DISEASE:
Heritable pulmonary arterial hypertension

NAME:	Heritable pulmonary arterial hypertension
DESCRIPTION:	Heritable pulmonary arterial hypertension (HPAH) is a form of pulmonary arterial hypertension (PAH, see this term), occurring due to mutations in PAH predisposing genes or in a familial context. HPAH is characterized by elevated pulmonary arterial resistance leading to right heart failure. HPAH is progressive and potentially fatal.
ORPHACODE:	275777
SYNONYMS:	FPAH Familial pulmonary arterial hypertension HPAH Hereditary pulmonary arterial hypertension
XREF(S):	Orphanet OMIM ICD-10 OMIM

ANALYTE(S):	ATP13A3 ACVRL1 BMPR2 TBX4 ENG CAV1 SMAD9 KCNK3 EIF2AK4
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RELATED CONTENT

Related Genetic Tests

- Arteriovenous malformation (gene panel)
- Primary Arterial Hypertension (gene panel)
- Pulmonary Arterial Hypertension / Rendu Osler Weber disease (gene panel - 24 genes)
- Rendu-Osler-Weber disease (4 genes)
- Trombosis - Hemostasis (gene panel)

Related Laboratories

- Centre de Génétique Humaine - Erasme ULB
- Centre de Génétique Médicale UCL
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- activin A receptor like type 1
- ATPase 13A3
- bone morphogenetic protein receptor type 2
- caveolin 1
- eukaryotic translation initiation factor 2 alpha kinase 4
- endoglin
- potassium two pore domain channel subfamily K member 3
- SMAD family member 9
- T-box transcription factor 4

Related Gene Panels

- Congenital heart disease (29 genes) - VUB
- Primary Arterial Hypertension (19 genes) - KUL
- Pulmonary Arterial Hypertension (24 genes) - ULB
- Rendu/Osler/weber (4 genes) - UCL
- Trombosis - Hemostasis (107 genes) - KUL
