Prices listed correspond to *institutional rates only*; please contact the lab for insurance rates.

Genetic Test	Institutional Price (USD$) June 2014	CPT codes 2014 (Current)	TAT **	Specimen Requirements
Neurofibromatosis Type 1 - *NF1* and Legius syndrome – *SPRED1*				
NF1/SPRED1 Combo Comprehensive (NFSP1) **RNA/cDNA-based** sequencing *and* dosage (del/dup) analysis by MLPA and FISH (if a micro-deletion is identified) for the ***NF1*** gene in combination with **Comprehensive DNA-based** sequencing and dosage (del/dup) analysis for the ***SPRED1*** gene. For each patient, both *NF1* and *SPRED1* genes are analyzed comprehensively, but price depends on test result obtained.	$1400 (NF1/SPRED1 negative)	81408, 81407, 88230, 81404	25 15 (RUSH‡)	**(1)** 10 ml of whole blood in EDTA (purple topped) tubes (Minimum of 3 ml for pediatric patients) Blood samples must be received within 60-72 hours
	$1200 (NF1 mutation identified)	81408, 81407, 88230		
	$500 (NF1 TGD identified)	81407		
	$400 (SPRED1 mutation identified)	81405, 81404		
NF1 Comprehensive (NF11) **RNA/cDNA-based** sequencing *and* dosage (del/dup) analysis by MLPA and FISH (if a micro-deletion is identified) for the ***NF1*** gene only. A reduced price is charged if a micro-deletion (TGD) is found during analysis.	$1200	81408, 81407, 88230	25 15 (RUSH‡)	
	$500 (NF1 TGD identified)	81407		
NF1 DNA (NF1D) **DNA-based** sequencing *and* dosage (del/dup) analysis by MLPA for the ***NF1*** gene starting from a DNA sample or from a blood sample that arrived >72 hours after blood draw. ****Please note:** this is currently not the preferred method of NF1 testing for our lab, please contact us for more details**	$1800	81407, 81408	35	**(1)** 10 ml of whole blood in EDTA (purple topped) tubes **(2)** DNA sample Applicable for those samples that cannot be received within 60-72 hours after blood draw
SPRED1 Comprehensive (SPD1) **DNA-based** sequencing *and* dosage (del/dup) analysis by MLPA for the ***SPRED1*** gene only. A reduced price is charged if a pathogenic mutation is found during sequencing.	$650	81405, 81404	15	
	$400 (mutation identified during sequencing)	81405		
NF1/SPRED1 Known Mutation (KT2) **Targeted** detection of a specific, previously known, ***NF1* and/or *SPRED1*** mutation in at-risk family members by sequence, MLPA, and/or FISH analysis Note: FISH analysis is performed in the UAB Cytogenetic Laboratory, directed by Prof. Dr. A. Carroll	$250	81403	15	**(1)** 1 ml whole blood in EDTA (purple topped) tubes **(2)** Buccal swab sample for sequence analysis only **(3)** DNA sample
NF1 or SPRED1 Prenatal (PT2) **Targeted** detection of a specific, previously known, ***NF1*** or ***SPRED1*** mutation (includes maternal cell contamination (MCC†) analysis)	$750	81403, 81265	6	**(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) Please also send 1-5 ml of blood or buccal swab sample from mother for MCC studies.

UAB Medical Genomics Laboratory
Ph. (205) 934-5562 Fax (205) 996-2929
www.genetics.uab.edu/medgenomics

