UAB MEDICAL GENOMICS LABORATORY

Next-Gen Sequencing-based Known Variant Testing (KT2-NG)

Ordering Information

Acceptable specimen types:

- Fresh blood sample (3-6 ml EDTA; no time limitations associated with receipt)
- Saliva (OGR-575 DNA Genotek; kits are provided upon request)
- DNA (extracted from lymphocyte cells; a minimum volume of 25µL at 3µg; O.D. of 260:280nm ≥1.8; must be extracted in a CLIA or equivalent certified lab)
- Fresh, sterile semen collection using a local sperm bank/cryobank facility

Turnaround time:

30 working days

Price, CPT codes, and Z code:

$800 (USD – institutional/self-pay);

CPT: 81479

Z code: ZB67W

Candidates for this test:

Patients who want to prepare for prenatal/pre-implantation diagnosis and for predictive testing for individuals at risk of inheriting an already known pathogenic variant with detection of mosaicism for at least 3-5% of the variant allele fraction (VAF)

720 Twentieth Street South, Suite 330 Phone: (205) 934-5562
Birmingham, Alabama 35294-0005 Fax: (205) 996-2929
www.uab.edu/medicine/genetics/medical-genomics-laboratory

Specimen shipping and handling:

- Please find acceptable specimen type above.
- All submitted specimens must be sent at room temperature. DO NOT ship on ice.
- Specimens must be packaged to prevent breakage and absorbent material must be included in the package to absorb liquids in the event that breakage occurs. Also, the package must be shipped in double watertight containers (e.g. a specimen pouch + the shipping company's diagnostic envelope).
- To request a sample collection kit, please visit the website or email medgenomics@uabmc.edu to complete the specimen request form.
- Please contact the MGL (via email at medgenomics@uabmc.edu, or via phone at 205-934-5562) prior to sample shipment and provide us with the date of shipment and tracking number of the package so that we can better ensure receipt of the samples.

Required forms:

- Test Requisition Form
- Form for Customs (for international shipments)

Note: Detailed and accurate completion of this document is necessary for reporting purposes. The Medical Genomics Laboratory issues its clinical reports based on the demographic data provided by the referring institution on the lab requisition form. It is the responsibility of the referring institution to provide accurate information. If an amended report is necessary due to inaccurate or illegible documentation, additional reports will be drafted with charge.

Requests for testing may not be accepted for the following reasons:

- No label (patients full name and date of collection) on the specimens
- No referring physician's or genetic counselor's names and addresses
- No billing information

- DNA samples must be extracted in a CLIA or equivalent certified lab

For more information, test requisition forms, or sample collection and mailing kits, please call: 205-934-5562.

Test Description

We offer **targeted detection** of a previously characterized pathogenic variant within the family. DNA is extracted directly from the sample and the target region is amplified and analyzed using Next-Generation sequencing-based targeted testing with deep coverage of the alleles. This analysis provides detection of mosaicism for a known variant present in at least 3-5% variant allele fraction (or lower, depending on the gene analyzed). To offer this testing service, the proband's variant must be identified by our laboratory before testing relatives.

REFERENCES available on website.

Other related testing options:

- Prenatal Targeted Testing (PT2)
- RNA-based Known Variant Testing (RT2)
- Known Variant Testing (KT2)