

UAB MEDICAL GENOMICS LABORATORY

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Please note that prices listed correspond to institutional rates; please contact the lab for insurance rates.

Test	Description	Turn Around Time / Price	CPT codes	Specimen Requirements
Neurofibromatosis Type 1				
Test 1	Comprehensive test , using direct sequencing of the entire coding region at cDNA/DNA level + dosage analysis + microsatellite marker analysis + FISH analysis. Identifies >95% of mutations in non-founder patients fulfilling NIH diagnostic criteria	5-6 weeks; 2-3 weeks for RUSH testing \$1200,-USD for comprehensive analysis; Add \$600,-USD for RUSH We charge only \$500, -USD for NF1 comprehensive analysis if NF1 total gene deletion is identified	Comprehensive: 83891(x3), 83903(x1), 83913(x1), 83909(x9), 83894(x3), 83896(x2), 83898(x16), 83900(x1), 83902(1), 83904(x18), 83912(x1), 88230(x1) NF1 Total Gene Deletion identified during Comprehensive: 83891 (x3), 83913 (x1), 88230 (x1), 83909 (x3), 83896 (x3), 83898 (x3), 83912 (x1)	(1) 10 ml of whole blood in EDTA (purple topped) tubes. (Minimum of 3 ml for pediatric patients) Samples must be received within 60 hours
MLPA	Copy Number Analysis by MLPA	2-3 weeks \$500,-USD	83891(x1), 83909(x6), 83896(x6), 83898(x6), 83912(x1)	(1) 3 ml whole blood in EDTA (purple topped) tubes
Test 2	Targeted testing : detection of a specific, previously known <i>NF1</i> mutation in at risk family members	2-3 weeks \$250,-USD	83891(x1), 83894(x4), 83898(x4), 83904(x3), 83912(x1)	(1) 1 ml whole blood in EDTA (purple topped) tubes (2) Buccal swab sample
Test 3	Prenatal testing of specific, previously known <i>NF1</i> mutation	6 working days after sample is received \$750,-USD	83891(x3), 83894(x20), 83896(x6), 83898(x20), 83904(x8), 83912(1)	(1) Direct CVS, (minimum 10 mg pure villi) (2) Direct amniotic fluid (minimum 10 ml fluid) (3) 2 T25 flasks of cultured CVS (>70% confluent) (4) 2 T25 flasks of cultured amniocytes. (>70% confluent)
Test 4	Comprehensive testing on neural crest derived cells , cultured from affected body regions (CAL-spots and/or neurofibromas)	Dependent on size and quality of the tissue specimen received. 3-6 months \$2600,-USD	83891(x6), 83903(x2), 83913(x2), 83909(x18), 83894(x6), 83896(x4), 83898(x32), 83902(x2), 83904(x36), 83912(x2), 88233(x2)	(1) 2-3 5mm punch biopsies from CAL-spots. Special transport media required for shipping: contact us at 205-934-5562 (2) minimum 3 separate fresh neurofibromas. Special transport

				media required for shipping: contact us at 205-934-5562
SPRED1- Neurofibromatosis Type 1 Like Syndrome				
