

## PRIMARY CARE

### PEDIATRICS - INTERNAL MEDICINE - FAMILY PRACTICE

**Chromosome analysis:** Identifies numerical and structural chromosome abnormalities.

- Indications for testing:
  - Mental retardation
  - Multiple congenital anomalies
  - Dysmorphic features
  - Ambiguous genitalia and cryptorchidism
  - Multiple pregnancy losses
  - Males: infertility, azospermia or oligospermia
  - Females: infertility, primary or unexplained secondary amenorrhea, short stature
  - Pigmentary dysplasia and mental retardation
  - Congenital contractures and mental retardation
  - Possible microdeletion syndromes
- Parental studies may be required as a follow-up for abnormal cytogenetic studies
- Description of clinical manifestations, recurrence risks, prognostic indicators and pertinent literature references are provided on abnormal reports.
- Preliminary reports on samples from newborns are provided within 24-48 hours.

### Fluorescent in situ hybridization (FISH)

Microdeletion syndrome probes:

- 1p36
  - Angelman syndrome
  - Cri-du-chat syndrome
  - DiGeorge/Velocardiofacial syndrome
  - Kallman's syndrome
  - Miller-Dieker syndrome
  - Prader-Willi syndrome
  - Smith-Magenis syndrome
  - Sotos syndrome (5q35)
  - Steroid Sulfatase (STS) probe
  - Williams syndrome
  - Wolf-Hirschhorn Syndrome
- Telomere rearrangement panel  
SRY probe

*Note: since new probes are continually being developed, call the laboratory for availability of probes not listed.*

## **DNA Testing**

### **Angelman Syndrome**

- Confirmation of diagnosis or clinical suspicion of AS
- Confirmation of diagnosis in patients with a suspected diagnosis, but with negative cytogenetic studies

### **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**

- Confirmation of diagnosis of males or females with mental retardation or developmental delay of unknown etiology
- Confirmation of diagnosis of individuals previously diagnosed by cytogenetic methods
- Carrier testing for females with premature ovarian failure

### **Hereditary Hemochromatosis**

- Confirmation of diagnosis of affected individuals
- Carrier testing for persons with family history of hereditary or idiopathic hemochromatosis
- All patients in whom a diagnosis of hemochromatosis is being pursued

### **Hereditary Pancreatitis**

- Hereditary pancreatitis with onset before age 20 years, and/or at-least 2 relatives with pancreatitis
- Idiopathic pancreatitis with or without a positive family history
- A relative known to carry a mutation in the cationic trypsinogen gene associated with hereditary pancreatitis

### **Myotonic Dystrophy**

- Confirmation of diagnosis of affected individuals
- Carrier testing for persons with a family history of myotonic dystrophy

### **Prader Willi Syndrome**

- Confirmation of diagnosis or clinical suspicion of PWS
  - Infants with hypotonia of unknown origin
  - Older, obese, moderately retarded patients
- Confirmation of diagnosis in patients with a suspected diagnosis, but with negative cytogenetic studies

### **Multiple Endocrine Neoplasia (MEN), Type 2A and Type 2B**

- Confirmation of diagnosis and determination of specific RET mutation
- Presymptomatic screening of at-risk family members with a known familial RET mutation
- Documentation of a germline mutation to confirm familial inheritance of the disorder

### **Familial Medullary Thyroid Carcinoma (FMTC)**

- Confirmation of diagnosis and determination of specific RET mutation
- Presymptomatic screening of at-risk family members with a known familial RET mutation
- Documentation of a germline mutation to confirm familial inheritance of the disorder

**MTHFR 677 C→T**

- Documented hyperhomocysteinemia
- Recurrent cerebrovascular, peripheral vascular or coronary artery disease
- Recurrent venous thrombosis
- A relative known to carry the MTHFR 677C→T mutation
- Presence of another known genetic hypercoagulability mutation in an individual with a history of venous thrombosis
- As part of a comprehensive thrombophilia evaluation

**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), mucopolidosis type IV, Niemann-Pick (type A), and Tay-Sachs disease**

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