**Supplementary Data**

## Molecular Genetics and Clinical Features of Chinese IPAH and HPAH Patients

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**Table 1. Oligonucleotides for PCR and sequencing of the *BMPR2* gene**

| Exon | Sense primer | Antisense primer | Annealing (°C) |
|---|---|---|---|
| 1 | 5'-CTTCGTCCTTCCCGGCAGT-3' | 5'-TGGCGAAGGGCAAGCACA-3' | 63 |
| 2 | 5'-CAGTCATTTCAGGTAAGGAAAGT-3' | 5'-TGAACAGAAGAACGTCATTGAA-3' | 56 |
| 3 | 5'-TTGCAAAACTGTTTCATAGCTTA-3' | 5'-GCTGGCATTACAGGCATGA-3' | 56 |
| 4 | 5'-CATTTCCTTTGATGCAAAAACA-3' | 5'-TTGAGGCTGGGTGTATTTTG-3' | 59 |
| 5 | 5'-CTCCCAGAATTTGGCTTTCAT-3' | 5'-AAATAGAAGCCCCAGGAGACA-3' | 59 |
| 6 | 5'-TGGGTCTGGTAGGAGCTTCAT-3' | 5'-TGGCCTCAAGTGATCCACCT-3' | 59 |
| 7 | 5'-CCATCCCTTCCTCTCCTCTCT-3' | 5'-TGTGAATTTTGAACCCACATGA-3' | 59 |
| 8 | 5'-GAGTTGAAATTCCGATTTCTCT-3' | 5'-ACCAAAGTGCTGGGATTACA-3' | 55 |
| 9 | 5'-AATTTGCATCCTGCTGCTAA-3' | 5'-TTAATGACATGGTTAGGGTCAA-3' | 57 |
| 10 | 5'-TTTGTGGCATTAGGCAACTC-3' | 5'-ATGTGCCTGAAGGGGATGAA-3' | 57 |
| 11 | 5'-CCGTAATCCTTGAAGCCTAA-3' | 5'-CATTGAACTATTAGGCTGGTTTA-3' | 56 |
| 12-1 | 5'-GTGGGTAAAGCAAGCTAGAAC-3' | 5'-ATGTACGTTTGGAAGAAAATGA-3' | 55 |
| 12-2 | 5'-ATGGCCAGCTTGTTGCTCT-3' | 5'-AGCTAGACCCAAAAGAAGTTGA-3' | 58 |
| 12-3 | 5'-ATGTTGGCCAGGTTGGTCT-3' | 5'-CTATCTGGCCAAACAACCAA-3' | 58 |
| 13 | 5'-TTAGCGAGACTAAACAAAAGTGC-3' | 5'-TTTTCCTGGAAAAACATTGTCT-3' | 57 |

**Table 2. The damaging effect of the splice-site mutations predicted by the software**

| Patient ID | Mutation Location | Variant* | Splice Site Finder | MaxEnt | Nnsplice | Genesplicer | Comments |
|---|---|---|---|---|---|---|---|
| 498 | intron 7 | c.967+5G>C | Primary donor splice site slightly decreased (–13.9%) | Primary donor splice site slightly decreased (–19.5%) Primary acceptor splice site slightly decreased (–9.8%) | Primary donor splice site slightly decreased (–15.6%) | Primary donor splice- site destroyed | Possible effect on splicing |
| 370 | exon8 | c.969T>C | No effect predicted | | No effect predicted | Primary acceptor splice site destroyed | Possible effect on splicing |

* Abbreviations are in accord with nomenclature guidelines as recommended by the Human Genome Variation Society (http://www.hgvs.org/mutnomen/). The letter c. indicates coding DNA, where nucleotide 1 is the A of the ATG translation initiation codon.

**Table 3. Polymorphisms in the coding region found in the general population**

| Location | Domain | Nucleotide Change* | Amino Acid Change† | Allele Frequency |
|---|---|---|---|---|
| 5'UTR | | c.-93A>G | | 0.84% of G allele (n=4 of 474 chromosomes) in the Chinese population in this study |
| exon2 | ECD | c.165T>C | p.Asn55Asn | 0.14% of C allele (n=1 of 710 chromosomes) in the Chinese population in this study |
| exon12 | CD | c.2379A>C | p.Thr793Thr | 0.14% of C allele (n=1 of 710 chromosomes) in the Chinese population in this study |
| exon12 | CD | c.2811G>A | p.Arg937Arg | 7.04% of A allele (n=50 of 710 chromosomes) in the Chinese population in this study |

* Abbreviations are in agreement with nomenclature guidelines as recommended by the Human Genome Variation Society (http://www.hgvs.org/mutnomen/). The letter c. indicates the numbering of the base change, where nucleotide 1 is the A of the ATG translation initiation codon.

† The letter p. is used to indicate the change at the protein level.

UTR = untranslated region; ECD = extracellular domain; CD = cytoplasmic domain.