|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
|  |  |  | rs1164325688 | rs4915 | rs1454173648 | rs1554081948 |
| sample | MLPA result | proportion GDV |  VAF GDV  | c.462A>G p.Gln154Gln | c.835-44G>A | c.84C>T p.Ser28= |
| 1 | SMN1 ex7 CN0 SMN2 ex7 CN3 | 1 | 1.0 | no | 1.0 | no |
| 2 | SMN1 ex7 CN0 SMN2 ex7 CN4 | 1 | 0.998 | 0.998 | 0.998 | no |
| 3 | SMN1 ex7 CN2 SMN2 ex7 CN2 | 1/2 | 0.459 | 0.430 | 0.454 | 0.281 |
| 4 | SMN1 ex7 CN1 SMN2 ex7 CN2 | 2/3 | 0.631 | no | 0.610 | no |
| 5 | SMN1 ex7 CN3 SMN2 ex7 CN1  | 1/4 | 0.230 | 0.995 | 0.250 | no |
| 6 | SMN1 ex7 CN1 SMN2 ex7 CN3  | 3/4 | 0.740 | 0.50 | 0.763 | no |
| 7 | SMN1 ex7 CN2 SMN2 ex7 CN3  | 3/5 | 0.585 | 0.393 | 0.573 | no |
| 8, HG002 NA24385 | SMN1 ex7 CN2 SMN2 ex7 CN2 | 1/2 | 0.410 | 0.986 | 0.357 | 0.191 |