

**Table S7. Reference genotype replacement rule in a common deletion region.**

| Copy<br>Number |   | Original data |    |    |
|----------------|---|---------------|----|----|
|                |   | AA            | AB | BB |
| 0              | → | OO            | OO | OO |
| 1              | → | AO            | ?? | BO |
| 2              | → | BB            | AB | BB |

We assume there exist two normal alleles "A" and "B" along with the deletion allele "O" at the SNP locus within a copy number deletion region. Missing allele is indicated by "?".