

Genetic Test	Institutional Price (USD\$)	CPT Codes	Z codes	TAT (working days)	Specimen Requirements
Lymphocyte/WBC-based Comprehensive Testing via Next-Gen Sequencing					
NF1/SPRED1 and Other RASopathy Related Conditions on Blood/Saliva					
<p>NF1- only NGS testing and copy number analysis for the NF1 gene (NF1-NG)</p> <p>This testing includes an average coverage of >1600x to allow for the identification of mosaicism as low as 3-5% of the alleles. In addition, novel variants identified in the NF1 gene will be confirmed via RNA-based analysis at no additional charge. RNA-based testing will also be provided to non-founder, multigenerational families with “classic” NF1 at no additional charge if next-generation sequencing is found negative.</p>	<p>\$1,000 \$1,600 (RUSH)</p>	<p>81408 81479</p>	<p>ZB6A9</p>	<p>30 15 (RUSH)</p>	<p>(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)</p>
<p>SPRED1-only NGS testing and copy number analysis for SPRED1 (SPD1-NG)</p> <p>This testing includes an average coverage of >1600x to allow for the identification of mosaicism as low as 3-5% of the alleles after comprehensive NF1 analysis.</p>	<p>\$800 \$1,400 (RUSH)</p>	<p>81405 81479</p>	<p>ZB6AC</p>	<p>30 15 (RUSH)</p>	<p>(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)</p>
<p>NF1/SPRED1 NGS testing and copy number analysis for NF1 and SPRED1 (NFSP-NG)</p> <p>This testing includes an average coverage of >1600x to allow for the identification of mosaicism as low as 3-5% of the alleles. In addition, free of charge testing maybe available in scenarios summarized for NF1 -only above.</p>	<p>\$1,100 \$1,700 (RUSH)</p>	<p>81408 81405 81479 (x2)</p>	<p>ZB6A8</p>	<p>30 15 (RUSH)</p>	<p>(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)</p>
<p>Noonan-only NGS panel (NNP-NG)</p> <p>This testing includes BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, and SPRED1 as well as copy number analysis for SPRED1 and LZTR1 . An average coverage of >1600x will allow for the identification of mosaicism as low as 3-5% of the alleles</p>	<p>\$1,200 \$1,800 (RUSH)</p>	<p>81442 81479 (x2)</p>	<p>ZB6AD</p>	<p>30 15 (RUSH)</p>	<p>(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)</p>
<p>RASopathy NGS panel (RAS-NG)</p> <p>This testing includes BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SPRED1 and SOS2 as well as copy number analysis for NF1, LZTR1, and SPRED1 . An average coverage of >1600x will allow for the identification of mosaicism as low as 3-5% of the alleles. In addition, free of charge testing may be available in scenarios summarized for NF1 -only above.</p>	<p>\$1,500 \$2,100 (RUSH)</p>	<p>81442 81479 (x3)</p>	<p>ZB6A6</p>	<p>30 15 (RUSH)</p>	<p>(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)</p>

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McCune Albright on Blood/Saliva					
<p><i>GNAS-only NGS testing(GNAS-NG)</i> This testing includes analysis of GNAS by NGS, exons 8 and 9 only. An average coverage of >1600x will allow for the identification of mosaicism as low as 3% of the alleles.</p>	\$700 \$1,300 (RUSH)	81479	N/A	30 15 (RUSH)	(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
Costello Syndrome Testing on Blood/Saliva					
<p><i>Costello syndrome (CST-NG)</i> This testing includes analysis of HRAS by NGS. An average coverage of >1100x will allow for the identification of mosaicism as low as 3% of the alleles.</p>	\$700 \$1,300 (RUSH)	81404	ZB67V	30 15 (RUSH)	(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
Peripheral Nerve Sheath Tumor Testing on Blood/Saliva					
<p><i>Peripheral Nerve Sheath Tumor NGS panel (PNT-NG)</i> This testing includes KRAS, LZTR1, NF1, NF2, PTPN11, and SMARCB1 as well as copy number analysis for the LZTR1, NF1, NF2, and SMARCB1. An average coverage of >1600x will allow for the identification of mosaicism as low as 3-5% of the alleles.</p>	\$1,500 \$2,100 (RUSH)	81408 81406 81479 (x4) 81405	ZB6AE	30 15 (RUSH)	(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
NF2/Schwannomatosis/Meningiomatosis Panel Testing on Blood/Saliva					
<p><i>NF2- only NGS testing and copy number analysis for the NF2 gene (NF2-NG)</i> NF2 testing by NGS sequencing and deletion/duplication analysis by MLPA. This testing includes an average coverage of >1600x to allow for the identification of mosaicism as low as 3% of the alleles. In addition, novel variants identified in the NF2 gene will be confirmed via RNA-based analysis at no additional charge.</p>	\$800 \$1,400 (RUSH)	81406 81405	ZB6AA	30 15 (RUSH)	(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
<p><i>Schwannomatosis/Multiple Schwannomas NGS Panel (SCH-NG)</i> This testing includes LZTR1, NF2, and SMARCB1 by NGS as well as copy number analysis of LZTR1, NF2, and SMARCB1. An average coverage of >1600x will allow for the identification of mosaicism as low as 3% of the alleles.</p>	\$1,500 \$2,100 (RUSH)	81406 81405 81479 (x3)	ZB67Y	30 15 (RUSH)	(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
<p><i>Meningiomatosis/Multiple Meningioma NGS Panel (MEN-NG)</i> This testing includes NF2, SMARCB1, SMARCE1, and SUFU by NGS as well as copy number analysis of NF2 and SMARCB1. An average coverage of >1500x will allow for the identification of mosaicism as low as 3% of the alleles.</p>	\$1,500 \$2,100 (RUSH)	81406 81405 81479 (x2)	ZB67L	30 15 (RUSH)	(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)

