



UPD testing

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.

Test methods:

We offer UPD testing of chromosomes 6, 7, 14, and 15 by microsatellite analysis, which compares microsatellite markers from both parents and the child or fetus. In order for UPD to be determined, a significant number of informative microsatellite markers must be present. Although testing is possible if only one parent is available, the chance of obtaining a sufficient number of informative markers is decreased.

UPD testing

Sample specifications:	3 to 10 cc of blood from patient and both parents in a purple top (EDTA) tube
Cost:	\$540 (total for a patient's and both parents' blood samples)
CPT codes:	81402
Turn-around time:	2 - 4 weeks

UPD prenatal testing

Sample specifications:	2 T-25 flask of cultured CVS or amniotic fluid cells (80%+ confluent)
Cost:	\$740
CPT codes:	81402
Turn-around time:	2 - 4 weeks

Results

Results, along with an interpretive report, will be faxed to the referring physician. Additional reports will be provided as requested. All abnormal results will be reported by telephone.

For more information about our testing options, please visit our website at dnatesting.uchicago.edu or contact us at 773-834-0555.

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