Primary Ciliary Dyskinesia (PCD) is a genetic disorder causing chronic oto-sino-pulmonary disease. No single diagnostic test will detect all PCD cases. Transmission electron microscopy (TEM) of respiratory cilia was previously considered the gold standard diagnostic test for PCD, looking for absence of the outer dynein arms, inner dynein arms, or central pair, but 30% of all PCD cases have either normal ciliary ultrastructure or subtle changes which are non-diagnostic. These cases are identified through alternate diagnostic tests, including nasal nitric oxide measurement, high speed videomicroscopy analysis, immunofluorescent staining of axonemal proteins, and/or mutation analysis of various PCD causing genes. Autosomal recessive mutations in DNAH11 and HYDIN produce normal TEM ciliary ultrastructure, while mutations in genes encoding for radial spoke head proteins result in some cross-sections with non-diagnostic alterations in the central apparatus interspersed with normal ciliary cross-sections. Mutations in nexin link and dynein regulatory complex genes lead to a collection of different ciliary ultrastructures; mutations in CCDC65, CCDC164, and GAS8 produce normal ciliary ultrastructure, while mutations in CCDC39 and CCDC40 cause absent inner dynein arms and microtubule disorganization in some ciliary cross-sections. Mutations in CCNO and MCIDAS cause near complete absence of respiratory cilia due to defects in generation of multiple cellular basal bodies; however, the scant cilia generated may have normal ultrastructure. Lastly, a syndromic form of PCD with retinal degeneration results in normal ciliary ultrastructure through mutations in the RPGR gene.

Wednesday, February 14, 2018
11:30 am
Strathcona Anatomy Building
3640 University Street
Room 2/45 (Anatomy Museum)

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