UAB MEDICAL GENOMICS LABORATORY

Rhabdoid Tumor Predisposition Syndrome by Next-Gen Sequencing (RT-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)
- Flash frozen tumor sent on dry ice
- Fresh tumor or affected tissue biopsy, immersed in sterile culture media (PBS/RPMI)

Turnaround time:

30 working days for blood, saliva, or DNA; 40 working days for fresh/frozen tumor

Price, CPT codes, and Z code:

$1000 for blood, saliva, or DNA (USD – institutional/self-pay);

$1,500 for fresh/frozen tumor (USD – institutional/self-pay);

CPT: 81479 (x2)

Z code: ZB68B

Candidates for this test:

Patients with rhabdoid tumors with or without confirmed *SMARCB1*-loss identified by immunohistochemistry staining

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.

Disorder Background

Rhabdoid tumors are rare, aggressive childhood cancers that most often develop in the kidney (Malignant Rhabdoid Tumor, MRT) and central nervous system (Atypical Teratoid/Rhabdoid Tumor, AT/RT). These lesions can occur spontaneously or as part of hereditary Rhabdoid Tumor Predisposition Syndrome (RTPS). In comparison to sporadic isolated rhabdoid tumors, the syndromic form is associated with an increased risk of developing multiple tumors at younger ages and schwannomas (benign nerve sheath tumors) that present primarily in adulthood. (Sevenet N. et al, 1999: Hum. Mol. Genet. 8:2359-68). Both *SMARCB1* and *SMARCA4* have been associated with rhabdoid tumor predisposition syndrome.

Test Description

The **Rhabdoid Tumor Predisposition Syndrome by NGS** involves sequencing of *SMARCB1* [and *SMARCA4* when testing on blood, saliva, or extracted DNA] as well as **deletion/duplication analysis** of the entire coding region of *SMARCB1*. The test uses an extensively customized and optimized set of Agilent HaloPlex capture probes, followed by sequencing of overlapping amplicons within the regions of interest using 300bp paired-end Illumina sequencing chemistry. Each coding exon plus ~50bp of flanking intronic sequence are simultaneously sequenced. 5' and 3' untranslated sequences are not included.

The average coverage is ~1100x with 91% of the coding regions ≥350x and 95% ≥200x. The minimum coverage for any additional areas is >30x. Variant and copy number calls are made using a unique bioinformatics pipeline detecting all types of variants including single nucleotide substitutions, indels, and frameshifts caused by deletion/ duplication up to 112bp.

REFERENCES available on website.

Other related testing options:

- Rhabdoid Tumor Predisposition Syndrome Analysis on Tumor Block (SB14RT)
- Meningiomatosis/Multiple Meningioma Panel by Next-Gen Sequencing (MEN-NG)

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