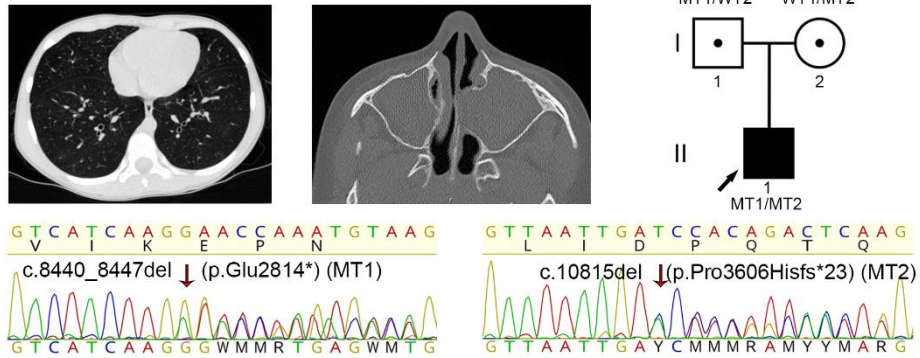
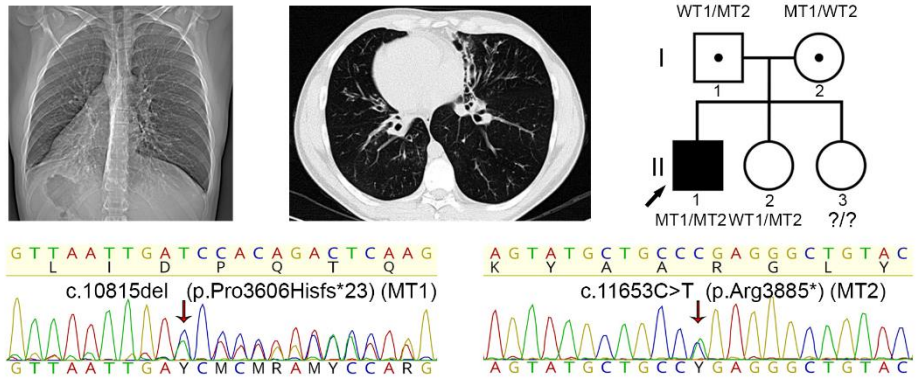


Additional file 4. Clinical and genetic analysis of PCD families with common truncating mutations in *DNAH5* and novel loss-of-function variants in *DNAH11*.

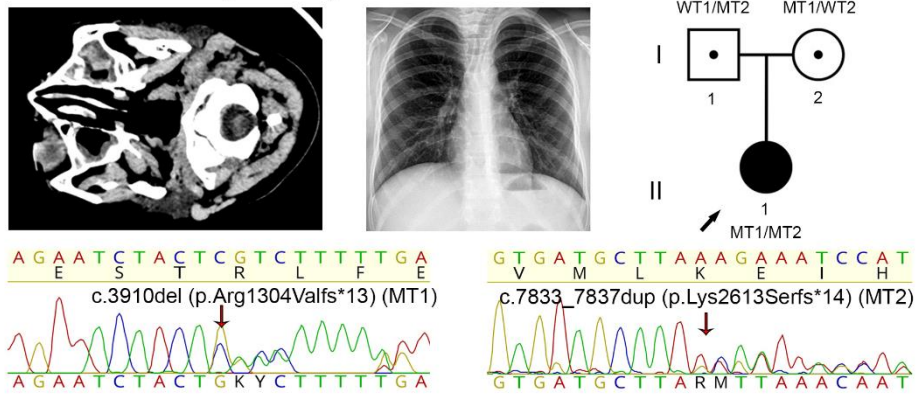
A Case PCD-#01 (*DNAH5*)



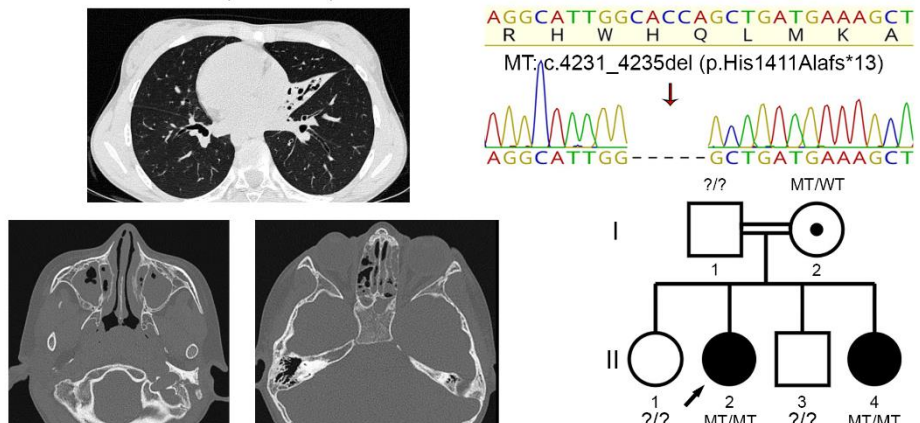
B Case PCD-#02 (*DNAH5*)



C Case PCD-#07 (*DNAH11*)



D Case PCD-#08 (*DNAH11*)



A Case PCD-#01. On the top panel: computed-tomography (CT) scans of the proband's lungs showing a tree-in-bud pattern (on the left) and paranasal sinuses demonstrating chronic hyperplastic rhinosinusitis (in the middle). On the lower panel: Sanger sequencing of the proband demonstrates compound heterozygosity for c.8440_8447del (MT1) and c.10815del (MT2) variants in *DNAH5*. **B** Case PCD-#02. On the top panel: CT scans of the proband showing situs inversus and bilateral interstitial changes in lungs, atelectasis with bronchiectatic disease (on the left, in the middle). On the lower panel: Sanger sequencing of the proband demonstrates compound heterozygosity for c.10815del (MT1) and c.11653C>T (MT2) variants in *DNAH5*. **C** Case PCD-#07. On the top panel: CT scans of the proband demonstrating maxilloethmoidal sinusitis, fibroatelectasis, and situs viscerum solitus (on the left, in the middle). On the lower panel: Sanger sequencing of the proband confirms compound heterozygosity for c.3910del (MT1) and c.7833_7837dup (MT2) variants in *DNAH11*. **D** Case PCD-#08. On the left, CT scans of the proband demonstrating bilateral bronchiectatic disease and total darkening of paranasal sinuses and ethmoid cells. On the top right, Sanger sequencing of the proband demonstrates homozygosity for c.4231_4235del (MT) variant in *DNAH11*. **A-D** On the right, the family pedigrees are shown.