Test 1	<p>Comprehensive test as a <u>reflex</u> test after NF1 testing (cDNA sequencing), using direct sequencing of the entire coding region at the RNA level and copy number analysis by MLPA and quantitative PCR</p> <p>OR</p> <p>Comprehensive test, (no NF1 testing previously done) using direct sequencing of the entire coding region at the DNA level and copy number analysis by MLPA and quantitative PCR</p>	<p>SPRED1 after NF1 testing \$500,-USD</p> <p>2 weeks</p> <p>No previous NF1 testing \$600,-USD</p> <p>3-4 weeks</p>	<p>SPRED1 reflex from NF1: 83891(x2), 83913(x1), 83894(x1), 83896(x3), 83898(x4), 83904(x4), 83909(x3), 83912(x1), 88230(x1)</p> <p>No previous NF1 testing (DNA-based): 83891(x1), 83896(x3), 83898(x11), 83904(x16), 83909(x3), 83912(x1)</p>	<p>(1) 10 ml of whole blood in EDTA (purple topped) tubes. (Minimum of 3 ml for pediatric patients)</p> <p>Samples must be received within 60 hours</p>
MLPA/QPCR	Copy Number Analysis by MLPA and Quantitative PCR (Note: Copy Number analysis is included as part of the Comprehensive Test1)	<p>2 weeks</p> <p>\$300,-USD</p>	83891(x1), 83909(x3), 83896(x3), 83898(x3), 83912(x1)	(1) 3 ml whole blood in EDTA (purple topped) tubes
Test 2	Targeted testing: detection of a specific, previously known SPRED1 mutation within family members	<p>2 weeks</p> <p>\$250,-USD</p>	83891(x1), 83894(x4), 83898(x4), 83904(x3), 83912(x1)	<p>(1) 1 ml whole blood in EDTA (purple topped) tubes</p> <p>(2) Buccal swab sample</p>
Neurofibromatosis Type 2				
Test 1	Comprehensive test , using direct sequencing of the entire coding region at cDNA/DNA level including dosage analysis (MLPA). Identifies 92% of mutations in non-founder NF2 patients	<p>4-5 weeks; 2-3 weeks for RUSH testing</p> <p>\$1250,-USD; Add \$600,-USD for RUSH</p>	83891(x2), 83913(x1), 83909(x9), 83894(x11), 83896(x7), 83898(x15), 83902(x1), 83904(x7), 83912(x1), 88230(x1)	<p>(1) 10 ml of whole blood in EDTA (purple topped) tubes. (Minimum of 3 ml for pediatric patients)</p> <p>Samples must be received within 60 hours</p>
MLPA	Copy Number Analysis by MLPA (Note: Copy Number analysis is included as part of the Comprehensive Test1)	<p>2-3 weeks</p> <p>\$300,-USD</p>	83891(x1), 83909(x3), 83896(x3), 83898(x3), 83912(x1)	(1) 3 ml whole blood in EDTA (purple topped) tubes
Test 2	Targeted testing: detection of a specific, previously known NF2 mutation in at risk family members	<p>2-3 weeks</p> <p>\$250,-USD</p>	83891(x1), 83894(x4), 83898(x4), 83904(x3), 83912(x1)	<p>(1) 1 ml whole blood in EDTA (purple topped) tubes</p> <p>(2) Buccal swab sample</p>

Test 3	Prenatal testing of specific, previously known <i>NF2</i> mutation	6 working days after sample is received \$750,-USD	83891(x3), 83909(x16), 83894(x4), 83896(x6), 83898(x20), 83904(x8), 83912(x1)	(1) Direct CVS, (minimum 10 mg pure villi) (2) Direct amniotic fluid (minimum 10 ml fluid) (3) 2 T25 flasks of cultured CVS (>70% confluent) (4) 2 T25 flasks of cultured amniocytes. (>70% confluent)
Test 4	Comprehensive testing on tumor blocks/frozen tissue using direct sequencing of all <i>NF2</i> exons at the gDNA level and including dosage analysis (MLPA) Or Using direct sequencing of the entire coding region at cDNA/DNA level including dosage analysis (MLPA) if a fresh biopsy was obtained	4-5 weeks \$1400,-USD	83891(x2), 83907(x2), 83909(x9), 83894(x14), 83896(x1), 83898(x16), 83904(x14), 83912(x1)	(1) tumor block (2) fresh tumor biopsy, immersed in special transport media required for shipping: contact us at 205-934-5562
