

Table S5. Primers used for CNVs molecular characterizations

| Designation | | Primer sequence (5'--> 3') | Primer localization * | Amplicon size | TA | Application |
|-----------------------|-----|------------------------------|--------------------------|---------------|--------|-----------------------------|
| <i>PT4_probe_ex12</i> | Fw | CAAGTATGACCGCATGAAGG | chr16:89337224-89337243 | 78 bp | 60°C | qPCR analysis |
| | Rev | ATCACAGAACTGGGCAGCA | chr16: 89337166-89337185 | | | |
| <i>PT14_probe1</i> | Fw | CCGCCTGCTCTCATTTCA | chr16:89555954-89555971 | 69 bp | 60°C | qPCR analysis |
| | Rev | CCGTGCTGGTCGTAGTTGTT | chr16:89556003-89556022 | | | |
| <i>PT14_probe2</i> | Fw | AGCGCCGTGAGTTAGTGC | chr16:89557095-89557112 | 67 bp | 60°C | qPCR analysis |
| | Rev | GACCGCCAGAGAGGCTGT | chr16: 89557144-89557161 | | | |
| <i>PT14_probe3</i> | Fw | AGGTGAACAAAATCGGTGCT | chr16:89557856-9557875 | 60 bp | 60°C | qPCR analysis |
| | Rev | AGTCCCAGTAACATGCAGAA | chr16:89557895-89557915 | | | |
| <i>PT14_probe4</i> | Fw | GAGCGCCTGATAAAAACAGC | chr16:89558644-89558663 | 91 bp | 60°C | qPCR analysis |
| | Rev | CTTCGTCCAGTGAGGCAAAT | chr16:89558715-89558734 | | | |
| <i>PT4_bkp_pcr</i> | Fw | TTAGAAATAGTGAGGCTGTTGTGAGGTG | chr16:89300930-89300957 | 39342 bp | 63.5°C | Bkps junction amplification |
| | Rev | CACTCTGAGCAGTGCACTAGGCTTT | chr16:89340247-89340271 | | | |
| <i>PT14_bkp_pcr</i> | Fw | <i>PT14_probe1_Fw</i> | | 2781 bp | 60°C | Bkps junction amplification |
| | Rev | <i>PT14_probe4_Rev</i> | | | | |

Fw, forward; Rev, reverse; TA, annealing temperatur

* Primer physical localization is based on GRCh37/hg19 human genome assembly.