Clinical Use
- Diagnose or exclude pheochromocytoma or paraganglioma caused by mutations in the SDHB (test code 19568X), SDHD (test code 19567X), and VHL (test code 19571X) genes
- Screen individuals who have a family history of pheochromocytoma and/or paraganglioma
- Assist in treatment selection for patients with a gene mutation

Reference Range
Negative (no mutations detected)

Interpretive Information
SDHB mutation present
- Abdominal or thoracic pheochromocytoma
- Paraganglioma type 4
SDHD mutation present
- Head or neck pheochromocytoma
- Paraganglioma type 1
VHL mutation present
- Adrenal or thoracic pheochromocytoma
- von Hippel-Lindau syndrome

Clinical Background
Pheochromocytomas are catecholamine-producing tumors that arise from chromaffin cells, the largest compact collection of which is in the adrenal medulla. Tumors that arise within the adrenal medulla are referred to as pheochromocytomas, whereas those arising outside the adrenal are called paragangliomas or extraadrenal pheochromocytomas. These tumors are characterized by hypertension and elevated levels of catecholamines and should be differentiated from ganglioneuromas. Although affecting only a small percentage of patients with hypertension, pheochromocytomas are usually curable; however, they are often fatal if left untreated. Pheochromocytoma is familial in up to 25% of affected individuals; age of onset is <30 years. The genes involved include NF-1; RET (see MEN and FMTC Mutations, test code 36587X); succinate dehydrogenase subunits B (SDHB), C (SDHC), and D (SDHD); and VHL. Pheochromocytoma causing NF-1 and SDHC mutations are rare.

Method
- Polymerase chain reaction (PCR) and DNA sequencing
- Analytical specificity: SDHB (8 exons) (test code 19568X), SDHD (4 exons) (test code 19567X), and VHL (3 exons) (test code 19571X); large deletions and deep intronic mutations affecting mRNA splicing are not detected

Specimen Requirements
5 mL room temperature whole blood
3 mL minimum
Collect in lavender-top (EDTA) tube