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Rhabdoid Tumor Predisposition Syndrome Panel Testing on Blood/Saliva					
<p>Rhabdoid Tumor Predisposition NGS (RT-NG) This testing includes analysis of SMARCB1 and SMARCA4 by NGS as well as copy number analysis SMARCB1. An average coverage of >1100x will allow for the identification of mosaicism as low as 3% of the alleles.</p>	\$1000 \$1,600 (RUSH)	81479 (x2)	ZB68B	30 15 (RUSH)	(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
Capillary Malformation Arteriovenous Malformation Syndrome Panel Testing on Blood/Saliva					
<p>Capillary Malformation-Arteriovenous Malformation NGS panel (RASA-NG) This testing includes analysis of RASA1 and EPHB4 by NGS as well as copy number analysis of RASA1. An average coverage of >1500x will allow for the identification of mosaicism as low as 3% of the alleles.</p>	\$1000 \$1,600 (RUSH)	81479 (x2)	ZB6AB	30 15 (RUSH)	(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
Tuberous Sclerosis Complex Testing on Blood/Saliva					
<p>Tuberous Sclerosis Complex NGS panel (TSC-NG) Testing of TSC1 and TSC2 by NGS sequencing and deletion/duplication analysis by MLPA. This testing includes an average coverage of >1600x to allow for the identification of mosaicism as low as 3% of the alleles.</p>	\$1,500 \$2,100 (RUSH)	81479 81406 81405	ZB68E	30 15 (RUSH)	(1) 3-6ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
Neurofibroma/CAL Spot Biopsies for Comprehensive Testing					
NF1/SPRED1 and Other RASopathy Panel Testing on Biopsied Samples					
<p>NF1 analysis on biopsied CALs, with reflex to SPRED1, if negative (NF14C) Comprehensive RNA/cDNA-based NF1 sequencing and copy number analysis with reflexive DNA-based SPRED1 sequencing and copy number 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**</p>	\$2,600 (for NF1 -only analysis) \$3,200 (For NF1 with SPRED1 reflex)	88233 (culture) 81408 (NF1) 81479 (NF1) 81405 (SPRED1) 81404 (SPRED1)	ZB6AG	120	Dependent on size and quality of the tissue specimen received (1) 2-3 3mm punch biopsies from CAL-spots. Special transport media required for shipping: contact us at 205-934-556 (2) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen

