MEMORANDUM #19

TO: UNC Medical Center Attending Physicians, Housestaff, Clinical Nurse Coordinators, Department Heads and Supervisors

FROM: Karen E. Weck MD, Director of Molecular Genetics Laboratory
Herbert C. Whinna MD, Medical Director, McLendon Clinical Laboratories

SUBJECT: Molecular Testing by NextGeneration Sequencing for mutations associated with Primary Ciliary Dyskinesia

DATE: January 24, 2017

Effective immediately, The UNC Hospitals Molecular Genetics Laboratory performs DNA sequencing of 35 genes associated with primary ciliary dyskinesia (PCD) and an additional two genes associated with conditions that present similarly to PCD. This test replaces previous testing in our laboratory for PCD involving Sanger sequencing of selected exons of two genes (*DNAI1* and *DNAH5*). Testing is performed by massively parallel (Next-Generation Sequencing) on an Illumina MiSeq instrument.

Clinical Indications for Molecular Genetic Testing:
PCD molecular genetic testing is performed for the purpose of diagnosis of PCD, to determine carrier status, or as confirmatory diagnostic testing. Indications include: 1) patients with clinical disease compatible with PCD, but without a defined etiology such as cystic fibrosis (CF), 2) neonatal respiratory distress in term neonates, 3) suppurative airways disease of unknown etiology, even with normal situs, 4) persistent/chronic cough and sinusitis, 5) non-CF bronchiectasis, 6) severe middle ear disease, 7) situs inversus totalis or situs ambiguus/heterotaxy, 8) congenital heart disease with situs inversus totalis or situs ambiguus 9) non-CF male infertility in conjunction with other features of PCD, for example airway disease or situs hydrocephalus, and 11) a family history of PCD/KS.

Specimen requirements: Requests for testing must be accompanied by a clinical criteria form (available on our website at http://www.pathology.unc.edu/labs/lablist_molpath.htm). The preferred sample is EDTA (purple top) or ACD (pale yellow top) anticoagulated blood which may be refrigerated up to 48 hours before analysis. Obtaining informed consent for testing is the responsibility of the ordering physician. Genetic counseling is recommended; for help with genetic counseling please call 919-966-4380.

For further information, consult the Molecular Genetics Laboratory website:
http://www.uncmedicalcenter.org/mclendon-clinical-laboratories/directory/molecular-pathology-and-genetics/ or call Dr. Weck at (984) 974-1825. E-mail: kweck@unc.edu