|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Tool | Intronic | Exonic | 1KB upstream | 10KB upstream | Total |
| Retroseq | 119 (45%) | 7 (3%) | 4 (2%) | 65 (24%) | 266 |
| Retroseq+ | 11 (35%) | 0 | 0 | 5 (16%) | 31 |
| Melt | 43 (45%) | 1 (1%) | 3 (3%) | 23 (24%) | 95 |
| Steak | 109 (29%) | 11 (3%) | 4 (1%) | 121 (32%) | 382 |
| Ervcaller | 15 (44%) | 0 | 1 (3%) | 7 (21%) | 34 |
| Mobster | NA | NA | NA | NA | NA |

Supplementary table 1.

Gene functional annotation of non-reference HERV-K insertions detected in the 50 whole genome sequencing samples.

The proportion of intronic HERV-Ks detected by each tool varied from 29% to 45%. Previous estimates of reference HERV-Ks (multiple subfamilies) suggest between 20% and 48% are expected to be intragenic [1,2,3,4,5]. The expected proportion of exonic HERV-Ks was reported to be between 0% and 5% in literature [2,3,4] which is consistent to our results. though one paper has reported exonic HERV-Ks (HML9) at a rate of 26% [5].

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