UAB MEDICAL GENOMICS LABORATORY

Known Mutation Testing (KT2)

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)

Turnaround time:

10 working days

Price, CPT codes, and Z code:

$250 (USD – institutional/self-pay);

CPT: 81403

Z code: ZB67K

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 variant.

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 specific, previously characterized variant in any gene that is available in our lab. Analysis can be performed by Sanger sequence, MLPA, and/or FISH. To offer this testing service, the proband's variant must be identified by our laboratory before testing relatives.

Important information regarding NF1:

With the largest dataset of NF1 genotypes matched with phenotypes, any genotype-phenotype correlations identified will be reported in real time. Confirmatory testing of reportable variants is performed using orthogonal methods as needed. For novel *NF1* variants of unknown significance, we offer *free of charge* targeted RNA-based testing to assess the effect of the variant on splicing and enhance the correct classification/ interpretation of this novel variant.

Relevant family members of a proband with a (novel or previously identified) variant of unknown significance are offered *free of charge* targeted analysis as long as accurate phenotypic data are provided by a health care professional to enhance the interpretation. There is no limitation to the number of relatives that can be tested free of charge in such families.

Mosaicism is often present in sporadic patients with an *NF1* microdeletion and has important repercussions for counseling. Evaluation by FISH analysis on 200 interphase chromosomes can be offered in such cases.

REFERENCES available on website.

Other related testing options:

- Prenatal Targeted Testing (PT2)
- RNA-based Known Mutation Testing (RT2)
- Next-Gen Sequencing-based Known Mutation Testing (KT2-NG)