

**Figure S3. Locations of rare deleterious PAH patient-derived *BM***

***PR2***

**variants within the two-dimensional protein structure.** Predicted damaging missense (D-Mis) variants are shown above the protein schematic; likely-gene-disrupting (LGD including stopgain, frameshift, in-frame deletion and whole exon deletion) variants are below the schematic. The vertical gray lines indicate exon borders. E1(2) indicates a deletion of exon 1 identified in two cases; similar designations are used for the other whole exon deletions.

**BM<sub>PR</sub>2**

