2014 USCAP Ghadially Award Winner

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Abstract: **Ciliary Inclusion Disease: Report of a New PCD Variant.** [JPG/2.1mb]

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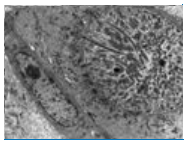
Background: Primary ciliary dyskinesia (PCD) is a rare, inherited disorder affecting cilia motility and thus impairing mucus clearance of the respiratory tract. With the exception of a single known mutation, all cilia dysfunction associated with PCD has been attributed to a variety of consistently observed structural abnormalities within individual cilia. While basal body migration abnormalities have been implicated in other ciliopathies, this mechanism has yet to be associated with congenital airway disease.



Results: Electron microscopic evaluation of each specimen showed that few cilia were present at the respiratory epithelial cell surface, however those that were observed displayed a normal ultrastructural appearance and axonemal architecture. Within many of the cells, including those deep within the epithelial cell layer, were large intracytoplasmic vesicular inclusions containing disorganized arrays of structurally normal cilia.

Conclusions: We have presented the ultrastructural findings of 5 patients in which the clinical history was highly suggestive of PCD; however, the derivation of cilia malfunction appears not the result of any recognized primary cause or secondary effect. Instead, the cases presented may illustrate a mechanism by which structurally normal appearing basal bodies and cilia are formed within the respiratory epithelial cells, but, due perhaps to some disruption in a membrane trafficking pathway, not anchored in their normal apical position. This new entity, termed Ciliary Inclusion Disease, represents a novel variant of PCD and should be considered when performing routine ultrastructural evaluation of ciliary biopsies.