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<p>NF1 analysis on biopsied neurofibromas (NF14N) Comprehensive RNA/cDNA-based NF1 sequencing and copy number analysis on cultured neural crest derived cells from neurofibromas from affected body regions 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**</p>	\$2,600	88233 (culture) 81408 81479	ZB67X	120	Dependent on size and quality of the tissue specimen received (1) minimum 2 separate fresh neurofibromas. Special transport media required for shipping: contact us at 205-934-5562 (2) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Fresh/Frozen Tumor Based Comprehensive Testing					
Noonan Syndrome/RASopathy Panel Testing on Fresh/Frozen Tumor					
<p>RASopathy NGS panel (RAS-NG) This testing includes BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SPRED1 and SOS2 as well as copy number analysis for NF1, LZTR1 and SPRED1. An average coverage of >1600x will allow for the identification of mosaicism as low as 3-5% of the alleles.</p>	\$2,500- tumor	81442 81479 (x3)	ZB6A6	40	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in sterile culture media (PBS/RPMI) (3) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Peripheral Nerve Sheath Tumor Testing on Fresh/Frozen Tumor					
<p>Peripheral Nerve Sheath Tumor NGS panel (PNT-NG) This testing includes KRAS, LZTR1, NF1, NF2, PTPN11, and SMARCB1 as well as copy number analysis for the LZTR1, NF1, NF2, and SMARCB1. An average coverage of >1600x will allow for the identification of mosaicism as low as 8% of the alleles.</p>	\$2,500- tumor	81408 81406 81479 (x4) 81405	ZB6AE	40	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in sterile culture media (PBS/RPMI) (3) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
NF2/Schwannomatosis/Meningiomatosis Panel Testing on Fresh/Frozen Tumor					
<p>NF2- only NGS testing and copy number analysis for the NF2 gene (NF2-NG) NF2 testing on fresh/frozen tumor by NGS sequencing and deletion/duplication analysis by MLPA. This testing includes an average coverage of >1600x to allow for the identification of mosaicism as low as 8% of the alleles. In addition, novel variants identified in the NF2 gene will be confirmed via RNA-based analysis at no additional charge.</p>	\$1,500- tumor	81406 81405	ZB6AA	40	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in sterile culture media (PBS/RPMI) (3) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
<p>Schwannomatosis/Multiple Schwannomas NGS Panel (SCH-NG) This testing includes LZTR1, NF2, and SMARCB1 by NGS as well as copy number analysis of LZTR1, NF2, and SMARCB1. An average coverage of >1600x will allow for the identification of mosaicism as low as 8% of the alleles.</p>	\$2,500- tumor	81406 81405 81479 (x3)	ZB67Y	40	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in sterile culture media (PBS/RPMI) (3) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen

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<p>Meningiomatosis/Multiple Meningiomas NGS Panel (MEN-NG)</p> <p>This testing includes NF2, SMARCB1, SMARCE1, and SUFU by NGS as well as copy number analysis of NF2 and SMARCB1. An average coverage of >1500x will allow for the identification of mosaicism as low as 8% of the alleles.</p>	\$2,500- tumor	81406 81405 81479 (x2)	ZB67L	40	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in sterile culture media (PBS/RPMI) (3) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Rhabdoid Tumor Predisposition Syndrome Panel Testing on Fresh/Frozen Tumor					
<p>Rhabdoid Tumor Predisposition NGS panel (RT-NG)</p> <p>This testing includes analysis of SMARCB1 and SMARCA4 by NGS as well as copy number analysis of SMARCB1. An average coverage of >1100x will allow for the identification of mosaicism as low as 8% of the alleles.</p>	\$1,500- tumor	81479 (x2)	ZB68B	40	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed sterile in culture media (PBS/RPMI) (3) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Tuberous Sclerosis Complex Testing on Fresh/Frozen Tumor or Affected Tissue					
<p>Tuberous Sclerosis Complex NGS panel (TSC-NG)</p> <p>Testing of TSC1 and TSC2 on fresh/frozen tissue by NGS and deletion/duplication analysis by MLPA. This testing includes an average coverage of >1600x to allow for the identification of mosaicism as low as 8% of the alleles.</p>	\$2,500- tumor	81479 81406 81405	ZB68E	40	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in sterile culture media (PBS/RPMI) (3) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Tumor Block Based Comprehensive Testing					
NF2/Schwannomatosis/Meningiomatosis Panel Testing On Tumor Block					
<p>NF2 Tumor Comprehensive Testing (NF24)</p> <p>Comprehensive gDNA-based NF2 testing on a minimum of 2 different tumor specimens by direct sequencing and copy number analysis by MLPA of the NF2 gene. Any variants 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.</p>	\$1,500- tumor	81406 81405	ZB67Z	40	(1) Tumor block only (2) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
<p>Schwannomatosis Panel (SCHP)</p> <p>Comprehensive <u>DNA-based</u> testing for Schwannomatosis and/or mosaic Neurofibromatosis type 2 on a minimum of 2 different tumor block specimens by direct sequencing and copy number analysis by MLPA of the SMARCB1, LZTR1, and NF2 genes. Any variants 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.</p>	\$2,500- tumor	81406 (x2) 81405 (x3) 81404	ZB68A	40	(1) Tumor block only (2) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen

