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Genetic Test	Institutional Price (USD$)	CPT Codes	Z codes	TAT (working days)	Specimen Requirements
Comprehensive Testing for Constitutional/Mosaic Mutations with Deep Coverage via NGS					
NF1/SPRED1 and Other RASopathy Related Conditions on Blood/Saliva					
NF1- only NGS testing and copy number analysis for the NF1 gene (NF1-NG) This testing includes an average coverage of 1800x to allow for the identification of mosaicism as low as 3-5% of the alleles. In addition, novel mutations 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.	$1,000 $1,600 (RUSH)	81408 81479	ZB6A9	25 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)
SPRED1-only NGS testing and copy number analysis for SPRED1 (SPD1-NG) This testing includes an average coverage of 1800x to allow for the identification of mosaicism as low as 3-5% of the alleles after comprehensive *NF1* mutation analysis.	$800 $1,400 (RUSH)	81405 81479	ZB6AC	25 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)
NF1/SPRED1 NGS testing and copy number analysis for NF1 and SPRED1 (NFSP-NG) This testing includes an average coverage of 1800x 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.	$1,100 $1,700 (RUSH)	81408 81405 81479 (x2)	ZB6A8	25 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)
Noonan-only NGS panel (NNP-NG) This testing includes ***BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SPRED1*** and ***SOS2*** as well as copy number analysis for the SPRED1gene. An average coverage of 1800x will allow for the identification of mosaicism as low as 3-5% of the alleles	$1,200 $1,800 (RUSH)	81442 81479	ZB6AD	25 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)
RASopathy NGS panel (RAS-NG) This testing includes ***BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SPRED1*** and ***SOS2*** as well as copy number analysis for the ***NF1*** and ***SPRED1*** gene. An average coverage of 1800x 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.	$1,500 $2,100 (RUSH)	81442 81479 (x2)	ZB6A6	25 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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Costello Syndrome Testing on Blood/Saliva					
Costello syndrome (CST-NG) This testing includes analysis of ***HRAS*** by NGS. An average coverage of 1800x will allow for the identification of mosaicism as low as 3% of the alleles.	$700 $1,300 (RUSH)	81404	ZB67V	25 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					
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 1800x will allow for the identification of mosaicism as low as 3-5% of the alleles.	$1,500 $2,100 (RUSH)	81408 81406 81479 (x4) 81405	ZB6AE	25 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					
NF2- only NGS testing and copy number analysis for the NF2 gene (NF2-NG) ***NF2*** testing on either blood or fresh/frozen tumor by NGS sequencing and deletion/duplication analysis by MLPA. This testing includes an average coverage of 1800x to allow for the identification of mosaicism as low as 3% of the alleles. In addition, novel mutations identified in the ***NF2*** gene will be confirmed via RNA-based analysis at no additional charge.	$800 $1,400 (RUSH)	81406 81405	ZB6AA	25 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)
Schwannomatoisis/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 1800x will allow for the identification of mosaicism as low as 3% of the alleles.	$1,500 $2,100 (RUSH)	81406 81405 81479 (x3)	ZB67Y	25 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)
Meningiomatosis/Multiple Meningioma NGS Panel (MEN-NG) This testing includes ***NF2, SMARCB1, SMARCE1,*** and ***SUFU*** by NGS as well as copy number analysis of ***NF2*** and ***SMARCB1*** . An average coverage of 1800x will allow for the identification of mosaicism as low as 3% of the alleles.	$1,500 $2,100 (RUSH)	81406 81405 81479 (x2)	ZB67L	25 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)
Rhabdoid Tumor Predisposition Syndrome Panel Testing on Blood/Saliva					
Rhabdoid Tumor Predisposition NGS (RT-NG) This testing includes analysis of **SMARCB1** by NGS as well as copy number analysis. An average coverage of 1800x will allow for the identification of mosaicism as low as 3% of the alleles.	$800 $1,400 (RUSH)	81479 (x2)	ZB68B	25 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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Capillary Malformation Arteriovenous Malformation Syndrome Panel Testing on Blood/Saliva					
Capillary Malformation-Arteriovenous Malformation NGS panel (RASA-NG) This testing includes analysis of **RASA1** by NGS as well as copy number analysis. An average coverage of 1800x will allow for the identification of mosaicism as low as 3% of the alleles.	$800 $1,400 (RUSH)	81479 (x2)	ZB6AB	25 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					
Tuberous Sclerosis Complex NGS panel (TSCP-NG) Testing of ***TSC1*** and ***TSC2*** on either blood or fresh/frozen tumor by **NGS** sequencing and deletion/duplication analysis by MLPA. This testing includes an average coverage of 1800x to allow for the identification of mosaicism as low as 3% of the alleles.	$1,500 $2,100 (RUSH)	81479 81406 81405	ZB68E	25 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 Biopsided Samples					
NF1 analysis on biopsied CALs, with reflex to SPRED1, if negative (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**	$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 5mm 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
NF1 analysis on biopsied neurofibromas (NF14N) Comprehensive RNA/cDNA-based ***NF1*** sequencing *and* dosage (del/dup) 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**	$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

