REPRODUCTIVE MEDICINE and PRENATAL TESTING

REPRODUCTIVE MEDICINE

Chromosome Analysis: identifies numerical or structural abnormalities

Couples with one or more miscarriage
Male infertility: azoospermia or oligospermia
Female infertility
Primary amenorrhea
Unexplained secondary amenorrhea
Family history of chromosome abnormality
(including parents with balanced translocation)
Stillborns
Products of conception - miscarriage (especially multiple)

DNA Testing

Congenital Bilateral Absence of the Vas Deferens (CBAVD)

- Patients with congenital absence of the vas deferens (unilateral or bilateral) who are either negative or heterozygous for a CF mutation
- Patients with CAVD or mild CF symptoms in whom one CF mutation has been previously identified

Fragile X Syndrome

- Determination of fragile X gene status in persons with a family history of fragile X syndrome or undiagnosed mental retardation
- Determination of fragile X gene status in females with premature ovarian failure

Ashkenazi Jewish disease panel. Tests can be ordered as a group or individually and include: Bloom syndrome, Canavan disease, Fanconi anemia (group C), familial dysautonomia, glycogen storage disease (1a), mucolipidosis type IV, Niemann-Pick (type A), and Tay-Sachs disease

- Determination of carrier status (for individuals of Ashkenazi Jewish ancestry only)

APPENDIX – Cytogenetics

PRENATAL TESTING

Amniocentesis or Chorionic Villus Sampling

Chromosome Analysis*:* identifies numerical and structural chromosome abnormalities in amniotic fluid or chorionic villus samples.

- Indications for testing:
  - Maternal age
  - Maternal serum screening risk is increased
  - Positive AFP profile
  - Fetal abnormality on ultrasound
  - Family history of chromosome abnormality
- Parental studies may be required as a follow-up for abnormal cytogenetic studies
- Final reports include a description of clinical manifestations, recurrence risks, prognostic indicators and pertinent literature references.

Fluorescent in situ hybridization (FISH)

Aneuploidy screening of amniotic fluid: identifies sex chromosome gain or loss, and trisomies of chromosome 13, 18 and 21. Analysis is performed on interphase cells and results are provided within 24-48 hours of sample receipt. Note: this is not a stand-alone test and should be performed as an adjunct to karyotype analysis.

DiGeorge syndrome testing: performed if a heart abnormality is detected on fetal ultrasound or of there is a family history of DiGeorge or velocardiofacial sydrome (22q deletion).

DNA Testing

Cystic Fibrosis

- Prenatal diagnosis when both parents are known carriers or when one parent is a known carrier and the other parent is not available for DNA testing
- Abnormal ultrasound (echogenic bowel)

Fragile X Syndrome

- Determination of fragile X gene status in an at-risk fetus. An at-risk fetus is one with a documented fragile X-positive family member

Myotonic Dystrophy

- Determination of myotonic dystrophy gene status in an at-risk fetus. An at-risk fetus is one with a documented affected family member

Prader-Willi/Angelman Syndrome

- Determination of PWS/AS methylation status in an at-risk fetus. An at-risk fetus is one with a documented affected family member

Sickle Cell Anemia/Hemoglobin C Disease

- Determination of fetal sickle cell and hemoglobin C disease status when both parents are known carriers or when one parent is a known carrier and the other parent is not available for testing

Ashkenazi Jewish diseases - tests can be ordered for an at-risk fetus and include: Bloom syndrome, Canavan disease, Fanconi anemia (group C), familial dysautonomia, glycogen storage disease (1a), mucolipidosis type IV, Niemann-Pick (type A), and Tay-Sachs disease

- Determination of gene status in an at-risk fetus. An at-risk fetus is one with a documented affected family member