McGill UNIVERSITY

Department of Anatomy and Cell Biology

Hosted by Dr. Huy Bui

How to Diagnose Primary Ciliary Dyskinesia

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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.

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Strathcona Anatomy Building
3640 University Street
Room 2/45 (Anatomy Museum)

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