This unique case of germline TBX4 variant highlights strikingly complex pulmonary histopathology leading to lethal cardiopulmonary failure. Over time and under the influence of abnormal Tbx4 signaling, all compartments of the lung continue to remodel, resulting in worsening PAH, impaired airway and distal airspace structure, and hypoxemia, which cause end stage lung disease and death. Studies focusing on signaling pathways downstream to Tbx4 are needed to intervene pulmonary remodeling and attenuate progressive PAH.

References