Schwannomatosis - INI1/SMARCB1 Testing				
Test 1	Comprehensive test , using direct sequencing of the entire coding region at cDNA/DNA level including dosage analysis (MLPA).	4-5 weeks \$800,-USD	RNA-based: 83891(x2), 83913(x1), 83909(x1), 83894(x2), 83896(x3), 83898(x3), 83902(x1), 83904(x8), 83912(x1), 88230(x1) DNA-based: 83891(x1), 83909(x1), 83894(x10), 83896(x3), 83898(x10), 83904(x18), 83912(x1)	(1) 10 ml of whole blood in EDTA (purple topped) tubes. (Minimum of 3 ml for pediatric patients) Samples must be received within 60 hours
MLPA	Copy Number Analysis by MLPA (Note: Copy Number analysis is included as part of the Comprehensive Test1)	2-3 weeks \$300,-USD	83891(x1), 83909(x3), 83896(x3), 83898(x3), 83912(x1)	(1) 3 ml whole blood in EDTA (purple topped) tubes
Test 2	Targeted testing : detection of a specific, previously known <i>INI1</i> mutation in at risk family members	2-3 weeks \$250,-USD	83891(x1), 83894(x4), 83898(x4), 83904(x3), 83912(x1)	(1) 1 ml whole blood in EDTA (purple topped) tubes (2) Buccal swab sample
Test 3	Prenatal testing of specific, previously known <i>INI1</i> mutation	6 working days after sample is received \$750,-USD	83891(x3), 83894(x20), 83896(x6), 83898(x20), 83904(x8), 83912(x1)	(1) Direct CVS, (minimum 10 mg pure villi) (2) Direct amniotic fluid (minimum 10 ml fluid) (3) 2 T25 flasks of cultured CVS (>70% confluent) (4) 2 T25 flasks of cultured amniocytes. (>70% confluent)
Test 4	Comprehensive testing on minimum 2 different tumor blocks/frozen tissue using direct sequencing of all <i>INI1</i> and <i>NF2</i> exons at the gDNA level and including dosage analysis (MLPA) Or Using direct sequencing of the entire coding region at cDNA/DNA level including	4-5 weeks \$2200,-USD	83891 (x1), 83907 (x1), 83909 (x10), 83894 (x24), 83896 (x4), 83898 (x26), 83904 (x32), 83912 (x1)	(1) tumor block (2) fresh tumor biopsy, immersed in special transport media required for shipping: contact us at 205-934-

	dosage analysis (MLPA) if a fresh tumor sample was obtained			5562
PTEN Hamartoma Tumor Syndrome / Macrocephaly-Autism Syndrome - PTEN				
Test 1	<p>Comprehensive testing using direct sequencing of the entire coding region at the cDNA or DNA level, sequencing of the core promoter region and dosage analysis (MLPA).</p> <p>Tier 1: Sequencing of coding region and core promoter region at the RNA or DNA level</p> <p>Tier 2: Copy number analysis by MLPA</p>	<p>5 weeks (2-3 weeks for copy number analysis by MLPA only)</p> <p>\$1100,-USD (Tier1)</p> <p>\$300,-USD (Tier2)</p>	<p>Tier 1-RNA-based 83891 (x2), 83894 (x2) 83898 (x2), 83904 (x8), 83912 (x1), 83913(x1), 88230(x1)</p> <p>Tier 1-DNA-based 83891 (x1), 83894 (x10), 83898 (x10), 83904 (x20), 83912 (x1)</p> <p>Tier 2 83891 (x1), 83896 (x3), 83898 (x3), 83909 (x3), 83912 (x1)</p>	<p>(1) 10 ml of whole blood in EDTA (purple topped) tubes. (Minimum of 3 ml for pediatric patients. For add-on Fragile testing for pediatric patients, we require a minimum of 5 ml)</p> <p>Samples must be received within 60 hours</p>
