M E M O R A N D U M # 17

TO: UNC Hospitals Attending Physicians, Housestaff, Nursing Coordinators, Department Heads and Supervisors

FROM: Karen Weck, MD, Director of Molecular Genetics
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SUBJECT: BRCA1 and BRCA2 Testing for Hereditary Breast and Ovarian Cancer

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The UNC Molecular Genetics Laboratory is pleased to announce the availability of a test to detect mutations in the *BRCA1* and *BRCA2* genes associated with hereditary breast and ovarian cancer.

Clinical indications for testing for *BRCA1* and *BRCA2* mutations include individuals with a personal or family history suggestive of hereditary breast or ovarian cancer. It is strongly recommended that patients receive genetic counseling prior to ordering *BRCA1/2* mutation testing (call the UNC Cancer Genetics Clinic at 919-843-8724 for an appointment).

Method:

1) **Full gene mutation analysis for *BRCA1* and *BRCA2*** is performed by massively parallel (next generation) sequencing to detect mutations, and multiplex ligation-dependent probe amplification (MLPA) to detect gene deletions and duplications.

The following two tests are also available:

2) **The *BRCA1/2* Ashkenazi Founder Mutation Panel** detects only the three founder mutations: *BRCA1* c.68_69delAG (previously called 185delAG), *BRCA1* c.5266dupC (previously called 5382insC), and *BRCA2* c.5946delT (previously called 6174delT).

3) **Test for known familial mutation in *BRCA1* or *BRCA2*** is performed by targeted dideoxy sequencing or MLPA, as indicated by the familial mutation.

Sample requirements:
3.0 mL peripheral blood collected in an EDTA (lavender-top) or ACD (pale yellow-top) tube.

To test a known familial mutation, a copy of the prior laboratory report for the affected family member is required, and a control blood sample from the affected family member is strongly preferred.

Turnaround time: The turn-around time of testing is two weeks.

For further information, contact the UNC Molecular Genetics Laboratory at 984-974-1825 or visit the laboratory website: http://www.uncmedicalcenter.org/uncmc/professional-education-services/mclendon-clinical-laboratories/directory/molecular-pathology-and-genetics/