Figure S4. Locations of rare deleterious PAH patient-derived other previously reported PAH risk gene variants within the two-dimensional protein structures. Predicted damaging missense (D-Mis) variants are shown above the protein schematics; likely-gene-disrupting (LGD including stopgain, frameshift, in-frame deletion and whole exon deletion) variants are shown below the schematics. The vertical gray lines indicate exon borders. For ACVRL1, E10 (1) indicates a deletion of exon 10 identified in one case. For EIF2AK4, patients 12-064 and 21-036 are homozygous for the given EIF2AK4 variants, and patients 12-014, 02-030 and 10-091 are compound heterozygotes.