

Supplemental table 1: Histopathological features of lung biopsy and explants.

| | Patient 12 (surgical biopsy) | Patient 3 (lung explant) | Patient 20 (lung explant) |
|--|---|------------------------------------|-------------------------------------|
| TBX4 mutation (nucleotide change) | c.(549+1_550-1)_(702+1_703-1)del | c.231G>A | c.143dup |
| Lung macroscopy | NA | Small size | Small size |
| Alveolar structure: Alveolar septa Emphysematous changes Interstitial fibrosis and inflammation | Thin alveolar septa Yes Patchy, peribronchiolar and perivascular | | |
| Bronchial anomalies | Peri-bronchial inflammation with cholesterol clefts and hypertrophy of smooth muscle | | |
| Vascular remodeling: - Small pulmonary arteries (<500µm) <i>Intimal and medial thickening</i> <i>Plexiform lesions</i> - Large (elastic-type pulmonary arteries) <i>Intimal thickening</i> <i>Organized thrombi</i> | Yes No | Yes Numerous | Yes Numerous |
| | NA | Mild No | Mild Yes |

NA: not available