Genetic Test	Institutional Price (USD$) June 2014	CPT codes 2014 (Current)	TAT **	Specimen Requirements
NF1 CALs/NFs (NF14N) Comprehensive RNA/cDNA-based *NF1* sequencing *and* dosage (del/dup) analysis on *cultured* neural crest derived cells from affected body regions (i.e. CAL-spots and/or neurofibromas) for possible segmental/mosaic NF1 cases ****Please note:** it is required to contact the laboratory at 205-934-5562 or email medgenomics@uabmc.edu **in advance** of sending specimens for this testing option**	$2600	88233(x2), 81408, 81407, 81402(x2)	120	Dependent on size and quality of the tissue specimen received **(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
NF1 CALs (NF14C) Comprehensive RNA/cDNA-based *NF1* sequencing *and* dosage (del/dup) analysis with reflexive **DNA-based *SPRED1*** sequencing *and* dosage (del/dup) analysis on *cultured* melanocytes from café-au-lait spots in affected body regions for possible segmental/mosaic NF1 and/or Legius syndrome cases with pigmentary findings only ****Please note:** it is required to contact the laboratory at 205-934-5562 or email medgenomics@uabmc.edu **in advance** of sending specimens for this testing option**	$2600	88233, 81408, 81407, 81405, 81404	120	
Neurofibromatosis Type 2 - *NF2*				
NF2 Comprehensive (NF21) RNA/cDNA-based sequencing *and* dosage (del/dup) analysis. A reduced price is charged if a pathogenic mutation is found during sequencing. Blood samples must be received within 60-72 hours	$1100	81406, 81405, 88230	25 15 (RUSH‡)	**(1)** 10 ml of whole blood in EDTA (purple topped) tubes. (Minimum of 3 ml for pediatric patients) **(2)** DNA sample
	$800 (mutation identified during sequencing)	81406, 88230		
NF2 DNA (NF2D) DNA-based sequencing *and* dosage (del/dup) analysis by MLPA for the ***NF2*** gene starting from a DNA sample or from a blood sample that arrived >72 hours after blood draw. A reduced price is charged if a pathogenic mutation is found during sequencing.	$1100	81406, 81405	25 15 (RUSH‡)	
	$800 (mutation identified during sequencing)	81406		
NF2 Known Mutation (KT2) Targeted detection of a specific, previously known, ***NF2*** mutation in at risk family members by sequence, MLPA, and/or FISH analysis ****Please note:** FISH analysis is performed in the UAB Cytogenetic Laboratory, directed by Prof. Dr. A. Carroll **	$250	81403	10	**(1)** 1 ml whole blood in EDTA (purple topped) tubes **(2)** Buccal swab sample for sequence analysis only **(3)** DNA sample
NF2 Prenatal (PT2) Targeted detection of a specific, previously known, ***NF2*** mutation (includes maternal cell contamination (MCC†) analysis)	$750	81403, 81265	6	**(1)** Direct CVS, (minimum 10 mg pure villi) **(2)** Direct amniotic fluid (min. 10 ml fluid) **(3)** 2 T25 flasks of cultured CVS (>70% confluent) **(4)** 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.
NF2 Tumor (NF24) Comprehensive gDNA-based *NF2* testing on a minimum of 2 **different** tumor specimens by direct sequencing *and* dosage (del/dup) analysis by MLPA of the ***NF2*** gene. Any mutations found in the tumor tissue will be confirmed in blood at no additional cost if the blood sample is submitted the same week as the tumor specimen.	$1400	81405, 81406, 81403(x2)	25	**(1)** Tumor block **(2)** Flash frozen tumor sent on dry ice **(3)** Fresh tumor biopsy, immersed in culture media (PBS/RPMI) **(4)** Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen