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Fresh/Frozen Tumor Based Comprehensive Testing					
Peripheral Nerve Sheath Tumor Testing on Fresh/Frozen Tumor					
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 1800x will allow for the identification of mosaicism as low as 8% of the alleles.	$2,500- tumor	81408 81406 81479 (x4) 81405	ZB6AE	30	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in 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					
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 1800x to allow for the identification of mosaicism as low as 8% of the alleles. In addition, novel mutations identified in the ***NF2*** gene will be confirmed via RNA-based analysis at no additional charge.	$1,500- tumor	81406 81405	ZB6AA	30	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in culture media (PBS/RPMI) (3) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Schwannomatoisis/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 1800x will allow for the identification of mosaicism as low as 8% of the alleles.	$2,500- tumor	81406 81405 81479 (x3)	ZB67Y	30	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in culture media (PBS/RPMI) (3) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Meningiomatosis/Multiple Meningiomas NGS Panel (MEN-NG) This testing includes ***NF2, SMARCB1, SMARCE1*** , and ***SUFU*** by NGS as well as copy number analysis of ***NF2*** and ***SMARCB1*** . An average coverage of 1800x will allow for the identification of mosaicism as low as 8% of the alleles.	$2,500- tumor	81406 81405 81479 (x2)	ZB67L	30	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in 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					
Rhabdoid Tumor Predisposition NGS panel (RT-NG) This testing includes analysis of **SMARCB1** by NGS as well as copy number analysis. An average coverage of 1800x will allow for the identification of mosaicism as low as 8% of the alleles.	$1,500- tumor	81479 (x2)	ZB68B	30	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in 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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Tuberous Sclerosis Complex Testing on Fresh/Frozen Tumor or Affected Tissue					
Tuberous Sclerosis Complex NGS panel (TSCP-NG) Testing of ***TSC1*** and ***TSC2*** on either blood or fresh/frozen tumor by **NGS** sequencing and deletion/duplication analysis by MLPA. This testing includes an average coverage of 1800x to allow for the identification of mosaicism as low as 8% of the alleles.	$2,500- tumor	81479 81406 81405	ZB68E	30	(1) Flash frozen tumor sent on dry ice (2) Fresh tumor or affected tissue biopsy, immersed in 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					
NF2 Tumor Comprehensive Testing (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.	$1,500- tumor	81406 81405	ZB67Z	30	**(1) Tumor block only** (2) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Schwannomatosis Panel (SCHP) Comprehensive DNA-based testing for Schwannomatosis and/or mosaic Neurofibromatosis type 2 on a minimum of 2 **different** tumor block specimens by direct sequencing *and* dosage (del/dup) analysis by MLPA of the ***SMARCB1, LZTR1, 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.	$2,500- tumor	81406 (x2) 81405 (x3) 81404	ZB68A	30	**(1) Tumor block only** (2) Blood specimen for confirmation at no additional charge if sent within a week of the tumor specimen
Rhabdoid Tumor Predisposition Syndrome Panel Testing on Tumor Block					
Rhabdoid Tumor Predisposition Testing (SB14RT) This testing includes Sanger sequencing and deletion/duplication analysis by MLPA of ***SMARCB1*** **.** 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.	$1,500- tumor	81405 81404	ZB68C	30	**(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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Sanger Sequencing Based Comprehensive Testing					
NF1/SPRED1 and Other RASopathy Panel Testing					
NF1 RNA-based Comprehensive Study (NF1-R) RNA/cDNA-based sequencing *and* dosage (del/dup) analysis by MLPA and FISH (if a micro-deletion is identified) for the ***NF1*** gene	$1,800	88230 (culture) 81408 81479	ZB6AF	15	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* 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	$2,000	88230 (culture) 81408 81479 (x2) 81405	ZB6AJ	15	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* dosage (del/dup) analysis by MLPA for the ***VHL*** gene. A reduced price is charged if a pathogenic mutation is found during sequencing	$650 $400 (if mutation identified during sequencing)	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* dosage (del/dup) analysis by MLPA for the ***PTEN*** gene. A reduced price is charged if a pathogenic mutation is found during sequencing	$1,100 $800 (if mutation 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/MCD2) **DNA-based** sanger sequencing begins with targeted testing of the c.985 A>G, p.K329E mutation followed by comprhensive 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)
Fragile X syndrome					
Fragile X syndrome (FRX) **PCR-based** analysis of the FMR1 gene followed by reflexive southern blot analysis, if necessary	$280 $280	81243 (PCR) 81404 (Southern)	ZB6A7	15	(1) 3-6 ml whole blood in EDTA (purple topped) tubes

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Known Mutation Testing					
Options Available for All Genes					
Known Mutation (KT2) **Targeted DNA-based** detection of a specific, previously known mutation in any gene that is available at our lab by sequence, MLPA, and/or FISH analysis	$250	81403	ZB67K	10	(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 Mutation (KT2-NG) **Next-generation sequencing-based** targeted testing with deep coverage of the alleles. This analysis provides detection of mosaicism for a known mutation present at least 3-5% mutant allele fraction (MAF).	700	81479	ZB67W	25	(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, mutation (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 Mutation (RT2) **RNA-based Targeted** detection of a specific, previously known mutation in the ***NF1, NF2, TSC,*** **or** ***SMARCB1*** 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 (cultute) 81403	ZB68I	20	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 Mutation (KT2) **Targeted** detection of specific, previously known, ***PKHD1*** mutation(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
	Sequencing 2 exons: $340	81403(x2)			
	MLPA: $250	81407			
ARPKD Prenatal Targeted (PT2) **Targeted** detection of specific, previously known, ***PKHD1*** mutations (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	

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