Idiopathic and/or familial pulmonary arterial hypertension

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Idiopathic and/or familial pulmonary arterial hypertension. ORPHA:422

Idiopathic and/or familial pulmonary arterial hypertension (IFPAH) is a form or pulmonary arterial hypertension (PAH, see his term) characterized by elevated pulmonary arterial resistance leading to right heart failure; it is progressive and potentially fatal. About 75% of heritable pulmonary arterial hypertension (HPAH, see this term) have an identified mutation. HPAH has been linked to mutations in BMPR2 in 75% of cases; other genes implicated in HPAH include ACVR1, BMPR1, CAV1, ENG and SMAD9 and CBLN2. (However, the majority of patients carrying an HPAH mutation do not develop PAH). Idiopathic pulmonary arterial hypertension (IFPAH; see this term) refers to those cases of pulmonary arterial hypertension in which etiology remains unknown .