

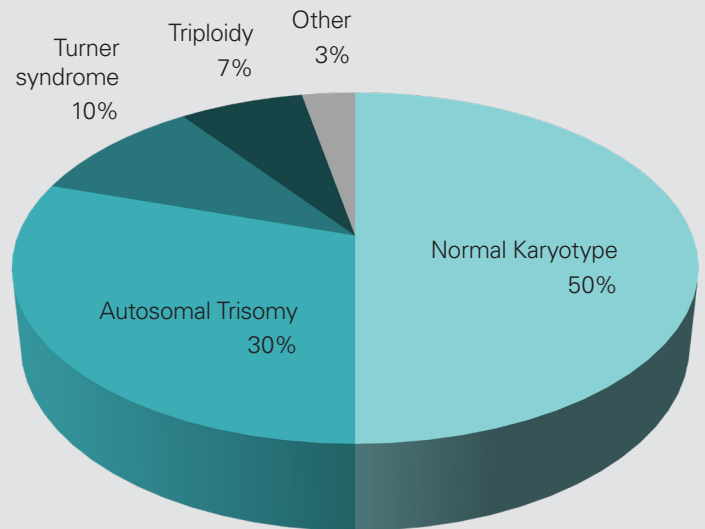
# Pregnancy Loss Testing Services

## Fetal Tissue | Products of Conception

v21.1

- Feelings of guilt and blame are common in women experiencing a loss; these symptoms are exacerbated for some women by misconceptions about the cause of their pregnancy loss.
- At least 10-15% of all recognized pregnancies are miscarried. Chromosome abnormalities cause more than 50% of first trimester losses and 15% of second trimester losses.<sup>1</sup>
- In most instances, identification of a chromosomal cause for pregnancy loss allows calculation of accurate recurrence risks; families and providers use risks to determine appropriate fertility and pregnancy management.
- Women with normal genetic test results may require further clinical evaluation, which may include additional laboratory studies, prior to their next pregnancy.

### Genetic Breakdown of Pregnancy Loss



Testing Options		
Patient History	Our Recommended Testing	Rationale
Woman with a history of recurrent pregnancy loss currently not pregnant	Chromosome Analysis for woman and her partner	Chromosome Analysis identifies a chromosomal rearrangement, such as a balanced translocation, in 5-8% of individuals with recurrent loss. <sup>2,3</sup>
Woman with a history of recurrent pregnancy loss currently experiencing a miscarriage	Pregnancy Loss Microarray performed on fetal tissue	Pregnancy Loss Microarray offers an increased detection rate and greater likelihood of obtaining results as compared to traditional chromosome analysis. <sup>4</sup>
Woman currently experiencing a stillbirth	Pregnancy Loss Microarray performed on fetal tissue	Pregnancy Loss Microarray offers an increased detection rate and greater likelihood of obtaining results as compared to traditional chromosome analysis. <sup>4</sup>
Woman without a history of recurrent pregnancy loss currently experiencing a miscarriage	Pregnancy Loss Microarray performed on fetal tissue	Testing may be appropriate for women experiencing their first or second loss, particularly when the family or pregnancy history is suspicious for a chromosomal cause of pregnancy loss.

1. Gardner, R.J.M. & Sutherland, G.R. (2004). Chromosome abnormalities and genetic counseling. 3rd ed. New York, NY: Oxford University Press.  
 2. Braekeleer, M.D. & Dao, T.N. (1990). Cytogenetic studies in couples experiencing repeated pregnancy losses. *Hum Reprod*, 5(5), 519-528.  
 3. Harger, J.H., Archer, D.F., Marchese, S.G., Muracca-Clemons, M., & Garver, K.L. (1983). Etiology of recurrent pregnancy losses and outcome of subsequent pregnancies *Obstet Gynecol*, 62(5),574-581.  
 4. Schaeffer, A.J., Chung, J., Heretis, K., Wong, A., Ledbetter, D.H., & Martin, C.L. (2004). Comparative genomic hybridization-array analysis enhances the detection of aneuploidies and submicroscopic imbalances in spontaneous miscarriages. *Am J Hum Genet*, 74(6), 1168-1174.

# Prenatal Testing Services

## Amniotic Fluid | Chorionic Villi Sampling

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### CHROMOSOME ANALYSIS

- Detects extra or missing whole chromosomes
  - Down syndrome
  - Trisomy 13
  - Trisomy 18
  - Triploidy
  - Turner syndrome
  - Other sex chromosome aneuploidies
- Detects structural aberrations
  - Large-scale deletions and duplications
  - Balanced and unbalanced rearrangements

### FLUORESCENCE *IN SITU* HYBRIDIZATION (FISH)

- Useful for targeted assessment of various chromosomal abnormalities
  - Common aneuploidies (13, 18, 21, X, and Y)
  - Familial chromosomal abnormalities
  - Microdeletion or microduplication syndromes
- Results for STAT indications communicated to referring health care provider within 24 hours of specimen receipt

### MICROARRAY ANALYSIS

- Detects submicroscopic copy number changes (microdeletions and microduplications) in targeted regions throughout the genome

### INDICATIONS FOR TESTING

#### CHROMOSOME ANALYSIS with the option of FISH

- ◆ Advanced maternal age
- ◆ Abnormal ultrasound findings
- ◆ Abnormal NIPT, 1st trimester, 2nd trimester, or combined maternal serum screening result
- ◆ Family history of genetic or chromosomal anomalies

#### PRENATAL MICROARRAY

- ◆ Abnormal ultrasound findings
- ◆ Abnormal screening test (NIPT) for microdeletions
- ◆ Family history of genetic or chromosomal anomalies
- ◆ History of pregnancy loss