

Supplementary material

TABLE S1: Lung histopathological analysis in patients with *SOX17* variants (n=8)

<i>Patient</i>	<i>I-1</i>	<i>I-2</i>	<i>II</i>	<i>VIII</i>	<i>X</i>	<i>XII-1</i>	<i>XIII</i>	<i>XVII</i>	<i>%</i>
Severe pulmonary arterial remodelling	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	8/8 (100%)
Congestive and/or haemorrhagic parenchyma	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	8/8 (100%)
Pleural and subpleural vessel dilation	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	8/8 (100%)
Dilated bronchial arteries*	Yes	Yes	Yes	No	Yes	No	No	Yes	5/8 (63%)

Main histological features included severe pulmonary artery remodelling, congestion or parenchymal haemorrhage, dilation of subpleural vessels and dilation of bronchial arteries. Pulmonary arteries were severely abnormal with major vascular wall thickening and plexiform lesions. Lung architecture was overall preserved, although congestion, parenchymal collapse (secondary to haemorrhagic infarction) as well as peripheral vessel dilation were constantly noticed in examining haematoxylin & eosin slides. Bronchial vessels also appeared altered, with dilation being observed in 5 out of 8 lung explants.

**Dilation of bronchial vessels could only be observed in central lung samples.*