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Rhabdoid Tumor Predisposition Syndrome Panel Testing on Tumor Block					
<p>Rhabdoid Tumor Predisposition Testing (SB14RT) This testing includes Sanger sequencing and deletion/duplication analysis by MLPA of SMARCB1. Any variants 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.</p>	\$1,500- tumor	81405 81404	ZB68C	40	(1) Tumor block only (2) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Sanger Sequencing Based Comprehensive Testing					
NF1/SPRED1 and Other RASopathy Panel Testing					
<p>NF1 RNA-based Comprehensive Study (NF1-R) RNA/cDNA-based sequencing <i>and</i> copy number analysis by MLPA and FISH (if a micro-deletion is identified) for the NF1 gene</p>	\$1,800	88230 (culture) 81408 81479	ZB6AF	22	3-6 ml of whole blood in EDTA (purple topped) tubes **Sample must arrive within 60-72hours of collection**
<p>NF1/SPRED1 RNA-based Comprehensive Study (NFSP-R) RNA/cDNA-based sequencing <i>and</i> copy number 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</p>	\$2,000	88230 (culture) 81408 81479 (x2) 81405	ZB6AJ	22	3-6 ml of whole blood in EDTA (purple topped) tubes **Sample must arrive within 60-72hours of collection**
Von Hippel Lindau Syndrome					
<p>VHL Comprehensive (VHL1) DNA-based sequencing <i>and</i> copy number analysis by MLPA for the VHL gene. A reduced price is charged if a pathogenic variant is found during sequencing</p>	\$650	81404 81403	ZB68F	15	(1) Minimum of 5 mL whole blood in EDTA (purple topped) tubes (2) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
PTEN Related Disorder Testing					
<p>PTEN Comprehensive (PTEN1) DNA-based testing by sequencing <i>and</i> copy number analysis by MLPA for the PTEN gene. A reduced price is charged if a pathogenic variant is found during sequencing</p>	\$1,100 \$800 (if variant identified during sequencing)	81321 81323	ZB6AH	15	(1) Minimum of 5 mL whole blood in EDTA (purple topped) tubes (2) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
Medium-chain Acyl-CoA Dehydrogenase deficiency (MCADD)					
<p>MCADD comprehensive analysis (DNA-based) (MCD1) DNA-based sanger sequencing begins with targeted testing of the c.985 A>G, p.K329E variant followed by comprehensive sequencing analysis of the ACADM gene</p>	\$250 \$550	81403 (Exon 11) 81406 (full gene)	ZB6AI	15	(1) 3-6 ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)

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Fragile X syndrome					
<p>Fragile X syndrome (FRX) PCR-based analysis of the FMR1 gene If necessary for confirmatory testing, a reflex to Southern blot analysis may be performed.</p>	\$280 \$280	81243 (PCR) 81404 (Southern)	ZB6A7	15	(1) 3-6 ml whole blood in EDTA (purple topped) tubes; confirmation test specimen can be provided before or after preliminary results
Known Variant Testing					
Options Available for All Genes					
<p>Known Variant (KT2) Targeted DNA-based detection of a specific, previously known variant in any gene that is available at our lab by sequence, MLPA, and/or FISH analysis</p>	\$250	81403	ZB67K	15	(1) 3-6 ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 \geq 1.8)
<p>NGS-based Known Variant (KT2-NG) Next-generation sequencing-based targeted testing with deep coverage of the alleles. This analysis provides detection of mosaicism for a known variant present at least 3-5% variant allele fraction (VAF).</p>	\$700	81479	ZB67W	30	(1) 3-6 ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 \geq 1.8) (4) Fresh, sterile semen collection using a local sperm bank/cryobank facility
<p>Prenatal Testing (PT2) Targeted detection of a specific, previously known, variant (includes maternal cell contamination (MCC) analysis)</p>	\$750	81265 (MCC) 81403	ZB67M	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) (5) Maternal blood specimen for maternal cell contamination
Options Available for Select Genes					
<p>RNA-based Known Variant (RT2) RNA-based Targeted detection of a specific, previously known variant in the NF1 or NF2 gene. **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**</p>	\$500	88230 (culture) 81403	ZB68I	22	3-6 ml whole blood in EDTA (purple topped) tubes **Sample must arrive within 60-72hours of collection**

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Autosomal Recessive Polycystic Kidney Disease - PKHD1					
ARPKD Known Variant (KT2) Targeted detection of specific, previously known, PKHD1 variant(s) in family members by sequence analysis	Sequencing 1 exon: \$250	81403	ZB67K	15	(1) 3-6 ml whole blood in EDTA (purple topped) tubes (2) Oragene 575 saliva kit (provided by the MGL) (3) DNA sample (25ul volume at 3ug, O.D. value at 260:280 ≥1.8)
	Sequencing 2 exons: \$340	81403(x2)			
	MLPA: \$250	81407			
ARPKD Prenatal Targeted (PT2) Targeted detection of specific, previously known, PKHD1 variants (includes maternal cell contamination (MCC) analysis)	\$1,000	81403(x2) 81265 (MCC)	ZB67M	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) (5) Maternal blood specimen for maternal cell contamination
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	ZB67G	20	Please send samples from the parents and their children for informativity results (1) 3-6 ml whole blood in EDTA (purple topped) tubes (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 (4) Maternal blood specimen for maternal cell contamination
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	ZB67G	6	(1) 3-6 ml whole blood in EDTA (purple topped) tubes (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 (4) Maternal blood specimen for maternal cell contamination