PREGNANCY LOSS MICROARRAY

Information for clinicians

Background
Over half of early pregnancy losses are caused by a chromosomal abnormality and a genetic cause for a loss may be identified at any gestational age [1]. Microarray detects most clinically significant cytogenomic abnormalities, including gross chromosomal aneuploidies and triploidy, as well as submicroscopic (microdeletion/microduplication) copy number changes.

Advantages of microarray over conventional cytogenetics for evaluation of pregnancy loss
- Viable cells for cell culture are not required; therefore, the test failure rate is significantly reduced [2,3].
- A higher diagnostic yield is achieved as small clinically significant abnormalities can be detected that would be missed by karyotype [2,3].
- The additional information provided may enable more accurate calculation of recurrence risks for future pregnancies and inform future reproductive decisions.
- In the presence of maternal cell contamination, microarray can reduce the possibility of an inaccurate result.

Limitations and complexities of microarray testing
- Microarray cannot provide structural information. For this reason, follow-up parental karyotype analysis may be recommended for assessment of recurrence risk in future pregnancies.
- Genetic variants of uncertain clinical significance are sometimes identified. Parental testing may be useful in determining the significance of these variants.
- Microarray may reveal secondary findings with implications for the extended family. These may include variants associated with disease in later life or with susceptibility to neurodevelopmental disorders, and parental relatedness. Genetic counselling will be helpful to resolve issues with such findings.
- Although microarray offers a higher sensitivity than conventional cytogenetics, it cannot identify all genetic causes of pregnancy loss (e.g. sequence variants and epimutations).

Requirements
- **Sample requirements**: Fetal tissue or products of conception, transported in a universal sterile container with PBS or normal saline
- **Accompanying paperwork**: A request form indicating the test to be performed (available on the PathWest website).
  Please note, a “Fetal chromosomal microarray - Information and consent” form is available on the PathWest website. The laboratory is not required to sight a copy of the consent but an indication that consent has been obtained should be documented.

Turnaround time and billing
- Results are typically available within 6 weeks.
- The genetic analysis of pregnancy loss samples by microarray is covered by Medicare (Item 73287).

References