**Ciliary Disorders**

Primary Ciliary Dyskinesis (PCD) is a heterogeneous congenital disease inherited in an autosomal recessive fashion that is thought to affect 1 in 10,000 to 40,000 individuals. It is characterized by abnormal mucociliary transport due to abnormal ciliary motility generally related to ultrastructural changes within cilia. Patients often present with recurrent lower respiratory infections, sinusitis, & otitis. The condition may also be associated with situs inversus (~50% of patients) and impaired fertility.

The diagnosis of PCD is supported by examination of cross sections of cilia by transmission electron microscopy to confirm pathognomonic abnormalities associated with the condition.

Synonyms: Immotile Cilia Syndrome, ciliopathies

**Specimen Requirements**

Respiratory samples are procured by brushing or scraping the inferior nasal turbinate, avoiding bone fragments. Samples are fixed as soon as possible in appropriate EM fixative\*. Submit the brush/scraper intact, submerged in the EM fixative.

\*An appropriate EM Fixative contains 1.5 – 3.0% glutaraldehyde in a stable buffer (pH 6.8-7.3). It may also contain formalin up to 4%. Pieces should be no bigger than 3 mm in any one dimension. The fixative can be obtained free of charge by calling the UNMC Electron Microscopy Laboratory at 402-559-6420.