Genetic Test	Institutional Price (USD$) June 2014	CPT codes 2014 (Current)	TAT **	Specimen Requirements
Schwannomatosis/Rhabdoid Tumor Predisposition Syndrome (RTPS) - *SMARCB1* and *LZTR1*				
SMARCB1 DNA (SB1D) **DNA-based** sequencing *and* dosage (del/dup) analysis by MLPA for the ***SMARCB1*** gene starting from a DNA sample or from a blood sample that arrived >72 hours after blood draw.	$800	81405, 81404	25 15 (RUSH‡)	**(1)** 10 ml of whole blood in EDTA (purple topped) tubes. (Minimum of 3 ml for pediatric patients) **(2)** DNA sample
LZTR1 Sequencing on Blood (LZTR1) **DNA-based** sequencing for the ***LZTR1*** gene starting from a DNA sample or from a blood sample that arrived >72 hours after blood draw.	$800	81406	20	**(1)** 10 ml of whole blood in EDTA (purple topped) tubes. (Minimum of 3 ml for pediatric patients) **(2)** DNA sample
LZTR1 or SMARCB1 Targeted (KT2) **Targeted** detection of a specific, previously known, ***SMARCB1or LZTR1*** mutation in at risk family members by sequence, MLPA, and/or FISH analysis ****Please note:** FISH analysis is performed in the UAB Cytogenetic Laboratory, directed by Prof. Dr. A. Carroll**	$250	81403	10	**(1)** 1 ml whole blood in EDTA (purple topped) tubes **(2)** Buccal swab sample for sequence analysis only **(3)** DNA sample
SMARCB1 Prenatal (PT2) **Targeted** detection of a specific, previously known, ***SMARCB1*** mutation (includes maternal cell contamination (MCC)† analysis)	$750	81403, 81265	6	**(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) Please also send 1-5 ml of blood or buccal swab sample from mother for maternal contamination studies.
SMARCB1 Schwannoma (SB14Sch) **Comprehensive DNA-based testing for Schwannomatosis** on a minimum of 2 **different** tumor specimens by direct sequencing *and* dosage (del/dup) analysis by MLPA of both the ***SMARCB1*** and ***NF2*** genes. Any mutations found in the tumor tissue will be confirmed in blood at no additional cost if the blood sample is submitted the same week as the tumor specimen.	$2200	81405(x2), 81404, 81406, 81402, 81403	30	**(1)** Tumor block **(2)** Flash frozen tumor sent on dry ice **(3)** Fresh tumor biopsy, immersed in culture media (PBS/RPMI) **(4)** Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
LZTR1 Reflexive testing on tumor (SB14Sch) **DNA-based testing for Schwannomatosis** on tumor specimens previously received by the laboratory for *SMARCB1* analysis which revealed no identifiable first hit. This testing includes direct sequencing of the ***LZTR1*** gene. Any mutations found in the tumor tissue will be confirmed in blood at no additional cost.	$800	81406	20	**(1)** Tumor block **(2)** Flash frozen tumor sent on dry ice **(3)** Fresh tumor biopsy, immersed in culture media (PBS/RPMI) **(4)** Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
SMARCB1 Rhabdoid (SB14RT) **Comprehensive DNA-based testing for RTPS** on a tumor specimen by direct sequencing *and* dosage (del/dup) analysis by MLPA of the ***SMARCB1*** gene. Any mutations found in the tumor tissue will be confirmed in blood at no additional cost if the blood sample is submitted the same week as the tumor specimen.	$1400	81405, 81404, 81402, 81403 (x2)	25	**(1)** Tumor block **(2)** Flash frozen tumor sent on dry ice **(3)** Fresh tumor biopsy, immersed in culture media (PBS/RPMI) **(4)** Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Tuberous Sclerosis Complex –TSC1/TSC2				
TSC1 Sequencing only (TSC1-S) **DNA-based** sequencing for the **TSC1** gene	$1000	81406	20	**(1)** Minimum of 5 mL whole blood in EDTA (purple topped) tubes **(2)** DNA sample

Genetic Test	Institutional Price (USD$) June 2014	CPT codes 2014 (Current)	TAT **	Specimen Requirements
TSC1 MLPA only (TSC1-M) **DNA-based** dosage (del/dup) analysis by MLPA for the **TSC1** gene	$500	81405	20	**(1)** 1 ml whole blood in EDTA (purple topped) tubes **(2)** DNA sample
TSC2 Sequencing only (TSC2-S) **DNA-based** sequencing for the **TSC2** gene	$1400	81407	20	**(1)** Minimum of 5 mL whole blood in EDTA (purple topped) tubes **(2)** DNA sample
TSC2 MLPA only (TSC2-M) **DNA-based** dosage (del/dup) analysis by MLPA for the **TSC2** gene	$500	81406	20	**(1)** 1 ml whole blood in EDTA (purple topped) tubes **(2)** DNA sample
Complete Tuberous Sclerosis Panel Testing (TSCP) **DNA-based** sequencing and dosage (del/dup) analysis by MLPA for the **TSC1** and **TSC2** gene	$2400	81408, 81407	20	**1)** 1 ml whole blood in EDTA (purple topped) tubes **(2)** DNA sample
TSC Known Mutation (KT2) **Targeted** detection of a specific, previously known, mutation in any of the genes mentioned above in at risk family members by sequence analysis	$250	81403	10	**(1)** 1 ml whole blood in EDTA (purple topped) tubes **(2)** DNA sample
TSC Prenatal (PT2) **Targeted** detection of a specific, previously known, : ***TSC1 or TSC2*** mutation (includes maternal cell contamination (MCC)[†] analysis)	$750	81322, 81265	6	**(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) Please also send 1-5 ml of blood or buccal swab sample from mother for maternal contamination studies.
Ras-o-pathy Panels – Noonan, Cardio-facio-cutaneous (CFC), and LEOPARD syndromes				
Noonan syndrome panel (NNP) **DNA-based** sequencing of 13 genes associated with a Noonan or Noonan-like phenotype: ***PTPN11, SOS1, RAF1, KRAS, NRAS, SHOC2, CBL, BRAF, MAP2K1, MAP2K2, HRAS, RIT1, and SPRED1***. In addition, this testing also includes dosage (del/dup) analysis by MLPA for the ***SPRED1*** gene.	$1500	81408, 81404	20	**(1)** Minimum of 5 mL whole blood in EDTA (purple topped) tubes
Cardio-facio-cutaneous (CFC) syndrome panel (CFCP) **DNA-based** sequencing of 4 genes associated with **CFC: *BRAF, MAP2K1, MAP2K2,* and *KRAS***	$1100	81407	20	**(1)** Minimum of 5 mL whole blood in EDTA (purple topped) tubes
LEOPARD syndrome panel (LPDP) **DNA-based** sequencing of 3 genes associated with LEOPARD syndrome: ***PTPN11, RAF1,* and *BRAF***	$1100	81407	20	**(1)** Minimum of 5 mL whole blood in EDTA (purple topped) tubes
Ras-o-pathy Single Gene Testing				
Costello syndrome (CST1) **DNA-based** sequencing of the ***HRAS*** gene	$300	81404	15	**(1)** Minimum of 5 mL whole blood in EDTA (purple topped) tubes
Metachondromatosis (MTM1) **DNA-based** sequencing of the ***PTPN11*** gene	$900	81406	15	**(1)** Minimum of 5 mL whole blood in EDTA (purple topped) tubes