Promoter	<p>Sequencing of the core promoter region Note: Sequencing of the core promoter region is included as part of the comprehensive test 1</p>	<p>2-3 weeks</p> <p>\$350,-USD</p>	83891(x1), 83894(x2), 83898(x2), 83904(x4), 83912(x1)	(1) 3 ml whole blood in EDTA (purple topped) tubes
Test 2	<p>Targeted testing: detection of a specific, previously known <i>PTEN</i> mutation in at risk family members</p>	<p>2-3 weeks</p> <p>\$250,-USD</p>	83891 (x1), 83894 (x4), 83898 (x4), 83904 (x3), 83912 (x1)	(1) 1 ml whole blood in EDTA (purple topped) tubes
Test 3	<p>Prenatal testing of specific, previously known <i>PTEN</i> mutation</p>	<p>6 working days after sample is received</p> <p>\$750,-USD</p>	83891(x3), 83894(x20), 83896(x6), 83898(x20), 83904(x8), 83912(x1)	<p>(1) Direct CVS, (minimum 10 mg pure villi) (2) Direct amniotic fluid (minimum 10 ml fluid) (3) 2 T25 flasks of cultured CVS (>70% confluent) (4) 2 T25 flasks of cultured amniocytes. (>70% confluent)</p>
Autosomal Recessive Polycystic Kidney Disease (ARPKD)				
Test 1	<p>Sequence analysis of entire <i>PKHD1</i> longest open reading frame and copy number analysis by MLPA, detecting >82% of mutations</p> <p>Tier 1: Sequencing of 23 exons that contain as many as 80% of identifiable pathogenic mutations</p> <p>Tier 2: Sequencing of the remaining exons of the entire longest open reading frame</p> <p>Tier 3: Copy number analysis by MLPA/qPCR</p>	<p>Maximum of 4 weeks</p> <p>Tier 1-3: \$3350,-USD</p> <p>Tier 1: \$1200,-USD</p> <p>Tier 1+2: \$2800,-USD</p> <p>Tier 3: \$600,-USD</p> <p>RUSH: \$3500,-USD (Tier 1 +Tier 2 only) 8 working days</p>	<p>Tier 1-3: 83891(x1) 83896 (x6) 83898(x86) 83904(x160) 83909(x6) 83912(x1)</p> <p>Tier1 : 83891(x1), 83898(x32), 83904(x64), 83912(x1)</p> <p>Tier1+2 : 83891(x1), 83898(x80), 83904(x160), 83912(x1)</p> <p>Tier 3 : 83891(x1) 83909(x6) 83896(x6) 83898(x6) 83912(x1)</p>	<p>(1) 5 ml whole blood in EDTA (purple tubes)</p> <p>(2) biopsy from skin/liver/spleen... (fresh or frozen)</p> <p>(3) 2 T25 flasks of cultured cells</p>

Test 2	Targeted testing of specific, well-defined <i>PKHD1</i> mutations previously identified in the family	2-3 weeks Free of charge for both parents of the proband if samples submitted same week as proband Other relatives: \$190,-USD (1 exon) \$340,-USD (2 exons)	1 exon: 83891(x1), 83898(x4), 83904(x3), 83912(x1) 2 exons: 83891(x1), 83898(x8), 83904(x6), 83912(x1)	(1) 5 ml whole blood in EDTA (purple topped) tubes (2) Buccal swab sample
Test 3	Prenatal testing of specific, well-defined <i>PKHD1</i> mutations, previously identified in the proband	6 working days after sample is received \$1000,-USD	83891(x3), 83909(x6), 83898(x20), 83904(x16), 83912(x1)	(1) Direct CVS, (minimum 10 mg pure villi) (2) 2 T25 flasks of cultured CVS. (>70% confluent) (3) 2 T25 flasks of cultured amniocytes. (>70% confluent) Please also send 1-5 ml of blood or buccal swab sample from mother for maternal contamination studies.
