Last Updated February 2023

Genetic Test	Institutional Price (USD$)	CPT Codes	Z codes	Average TAT (working days)	Specimen Requirements
Rhabdoid Tumor Predisposition Syndrome Panel Testing on Tumor Block					
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.	$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					
NF1 RNA-based Comprehensive Study (NF1-R) RNA/cDNA-based sequencing *and* copy number analysis by MLPA for the ***NF1*** gene	$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**
NF1/SPRED1 RNA-based Comprehensive Study (NFSP-R) RNA/cDNA-based sequencing *and* copy number analysis by MLPA for the ***NF1*** gene in combination with Comprehensive DNA-based sequencing and dosage (del/dup) analysis for the ***SPRED1*** gene	$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					
VHL Comprehensive (VHL1) DNA-based sequencing *and* copy number analysis by MLPA for the ***VHL*** gene.	$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					
PTEN Comprehensive (PTEN1) DNA-based testing by sequencing *and* copy number analysis by MLPA for the ***PTEN*** gene. A reduced price is charged if a pathogenic variant is found during sequencing	$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)					
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	$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					
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.	$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					
Known Variant (KT2) **Targeted DNA-based** detection of a specific, previously known variant in any gene that is available at our lab by sequence and MLPA.	$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)
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).	$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 ≥1.8) (4) Fresh, sterile semen collection using a local sperm bank/cryobank facility
Prenatal Testing (PT2) **Targeted** detection of a specific, previously known, variant (includes maternal cell contamination (MCC) analysis)	$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					
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**	$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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Genetic Test	Institutional Price (USD$)	CPT Codes	Z codes	TAT (working days)	Specimen Requirements
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	

*In the event of failure via Next Generation Sequencing (NGS), the sample will be analyzed via Sanger sequencing when possible which adds 20 working days to the average TAT.
**The TAT is not an average for prenatal analysis. The results are guaranteed within 6 working days for prenatal samples.

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