Genetic Test	Institutional Price (USD$) June 2014	CPT codes 2014 (Current)	TAT **	Specimen Requirements
Single Gene Sequencing (SGS1) **DNA-based** sequencing of any single gene offered on the Ras-o-pathy panels: ***PTPN11, SOS1, RAF1, KRAS, NRAS, SHOC2, CBL, BRAF, MAP2K1, MAP2K2, SPRED1, RIT1, and HRAS*** **Please note:** it is required to contact the laboratory at 205-934-5562 or email medgenomics@uabmc.edu **in advance** of sending specimens for this testing option**	$900 (PTPN11)	81406	20	**(1)** Minimum of 5 mL whole blood in EDTA (purple topped) tubes
	$550 (SOS1, RAF1, BRAF, MAP2K1, MAP2K2)	81406		
	$300 (RIT1, KRAS, NRAS, CBL, HRAS)	81404		
	$250 (SHOC2)	81403		
	$400/$650 (SPRED1 Seq and del/dup)	Please refer to page 1 for more details.		
Ras-o-pathy Known Mutation (KT2) **Targeted** detection of a specific, previously known, mutation in any of the genes mentioned above in at risk family members by sequence analysis	$250	81403	10	**(1)** 1 ml whole blood in EDTA (purple topped) tubes **(2)** DNA sample
Ras-o-pathy Prenatal (PT2) **Targeted** detection of a specific, previously known, : ***PTPN11, SOS1, RAF1, KRAS, NRAS, SHOC2, CBL, BRAF, MAP2K1, MAP2K2, SPRED1, RIT1, or HRAS*** mutation (includes maternal cell contamination (MCC)† analysis)	$750	81322, 81265	6	**(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) Please also send 1-5 ml of blood or buccal swab sample from mother for maternal contamination studies.
PTEN Hamartoma Tumor Syndrome / Macrocephaly-Autism Syndrome – *PTEN*				
PTEN Comprehensive (PTEN1) **DNA-based** testing by sequencing *and* dosage (del/dup) analysis by MLPA for the ***PTEN*** gene. A reduced price is charged if a pathogenic mutation is found during sequencing	$1100	81323, 81321	25	**(1)** 10 ml of whole blood in EDTA (purple topped) tubes. (Minimum of 3 ml for pediatric patients; For add-on Fragile X testing for pediatric patients, we require a minimum of 5 ml) **(2)** DNA sample
	$800 (if mutation identified during sequencing)	81321		
PTEN Known Mutation (KT2) **Targeted** detection of a specific, previously known, ***PTEN*** mutation in at risk family members by sequence and/or MLPA analysis	$250	81322	10	**(1)** 1 ml whole blood in EDTA (purple topped) tubes **(2)** DNA sample
PTEN Prenatal (PT2) **Targeted** detection of a specific, previously known, ***PTEN*** mutation (includes maternal cell contamination (MCC)† analysis)	$750	81322, 81265	6	**(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) Please also send 1-5 ml of blood or buccal swab sample from mother for maternal contamination studies.
Von-Hippel Lindau Syndrome – *VHL*				
VHL Comprehensive (VHL1) **DNA-based** sequencing *and* dosage (del/dup) analysis by MLPA for the ***VHL*** gene. A reduced price is charged if a pathogenic mutation is found during sequencing	$650	81404, 81403	15	**(1)** Minimum of 5 mL whole blood in EDTA (purple topped) tubes **(2)** DNA sample
	$400 (if mutation identified during sequencing)	81404		