Tests 4 & 5	Haplotype analysis by PCR of 7 markers within and flanking the disease interval of chromosome 6p21-cen	Test 4: \$400,-USD per individual tested 4 weeks for informativity, (Test 4) Test 5: \$500,-USD per individual tested 6 working days for prenatal samples, (Test 5)	Test 4: 83891(x1), 83909(x3), 83896(x7), 83898(x7), 83912(x1) Test 5: 83891(x2), 83909(x6), 83896(x14), 83898(x14), 83912(x1)	Please send samples from the parents and their children for informativity results (1) 10ml of whole blood in EDTA (purple topped) tubes. (2 ml minimum for pediatric patients) (2) Paraffin-embedded tissue blocks or whole tissue from affected individual (3) For prenatal samples, please send 2 T25 flasks of cultured CVS or 2 T25 flasks of cultured amniocytes.
Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)				
Test 1	Targeted analysis for c.985A>G (Tier1), complete sequence analysis of <i>ACADM</i> (Tier2)	2 weeks \$190,-USD (Tier 1) \$600,-USD (Tier 2)	Tier1: 83891(x1), 83894(x3), 83898(x3), 83904(x2), 83912(x1) Tier2: 83891(x1), 83894(x10), 83898(x10), 83904(x20), 83912(x1)	(1) Minimum of 5 mL whole blood in EDTA (purple topped) tubes
Test 2	Targeted testing of specific, well-defined <i>ACADM</i> mutations previously identified in the family	1-2 weeks \$250 per amplicon,-USD Free of charge for both parents of the proband if	83891(x1), 83894(x4), 83898(x4), 83904(x3), 83912(x1)	(1) 1 ml whole blood in EDTA (purple topped) tubes (2) Buccal swab sample

		samples submitted same week as proband		
Von Hippel-Lindau Syndrome (VHL)				
Test 1	<p>Comprehensive test, using complete sequencing and MLPA analysis for dosage alterations</p> <p>Tier 1: Sequencing of coding region</p> <p>Tier 2: Copy number analysis by MLPA</p>	<p>3 weeks,</p> <p>\$400,-USD (Tier 1)</p> <p>\$300,-USD (Tier 2)</p> <p>\$650,-USD (Tier 1 and Tier 2)</p>	<p>Tier 1: 83891(x1), 83894(x3), 83898(x3), 83904(x6), 83912(x1)</p> <p>Tier 2: 83891(x1), 83909(x3), 83896(x3), 83898(x3), 83912(x1)</p> <p>Tier 1 and Tier 2: 83891(x1), 83909(x3), 83894(x3), 83896(x3), 83898(x3), 83904(x6), 83912(x1)</p>	(1) Minimum of 5 mL whole blood in EDTA (purple topped) tubes
Test 2	Targeted testing : detection of a specific, previously known <i>VHL</i> mutation in at risk family members	<p>2 weeks</p> <p>\$250,-USD</p>	83891(x1), 83894(x4), 83898(x4), 83904(x3), 83912(x1)	(1) 1 ml whole blood in EDTA (purple topped) tubes
Test 3	Prenatal testing of specific, previously known <i>VHL</i> mutation	<p>6 working days after sample is received</p> <p>\$750,-USD</p>	83891(x3), 83894(x20), 83896(x6), 83898(x20), 83904(x8), 83912(x1)	<p>(1) Direct CVS, (minimum 10 mg pure villi)</p> <p>(2) Direct amniotic fluid (minimum 10 ml fluid)</p> <p>(3) 2 T25 flasks of cultured CVS (>70% confluent)</p> <p>(4) 2 T25 flasks of cultured amniocytes. (>70% confluent)</p>
Fragile X				
Fragile X Syndrome	Analysis of CGG expansion in the <i>FMR1</i> gene by Southern blot and PCR	<p>2-3 weeks</p> <p>\$280,-USD</p>	83891(x1), 83892(x2), 83909(x2), 83894(x1), 83896(x2), 83897(x1), 83898(x1), 83912(x1)	(1) Minimum of 3 mL whole blood in EDTA (purple topped) tubes