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 syndrome (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.