Genetic Test	Institutional Price (USD$) June 2014	CPT codes 2014 (Current)	TAT **	Specimen Requirements
VHL Known Mutation (KT2) **Targeted** detection of a specific, previously known, ***VHL*** mutation in at risk family members by sequence and/or MLPA analysis	$250	81403	10	**(1)** 1 ml whole blood in EDTA (purple topped) tubes **(2)** DNA sample
VHL Prenatal (PT2) **Targeted** detection of a specific, previously known, ***VHL*** mutation (includes maternal cell contamination (MCC)[†] analysis)	$750	81403, 81265	6	**(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) Please also send 1-5 ml of blood or buccal swab sample from mother for maternal contamination studies.
Autosomal Recessive Polycystic Kidney Disease - *PKHD1*				
ARPKD Comprehensive (PKD1) **DNA-based** sequencing of the entire ***PKHD1*** longest open reading frame *and* dosage (del/dup) analysis by MLPA, detecting >82% of mutations. This includes automatically reflexing through Tiers 1-3: **Tier 1**: Sequencing of 23 exons which will detect 2 clear pathogenic mutations in ~64% of ARPKD patients **Tier 2**: Sequencing of the remaining exons of the entire longest open reading frame *and* targeted analysis for a deep intronic splicing alteration in intron 46 **Tier 3**: Dosage (del/dup) analysis by MLPA/qPCR A reduced price is charged based on which tiers are necessary to identify two clear pathogenic mutations in a proband.	$2600 (Tiers 1+2+3 necessary)	81408, 81407	20 8 for Tier1+2 RUSH	**(1)** 5 ml whole blood in EDTA (purple tubes) **(2)** biopsy from skin/liver/spleen. (fresh or frozen) **(3)** 2 T25 flasks of cultured cells
	$2200 (Tiers 1+2 necessary)	81408		
	$1100 (only Tier 1 was necessary)	81406		
ARPKD Sequencing (PKDS) **DNA-based** sequencing-only of the entire ***PKHD1*** longest open reading frame, detecting ~80% of mutations **Tier 1**: Sequencing of 23 exons which will detect 2 clear pathogenic mutations in ~64% of ARPKD patients **Tier 2**: Sequencing of the remaining exons of the entire longest open reading frame *and* targeted analysis for a deep intronic splicing alteration IVS46+653 A>G (Tier 1+2 **can** be ordered as a standalone test)	$2200 (Tiers 1+2 necessary) $3200 (for RUSH):	81408		
	$1100 (only Tier 1 is necessary)	81406		
ARPKD Known Mutation (KT2) **Targeted** detection of specific, previously known, ***PKHD1*** mutation(s) in family members by sequence and/or MLPA analysis	Sequencing 1 exon: $250	1 exon: 81403	15	**(1)** 5 ml whole blood in EDTA (purple topped) tubes **(2)** Buccal swab sample
	Sequencing 2 exons: $340	2 exons: 81403(x2)		
	MLPA: $250	MLPA: 81407		
ARPKD Prenatal Targeted (PT2) **Targeted** detection of specific, previously known, ***PKHD1*** mutations (includes maternal cell contamination (MCC)[†] analysis)	$1000	81403(x2), 81265	6	**(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) Please also send 1-5 ml of blood or buccal swab sample from mother for maternal contamination studies.

ARPKD Informativity (PKDL) **Haplotype analysis** by PCR of 7 markers within and flanking the ***PKHD1*** disease interval on chromosome 6p21-cen	$400/ individual tested	81265	20	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 Please also send 1-5 ml of blood or buccal swab sample from mother for maternal contamination studies.
ARPKD Prenatal Linkage (PKDPL) **Haplotype analysis** by PCR of 7 markers within and flanking the ***PKHD1*** disease interval on chromosome 6p21-cen as a prenatal test (includes maternal cell contamination (MCC)[†] analysis)	$500/ individual tested	81265	6	

Note: A **comprehensive test** is a direct test that uses multiple complementary assays to detect all types of lesions within the entire coding region. A **targeted test** is a test that solely detects a previously characterized specific mutation within the family. A **prenatal test** is a test that detects a previously characterized specific mutation within a family on a prenatal sample and testing includes maternal cell contamination analysis.

*****Targeting testing for parents is free of charge if both parents of the proband are submitted during the same week as the proband.

******Turn-around time is **working days** starting from the day after receipt of sample.

[†] Maternal Cell Contamination Analysis is abbreviated as MCC and is performed as a part of prenatal testing.

[‡] RUSH fee for *NF1*, *NF2,* and *SMARCB1* is an additional $600