**Supplemental Table S2: Relative dosage values displayed per-exon.**

|  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | **Exon number** (genomic coordinates) | | | | | | | | | | |
| **Sample** | **2**  (48573417-48573665) | **3**  (48575056-48575230) | **4**  (48575665-48575694) | **5**  (48581151-48581363) | **6**  (48584495-48584614) | **7**  (48584710-48584826) | **8**  (48586236-48586286) | **9**  (48591793-48591976) | **10**  (48593389-48593557) | **11**  (48603008-48603146) | **12**  (48604626-48604837) |
| 1 | 1.48 | 1.51 | **1.06** | 1.46 | 1.54 | 1.48 | **1.34** | 1.41 | **1.38** | **1.38** | 1.45 |
| 2 | 1.43 | 1.50 | **1.02** | 1.50 | 1.50 | 1.50 | **1.24** | 1.49 | 1.49 | 1.58 | 1.45 |
| 3 | 1.52 | 1.61 | **1.08** | 1.63 | 1.55 | 1.51 | **1.32** | 1.56 | 1.67 | 1.55 | 1.40 |
| 4 | 1.47 | 1.45 | **1.01** | 1.53 | 1.52 | 1.54 | **1.29** | 1.53 | 1.52 | 1.53 | 1.42 |
| 5 | 1.51 | 1.51 | **0.99** | 1.57 | 1.50 | 1.62 | **1.25** | 1.49 | 1.62 | 1.67 | 1.41 |
| Average | 1.48 | 1.52 | **1.03** | 1.54 | 1.52 | 1.53 | **1.29** | 1.50 | 1.54 | 1.54 | 1.43 |

Chr.: Chromosome. Exon 1 contains 5’UTR sequence and was not targeted with hybridisation capture probes. Exon numbering is in accordance to transcript NM\_005359.5. Values < 1.40 are in bold. Genomic coordinates are provided for coding nucleotides and are described according to human genome build hg19.