Detection of chromosome abnormalities: Beyond conventional Karyotyping

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Editorial Note

Pregnancy loss is a gravely unhappy and emotionally stressful event for any couple. Besides having to address what may be a couple's psychosocial embarrassment due to their loss, one of the challenges for most researchers is how to identify these pregnancy loss cases with genetic defects that are destined to miscarry from other treatable cases. It remains indisputable that chromosomal karyotyping is the gold standard for prenatal diagnosis, including pregnancy loss. However, researchers can obtain virtually the same or better diagnostic information for detecting gains and losses of genetic material across the genome using microarray analysis, also known as molecular karyotyping and/or chromosomal array analysis, including array-based comparative genomic hybridization and single nucleotide polymorphism (SNP) array.

All of these different arrays create an entire genome scanning panel. Furthermore, it is reported that the full cohort of identifiable anomalies, regardless of known clinical significance, in a large-scale cohort of post miscarriage products of-conception samples analysed using a high-resolution SNP based microarray platform. They concluded that using SNPs extends the scope of detectable genomic abnormalities and facilitates reporting true foetal results, supporting the use of SNP chromosomal microarray analysis for cytogenomic evaluation of miscarriage specimens when clinically indicated.

Although few researchers confirmed the validity of SNP microarray analysis, the obvious question is: when should these tests be ordered? Currently, cytogenetic evaluation of products of conception is not routinely recommended. It may be that the advent of this new technology could compel us to rethink that policy and start testing this tissue more liberally. Approximately 25% of all recognized pregnancies ended in spontaneous miscarriage, with the majority caused by sporadic aneuploidy. Most authorities do not believe that karyotyping products of conception after one miscarriage will be beneficial for these parents, because the result would not change treatment for the vast majority of couples and these evaluations are too expensive.

Although cytogenetic investigation is suggested for those parents with recurrent pregnancy loss, this recommendation applies to obtaining parental karyotypes and not the karyotype of products of conception. Additionally, confirming a genetic abnormality in products of conception, resulting in a lack of further parental evaluation, could lead to failure to identify another remediable cause for miscarriage. Finally, testing all products of conception might be considered for purposes of reassuring the parents. However, the basic cost of the test, coupled with the fact that 50% of the results would be euploid and could not provide an explanation for the pregnancy loss, argue against such a practice.

Other arguments associated with article were that 10.3% of miscarriages occurred in the second trimester, which requires further discussion. Testing foetal tissue after the second or third trimester pregnancy loss might obviate the need to do additional studies, such as imaging for uterine anomalies or testing for antiphospholipid antibody syndrome. Few scientists suggest that while the present genomic diagnostic technology with access to prenatal testing following an informed consent process recommendation would probably become a future standard of care for prenatal screening/diagnosis/therapy, when specific foetal abnormalities/syndrome are identified.

In summary, SNP-based microarray analysis of products of conception is likely to give accurate results for the majority of parents; however, whether and when it is appropriate to perform this analysis remains uncertain.

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