Department of Genetics

Medical Genomics Laboratory

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Schwannomatosis/Meningiomatosis testing at UAB

The UAB Medical Genomics Laboratory (MGL) is pleased to offer several custom-designed next generation sequencing panels for patients with a Schwannomatosis related phenotype. The Schwannomatosis NGS panel includes 3 genes (*NF2, SMARCB1, LZTR1*) and the Meningiomatosis panel includes 4 genes (*NF2, SMARCB1, SMARCE1, SUFU*). Each panel covers all of the coding regions and ~50bp intronic sequences flanking all exons of the targeted genes with deep coverage (average coverage >1500x with 99% of the coding region covered at ≥350x and 100% covered at ≥200x). This allows for detection of very low-level mosaicism by sequencing, as low as 3-5% of the alleles (regions covered by 350x and 200x respectively) with >95% confidence.

Important Consideration	Meningiomatosis Panel	Schwannomatosis Panel
Indications of Testing	• Patients with multiple meningiomas with or without the presence of schwannomas • Patients who have at least one meningioma with negative sanger sequencing analysis for the schwannomatosis genes via blood or tumor	• Patients with an ambiguous phenotype including one or more schwannomas, with/without additional features associated with *NF2*-related schwannomatosis. • Sporadic patients who are concerning for mosaicism for schwannomatosis
Specimen Types	Blood, Saliva, DNA, Fresh/Frozen Tumor	Blood, Saliva, DNA, Fresh/Frozen Tumor, FFPE tumor
Detection of Mosaicism	100% of the coding regions provides deeper coverage with the ability to identify substitution variants as low as 3% of the alleles.	Mosaicism detection is the same as described for the meningiomatosis panel for specimens analyzed via next-generation sequencing Tumor blocks are sent to the UAB pathology department to confirm that the specimen contains >70% tumor cells. Sanger sequencing can identify mutations present in >30% of the tumor cells analyzed. Mosaicism detection is dependent upon the quality of the material provided.
Suggested Testing Algorithm	Analysis of Fresh/Frozen tumor, if available. If unavailable, analysis of a blood specimen is suggested.	Analysis of Fresh/Frozen tumor, if available. Tumor testing via FFPE tumor is also an option. If tumor is unavailable, analysis of a blood specimen is suggested.
Copy Number Analysis	Provided via MLPA	Provided via MLPA
Tested Genes	*NF2, SMARCB1, SMARCE1, SUFU*	*NF2, SMARCB1, LZTR1*