

Table S1 Genomic information of the seven PFV associated genes

Gene	Cytogenetic locus	gDNA	mRNA	Protein
<i>NFKB1</i>	4q24	NC_000004.11	NM_003998	NP_003989
<i>VEGFA</i>	6p21.1	NC_000006.11	NM_001025366	NP_001020537
<i>EDN1</i>	6p24.1	NC_000006.11	NM_001955	NP_001946
<i>CDKN2A</i>	9p21.3	NC_000009.11	NM_000077	NP_000068
<i>ATOH7</i>	10q21.3	NC_000010.10	NM_145178	NP_660161
<i>TGFB1</i>	19q13.2	NC_000019.9	NM_000660	NP_000651
<i>NDP</i>	Xp11.3	NC_000023.10	NM_000266	NP_000257

The human genome information was based on NCBI build 37/hg19. PFV, persistent fetal vasculature.

Table S2 The list of the SNPs and the haplotype analysis of the probands in family 1, 2 and 4

#Chromosome	Position	Intermarker distance (bp)	SNP ID	Haplotype of the mutant <i>LIM2</i> allele		
				F1-proband	F2-proband	F4-proband
19	51,608,074	–	rs2691249	C	C	A
19	51,689,700	81,626	rs12461542	T	C*	C*
19	51,837,332	147,632	rs10418551	G*	G*	G*
19	51,883,831	46,499	Mutation	A*	A*	A*
19	52,032,964	149,133	rs3976745	G*	G*	C
19	52,095,980	63,016	rs3794984	T*	T*	G
19	52,130,337	34,357	rs8105105	C	T	T
Distance of both ends of the shared haplotypes (bp)				258,648	406,280	194,131

* in haplotypes were the haplotypes shared between the three families. The human genome information of SNPs was based on UCSC NCBI build 37/hg19. SNP, single nucleotide polymorphism.



Figure S1 The geographical positions of families 1, 2, 3 and 4 are shown in the map of China. The position of family 1 was in Urumqi, Xinjiang province. The position of family 2 was in Ji'an, Jiangxi Province. The position of family 3 was in Changning, Hunan province. The position of family 4 was in Ganzhou, Jiangxi province. The geographical distance between family 1 and families 2&4 is more than 3,600 kilometers. The geographical distance between family 3 and families 2&4 is around 400 kilometers. The geographical distance between family 2 and family 4 is around 200 kilometers. The map of China was download from the China Standard Map Service Website (<http://bzdt.ch.mnr.gov.cn/>) with the map approval number GS(2019)1696.