

The University of Chicago Genetic Services Laboratories



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UPD TESTING

Thank you for your inquiry regarding testing for uniparental disomy (UPD). We offer analysis for chromosomes 6, 7, 14, and 15. Sample submission instructions and paperwork are included with this fax.

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.

Prenatal UPD studies are appropriate following the detection of a mosaic trisomy or a marker chromosome in a prenatal sample. UPD analysis is also considered when a prenatal test is performed to rule out the possibility that the fetus has inherited an unbalanced translocation from a parent carrying a balanced rearrangement and for pregnancies affected with a de novo translocation.

UPD has been demonstrated in patients with Prader-Willi syndrome (UPD15mat), Angelman syndrome (UPD15pat), and transient neonatal diabetes mellitus (UPD6pat). UPD7mat has been associated with pre- and postnatal growth retardation and Russell-Silver syndrome. UPD14mat has been associated with growth retardation and precocious puberty. UPD14pat has been associated with mental retardation and skeletal abnormalities.

Sample specifications:	3-10 cc of blood from the patient and BOTH parents in purple top (EDTA) tubes 2 T-25 flask of cultured CVS or amniotic fluid cells (80% ⁺ confluent) for PRENATAL testing
Cost:	\$540 (total for a patient's and both parents' blood samples) \$740 (total for a prenatal sample and both parents' blood samples)
CPT codes:	83891, 83898 x 4, 83894, 83912 (+ 99051 for prenatal cases)
Turn-around time:	15-20 days