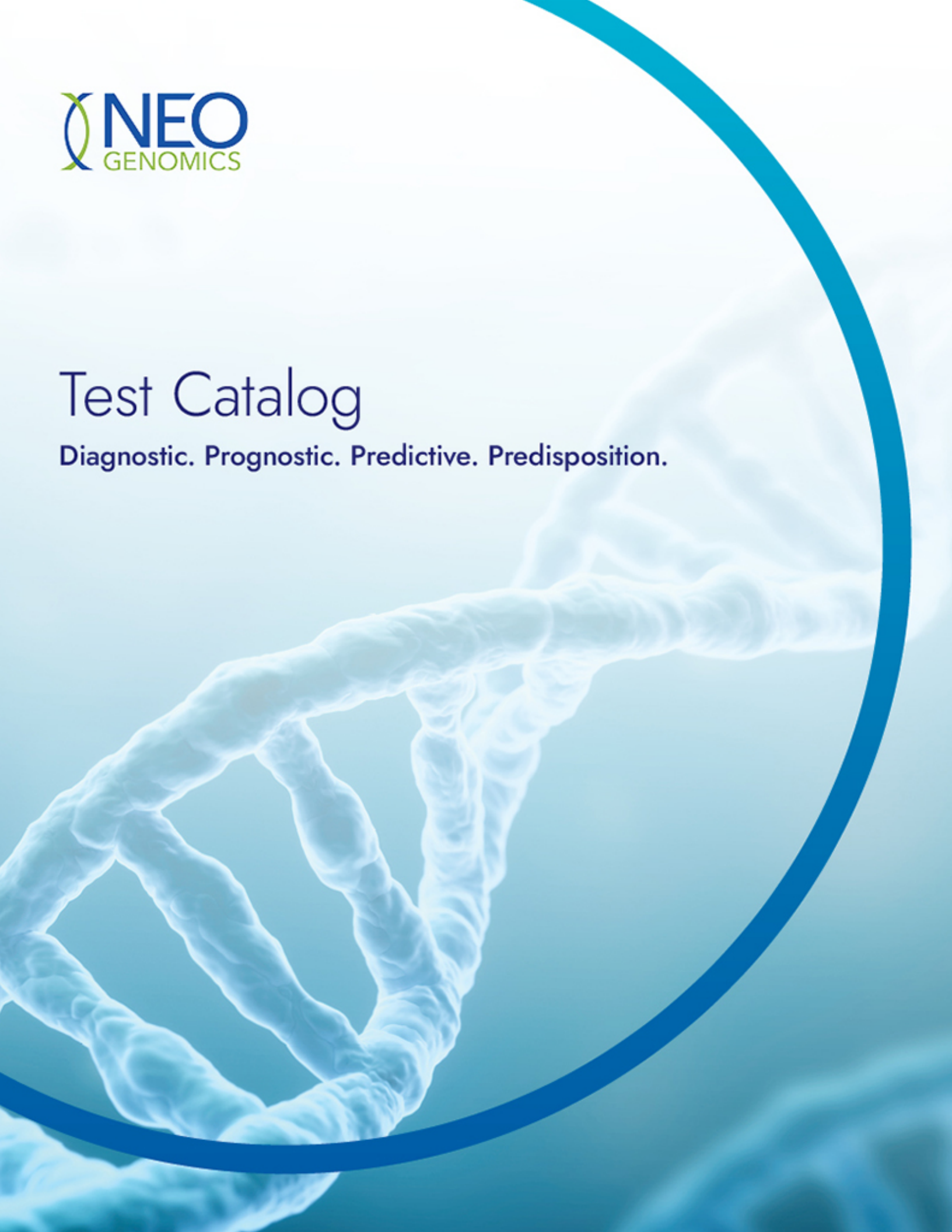
NEO GENOMICS

Test Catalog

Diagnostic. Prognostic. Predictive. Predisposition.



BRCA1/2 Mutation Analysis for Tumors

Methodology
Molecular

Test Description
BRCA1 and BRCA2 mutation analysis is performed by next-generation sequencing of all coding exons of the BRCA1 and BRCA2 genes to detect point mutations and small insertions/deletions. This test does not detect large deletions or duplications.

This test is specifically for tumor specimens; please see our Hereditary Cancer Testing menu for germline (peripheral blood) testing requirements.

Clinical Significance
BRCA1 and BRCA2 mutations account for a significant fraction of hereditary breast and ovarian cancer (HBOC) and impart increased risks for additional cancers including prostate, pancreatic, and melanoma. Both genes have roles in tumor suppression and DNA repair. Tumors with mutations may respond to PARP inhibitors and be sensitive to platinum-based therapy. Genetic counseling and germline testing may be considered if a tumor mutation is detected as tumor mutations may be somatic or germline. Large gene deletions and duplications account for approximately 10% of mutations and will not be detected by this test.

Specimen Requirements
- **FFPE tissue:** Paraffin block is preferred. Alternatively, send 1 H&E slide plus 5-10 unstained slides cut at 5 or more microns. Please use positively-charged slides and 10% NBF fixative. Do not use zinc fixatives.

Storage & Transportation
Use cold pack for transport. Make sure cold pack is not in direct contact with specimen.

CPT Code(s)*
81163x1

New York Approved
Yes

Level of Service
Global

Turnaround Time
14 days

References

1. Petrucelli N, Daly MB, Pal T. BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer. GeneReviews. https://www.ncbi.nlm.nih.gov/books/NBK1247/ Updated December 15, 2016. Accessed February 21, 2019.
2. Meric-Bernstam F, Brusco L, Daniels M, et al. Incidental germline variants in 1000 advanced cancers on a prospective somatic germline profiling protocol. Ann Oncol. 2016;27:795-800.
3. Judkins T, Rosenthal E, Arnell C, et al. Clinical signficiance of large rearrangements in BRCA1 and BRCA2. Cancer. 2012; 118:5210-5216.
4. Ewald IP, Ribeiro PLI, Palmero EI, et al. Genomic rearrangements in BRCA1 and BRCA2: A literature review. Genet Mol Biol. 2009;32(3):437-446.;

*The CPT codes provided with our test descriptions are based on AMA guidelines and are for informational purposes only. Correct CPT coding is the sole responsibility of the billing party.

Please direct any questions regarding coding to the payor being billed.

NeoGenomics Laboratories is a specialized oncology reference laboratory providing the latest technologies, testing partnership opportunities, and interactive education to the oncology and pathology communities. We offer the complete spectrum of diagnostic services in molecular testing, FISH, cytogenetics, flow cytometry, and immunohistochemistry through our nation-wide network of CAP-accredited, CLIA-certified laboratories.

Committed to research as the means to improve patient care, we provide Pharma Services for pharmaceutical companies, in vitro diagnostic manufacturers, and academic scientist-clinicians. We promote joint publications with our client physicians. NeoGenomics welcomes your inquiries for collaborations. Please contact us for more information.

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