Henry Ford Health System
CENTER FOR PRECISION DIAGNOSTICS

Hereditary Neuroendocrine Tumor Disorders Risk Panel (13 genes)

FH	*MAX*	*MEN1*	*NF1*	*RET*	*SDHAF2*	*SDHB*	*SDHC*	*SDHD*	*TMEM127*
TSC1	*TSC2*	*VHL*							

Indication

A proportion of neuroendocrine cancer cases are caused by mutations in cancer predisposition genes. Neuroendocrine tumors from the gastrointestinal and pancreatobiliary tracts are heterogeneous tumors with diverse biologic and clinical behaviors that vary according to the primary tumor origin, type of neuroendocrine cell, and pathologic features. Pheochromocytomas and paragangliomas are genetically heterogeneous neural crest-derived cancers, with almost one third having a germline origin. This panel includes genes responsible for very rare hereditary cancer syndromes, such multiple endocrine neoplasia (*MEN1*, *RET*), tuberous sclerosis complex (*TSC1*, *TSC2*), neurofibromatosis type I (*NF1*), Von Hippel-Lindau syndrome (*VHL*), and hereditary paraganglioma-pheochromocytoma syndromes (*MAX*, *SDHAF2*, *SDHB*, *SDHC*, *SDHD*, *TMEM127*), along with another gene associated with increased risk for developing neuroendocrine tumors (*FH*).

Hereditary cancer syndrome is a genetic predisposition to develop certain types of cancers, often at an early age. Hereditary cancer risk assessment is performed to identify patients and families who may be at risk. Clues that a hereditary cancer syndrome may be present include the following:

- Cancer diagnosed at an unusually young age
- Several different types of cancer in the same person
- Multiple primary tumors
- Several close blood relatives that have the same type of cancer, especially when on the same side of the family
- Unusual presentation of a specific type of cancer
- The presence of birth defects that are known to be associated with inherited cancer syndromes
- Occurrence of certain types of adult cancer in which the probability of harboring a hereditary cancer syndrome is high (i.e. triple negative breast cancer; ovarian, tubal or peritoneal cancer; colorectal cancer or endometrial cancer with DNA mismatch repair deficiency)

Testing method

Next Generation Sequencing (NGS) provides coverage of all coding exons and noncoding DNA in exon-flanking regions (on average 50 bp) enriched using hybrid capture Illumina TruSight Cancer Sequencing Panel. Single base pair (point) mutations, small insertions/deletions (1-25 bp), complex insertions and deletions, or larger deletions and duplication (<100 bp) are detected using a combination of clinically validated computational data analysis methods for sequence variant calling, filtering, and annotation. Gross deletions and duplications at each targeted gene and exon are evaluated through comparative depth of coverage analysis of NGS targeted sequencing data using clinically-validated analysis algorithm. All reportable copy number variants are confirmed by independent methodology using gene-specific Multiplex Ligation-dependent Probe Amplification (MLPA) or genome-wide SNP microarray assay.

Turnaround time
5-10 business days

Sample requirements
3 ml peripheral blood in EDTA (lavender) top tube
Specimen stability: Ambient - 72 hours; Refrigerated - 1 week

CPT codes
81437, 81438, G0452

References

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Ship Specimens to:

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