

Supplement Table S4: Clinical characteristics of twelve patients with PAH carrying *ACVRL1* mutations

Variables	Patient *											
	1	2	3	4	5	6	7	8	9	10	11	12
Sex	M	F	M	F	M	F	M	M	F	F	M	F
Age at onset,(year)	4.8	13.5	10.4	2.9	2.6	28.8	4.6	36	10.6	27.5	4.6	8.4
Age at diagnosis(year)	4.9	15.8	10.6	3.2	3.8	29.5	5.1	36.5	11.1	27.6	6.6	8.9
Age at death(year)	-	17.4	-	-	5.8	31.8	-	-	-	27.6	9.2	-
Known features of PH												
Family history of PH	-	+	-	+	-	+	-	-	-	-	-	-
Symptoms at onset												
Breathlessness	+	+	-	+	-	-	+	+	-	+	-	-
Fatigue	+	+	+	+	+	+	+	+	+	+	+	+
Syncope	+	-	+	+	-	+	-	-	+	-	-	+
Chest pain	-	-	-	-	-	-	+	-	-	-	-	-
NYHA FC	III	III	III	III	II	II	IV	III	III	IV	III	II
BNP(pg/ml)	939	681	975.57	1248	698	478	5000	93	469	432	788.3	658
SPAP (cardiac echocardiography) mmHg	96	15	193	71	96	109	93	75	86	74	134	46
RHC parameters												
MPAP(mmHg)	95	68	70	Na	78	58	Na	50	82	Na	78	52
RAP(mmHg)	8	3	14	Na	17	9	Na	2	13	Na	11	7
PCWP(mmHg)	12	Na	9	Na	Na	6	Na	9	2	Na	15	12
PVR(wood units)	41.92	12.62	15.68	Na	25	12.99	Na	10.63	20.17	Na	23.4	9.26
CI(l/min/m ²)	2.87	3.48	2.8	Na	2.8	2.75	Na	1.95	2.4	Na	2.6	4.45
SvO ₂ (%)	61	75	66.65	Na	72	71	Na	68.5	54.7	Na	70	74
AVT (+, -)	-	-	-	Na	-	-	Na	-	-	Na	-	-

NYHA FC, New York Heart Association Function Class; CI, cardiac index, normal value: 2.6–4 l/min per m²; BNP, brain natriuretic peptide; PCWP, pulmonary capillary wedge pressure, normal value ≤15 mmHg; mPAP, mean pulmonary arterial pressure, normal range 10–20 mmHg; PVR, pulmonary vascular resistance, normal value 0.25–1.6 Wood units; SPAP, Systolic pulmonary arterial pressure, normal value <30 mmHg; Na, Data unavailable; AVT, acute pulmonary vasodilatation test; SvO₂, mixed venous oxygen saturation; *Plus (+) signs indicate the presence of a characteristic, and minus (-) signs indicate the absence of a characteristic and negative;