

Uniparental Disomy (UPD) Screen

DESCRIPTION:

- This is a DNA test to examine inheritance of nucleotide repeat polymorphisms on a selected chromosome for a patient and parents. Normal biparental inheritance rules out the presence of uniparental disomy in the patient.

REASONS FOR REFERRAL:

- Uniparental disomy may cause some cases of Prader-Willi, Angelman, Russell-Silver, and Beckwith-Wiedemann syndromes (see separate listings).
- Uniparental disomy has been reported for at least 15 autosomes and both sex chromosomes (Ledbetter and Engel, *Hum Mol Genet*, 1995, 4: 1757-1764). For chromosomes 2, 6, 7, 11, 14, 15, 16, 20, and X, abnormal phenotypes may result from UPD due to imprinted genes (genes with either maternal or paternal-only expression). An abnormal phenotype can also result when the child is functionally homozygous (uniparental isodisomy) for one parental chromosome which happens to carry a deleterious recessive allele. This has been reported for disorders such as Cystic Fibrosis (Spence, JE et al., *Am J Hum Genet*, 1988, 42: 217-226).
- UPD occurs in one third of cases of "trisomy rescue", often associated with a prenatal diagnosis of mosaic trisomy (Kalousek et al., *Prenat Diagn*, 1991, 11:743).

METHOD OF ANALYSIS:

- Size polymorphisms of nucleotide repeat alleles are determined by PCR amplification using specific primers and nucleotides labeled with fluorescent dye. Typically, ten or more loci are amplified in patient and parents. Alleles are determined by a high resolution laser-induced fluorescence capillary electrophoresis system with internal standard.
- Results are reported within 3 weeks or less of receipt of samples.
PRENATAL TURNAROUND TIME IS TARGETED AT 14 DAYS OR LESS.

REFERENCE RANGES:

- At least three informative loci must show biparental inheritance to rule out UPD. If maternal UPD is suggested, paternity is confirmed by loci on other chromosomes.

SAMPLE REQUIREMENTS:

- 5 to 10 milliliters of blood (minimum 1 ml) in EDTA (purple top) tubes should be sent by overnight carrier at room temperature.
- 2 confluent T-25 flasks of cultured amniocytes or CVS sent overnight at room temperature. Please contact the laboratory regarding specimens for prenatal analysis.
- Testing requires DNA from the patient and at least parent, both parents are recommended.

TEST CHARGE: \$732.00 (LIST PRICE EFFECTIVE June 1, 2009)

CPT 83890	DNA extraction X 3	\$ 111 (\$37 x 3)
CPT 83894	DNA separation	\$ 24
CPT 83898	DNA amplification X 3	\$ 270 (\$90 x 3)
CPT 83901	DNA amplification multiplex X 3	\$ 270 (\$90 x 3)
CPT 83912	DNA interpretation and report	\$ 57

Discounts from list price are available for institutional billing under contractual arrangement with the laboratory. Contact Ellen Livers at 800-447-6614 ext 7523.