

Appendix B

Table S1 Primer sequences for real-time PCR for confirmation of CNV regions

| | Primer name | Orientation | Primer sequence (5' to 3') |
|-----------|--------------------|-------------|----------------------------|
| Dup-CNV-1 | <i>EDN3</i> | F | GGTTGGAGTGACCAATGGAT |
| | <i>EDN3</i> | R | GGGTGGATGTTCGCATTGAG |
| | <i>C20orf174-1</i> | F | GTGCTGTTCAAGTACACAACCA |
| | <i>C20orf174-1</i> | R | TTGTCTGTGCCTCCTCTGGAA |
| | <i>ATP5e</i> | F | GCGTTGAAAGGTTCTGAAGTTG |
| | <i>ATP5e</i> | R | AGCGTGTCTCTCCCACAGCTA |
| | <i>TBB6</i> | F | TGTGGTCAGGAACGAGTGTGA |
| | <i>TBB6</i> | R | GCGAATGGATGAGCTGGAAT |
| Dup-CNV-2 | <i>CNV-2</i> | F | TGCTTCCCCATTAGTGTGTTTCT |
| | <i>CNV-2</i> | R | CAACAGGCTTTGGATGAGTATAATCA |
| Reference | <i>PCCA</i> | F | CAGACACACAGAGCCCATCTCT |
| | <i>PCCA</i> | R | TGGAGCAGTGGTGGCTGTT |

Table S2 Primer sequences for pyrosequencing

| Primer name | Orientation | Primer sequence (5' to 3') |
|-------------|-------------|----------------------------------|
| rs16172722 | F | GGGGGAAAAAGAATTTCTGTGT |
| rs16172722 | R | -biotin-CATGCAGGATCTCTATGAAGAAAA |
| rs16172722 | S | GTATTCTAGAAGCCATTCC |
| rs16172768 | F | ATCATAAACAAACAGCCAAATCAA |
| rs16172768 | R | -biotin-TTCGATAGAGGAAGCACGTAACAT |
| rs16172768 | S | GCCAAATCAAAACAGATG |

Table S3 Primer sequences for PCR to determine the boundaries

| Primer name | Orientation | Primer sequence (5' to 3') |
|--------------------|-------------|----------------------------|
| Dup1-3' & Dup2-3' | F | CCTCCTGTTGGGTTCTGACTG |
| Dup-1-3' & Dup2-3' | R | TGTGGATGGAGGATGGTGAGGC |
| Dup-1-5' & Dup2-5' | F | AGGAAGCAAGTTGTAAAGGGA |
| Dup-1-5' & Dup2-5' | R | ATCATTTCTTCAATTAACACCCAC |

Table S4 Primer sequences for mRNA qPCR assays

| Gene | Orientation | Chromosome | Primer sequence (5' to 3') |
|---------------|-------------|------------|--------------------------------|
| <i>EDN3</i> | F | 20 | CACACGCTCGCTCAGTAGCA |
| <i>EDN3</i> | R | 20 | TTCAGCAAGCAAACGTTAAACC |
| <i>SLMO2</i> | F | 20 | GGGAATCCCATCCATTGTGA |
| <i>SLMO2</i> | R | 20 | CCTGGACATACGTCTTCGTTCTG |
| <i>ATP5e</i> | F | 20 | CAGGCCGGACTCAGCTACAT |
| <i>ATP5e</i> | R | 20 | ATCGCCGCTCTGACAACCT |
| <i>TBB6</i> | F | 20 | CCCACTGTTTCGACCCTGACA |
| <i>TBB6</i> | R | 20 | CCCTTAGCCCAGTTGTTTCCA |
| <i>EDNRB</i> | F | 1 | TCTCAGAATCTGCCTGCTTCAC |
| <i>EDNRB</i> | R | 1 | CTTTAGCTTTCTTGTA AACATCATAAAGG |
| <i>EDNRB2</i> | F | 4 | TGAGCAAGAGAAATGGCATGA |
| <i>EDNRB2</i> | R | 4 | CCTCCCGACGCCGTTT |
| <i>TYRP2</i> | F | 1 | GTGGATGACCGGGAAAGGT |
| <i>TYRP2</i> | R | 1 | CAAAGTTTCCTGTGCACCAACA |
| <i>GAPDH</i> | F | 1 | TGACAAGTCCCTGAAAATTGTCA |
| <i>GAPDH</i> | R | 1 | AAGGGTGCCAGGCAGTTG |