

**Supplementary Table 2. Sequential whole exome filtering process for PCD-221 II:1**

<b>Sequentially listed filtering steps</b>	<b>Total variants</b>
Total variants	62,913
Variants in MAF<0.01 in 1000 Genomes Project	3,107
Homozygous variants	744
Homozygous nonsynonymous, splice-site, or insertion/deletion variants	76
Of these, any that appeared in less than 10/500 exomes from the UK10K_RARE cohort	8 ( <i>EEF1D</i> , <i>MYO1D</i> , <i>CXORF69</i> , <i>GPR112</i> , <i>EXPH5</i> , <i>PCK1</i> , <i>PMS1</i> , <i>ARMC4</i> )
Of these, any in genes in Cilia Proteome	3 ( <i>EEF1D</i> , <i>MYO1D</i> , <i>ARMC4</i> )
Of these, any in genes implicated in cilia motility*	1 ( <i>ARMC4</i> )

\* Identified in KM Lonergan *et al* (2006). Identification of novel lung genes in bronchial epithelium by serial analysis of gene expression. *Am J Respir Cell Mol Biol* 35 (6):651-661; and ET O'Toole *et al* (2012). Computer-assisted image analysis of human cilia and *Chlamydomonas* flagella reveals both similarities and differences in axoneme structure. *Cytoskeleton (Hoboken )* 69 (8):577-590.