

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.

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)