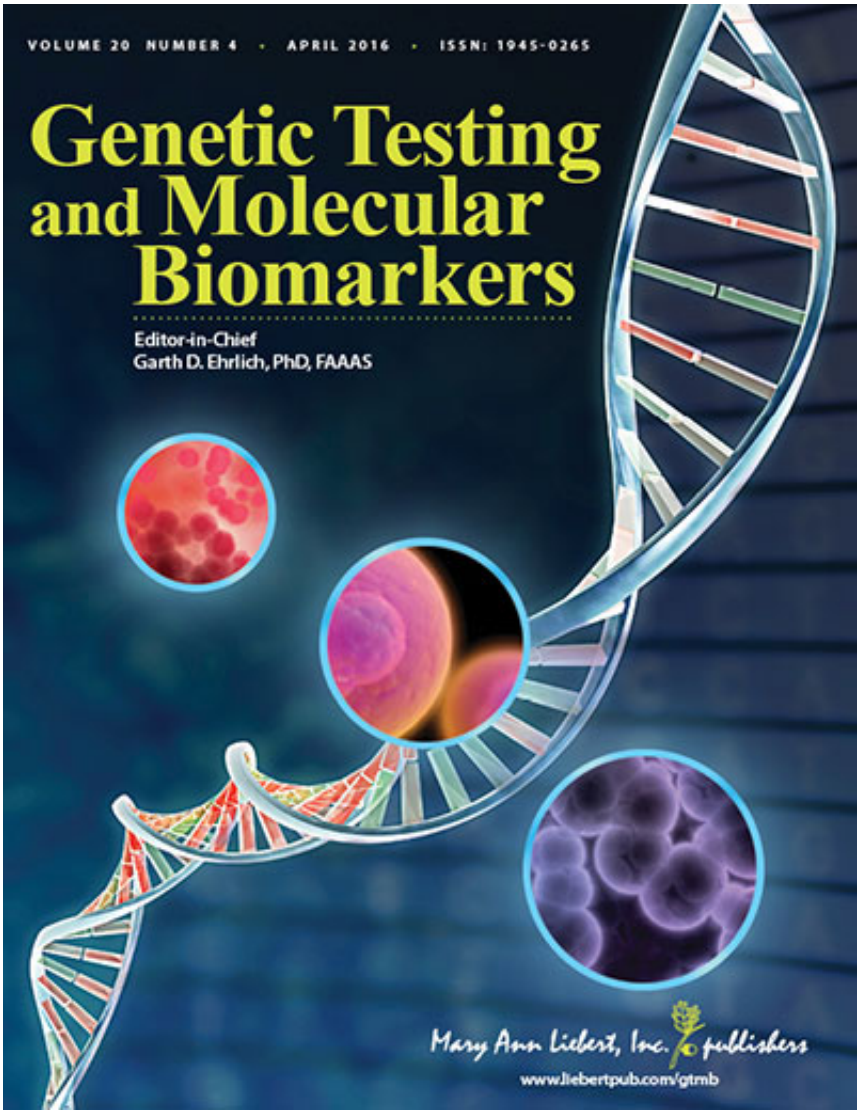

Credit: Mary Ann Liebert, Inc., publishers

U.S. sperm banks perform genetic testing to screen for and disqualify carriers of a limited number of recessive disease mutations, but more comprehensive and affordable DNA-based screening methods are now available that can detect many more disease-causing genetic variations. To protect future children from highly heritable diseases, sperm banks need to modernize their testing methods, according to an article published in *Genetic Testing and Molecular Biomarkers*, a peer-reviewed journal from Mary Ann Liebert, Inc., publishers. The article is available for download on the *Genetic Testing and Molecular Biomarkers* website.

In the article "Carrier Screening is a Deficient Strategy for Determining Sperm Donor Eligibility and Reducing Risk of Disease in Recipient Children," Ari Silver, Jessica Larson, Maxwell Silver, Regine Lim, Carlos Borroto, Brett Spurrier, Anne Morriss, and Lee Silver, Gene Peeks, Inc. (Cambridge, MA and New York, NY) and Princeton University (Princeton, NJ), compared the results of DNA-based screening of sperm donors using three commercial carrier-testing panels versus next generation DNA sequencing (NGS) technology. Whereas each carrier panel can identify serious disease mutations in specific, targeted genes that are included on the individual panel, NGS can detect variations throughout the donor's DNA. NGS analysis has advanced rapidly in recent years, becoming increasingly accessible and affordable for commercial uses.

"What this study shows is that all persons contain lethal recessive mutations," says *Genetic Testing and Molecular Biomarkers* Editor-in-Chief Garth D. Ehrlich, PhD, FAAAS, Center for Genomic Sciences and Center for Advanced Microbial Processing, Institute for Molecular Medicine and Infectious Disease, Drexel College of Medicine (Philadelphia, PA). "Thus, Silver et al have demonstrated that what is really needed in the realm of mate choice analyses are whole genome studies of both members of a potential mate-pair to ensure that they don't both carry recessive mutations in the same gene(s). Current sperm

bank testing, based on the use of target gene panels, will give prospective mothers a false sense of security that their unborn children will be free of the risk of recessive diseases."

More information: Ari J. Silver et al, Carrier Screening is a Deficient Strategy for Determining Sperm Donor Eligibility and Reducing Risk of Disease in Recipient Children, *Genetic Testing and Molecular Biomarkers* (2016). DOI: 10.1089/gtmb.2016.0014

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