
Lab Dept: **Anatomic Pathology**

Test Name: **UNIPARENTAL DISOMY CHROMOSOME 6**

General Information

Lab Order Codes: UPD6

Synonyms: UPD Testing for chromosome 6; Transient Neonatal Diabetes

CPT Codes: 81402 – Uniparental disomy (UPD) (eg, Russell-Silver syndrome, Prader-Willi/Angelman syndrome), short tandem repeat (STR) analysis

Test Includes: Testing of child and both parents.

Logistics

Test Indications: Uniparental disomy (UPD) describes a condition in which both homologs of a chromosome pair are derived from the same parent. UPD can be associated with phenotypic abnormalities if:

- The chromosome or chromosome segment involved carries genes that are imprinted. Genomic imprinting refers to the differential expression of alleles as determined by the parental origin of the allele.
- Homozygosity due to uniparental isodisomy results in the expression of an autosomal recessive condition from a single carrier parent.

Lab Testing Sections: Anatomic Pathology - Sendouts

Referred to: University of Chicago Genetic Services Laboratory

Phone Numbers: MIN Lab: 612-813-6280

STP Lab: 651-220-6550

Test Availability: Monday – Thursday, 24 hours

Turnaround Time: 2 - 4 weeks

Special Instructions: EDTA blood from both parents and the child are required.

Specimen

Specimen Type: Whole blood

Container:	Lavender top (EDTA) tube x3
Draw Volume:	10 mL (Minimum: 3 mL) blood from child and both parents
Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
Special Processing:	Lab Staff: Do Not centrifuge. Send specimens in original collection containers via overnight shipping. Store and ship at room temperature. Forward promptly. Note: Specimens can only be shipped Monday through Thursday. Specimens will not be processed at the University of Chicago on weekends or holidays. Hold specimens collected on weekends and holidays at refrigerated temperatures and ship at ambient temperatures on the next business day, Monday – Thursday.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens

Interpretive

Reference Range:	Narrative report
Methodology:	PCR amplification, gel electrophoresis
References:	University of Chicago Genetic Services Laboratory Web Page January 2013 Phone: 888-824-3637
Updates:	1/15/2013: CPT update