Conserved Ancestral Repeats

Although interspersed repeats are usually assumed to be non-functional, there are known cases where the insertion of repetitive sequences results in the formation of a functional regulatory element (Makalowski 1995). We thus examined the rare instances where an MCS overlaps an AR identified by RepeatMasker (A.F.A. Smit and P. Green, unpublished; http://repeatmasker.genome.washington.edu); in six cases, this overlap is longer than 50 bp (Table 3). While some of these regions may be erroneous RepeatMasker predictions, most show a fairly high levels of sequence identity (>68%) to the consensus for that repeat family. Interestingly, five of the six regions are tightly clustered in two locations, in one case just 1 kb upstream of an alternatively spliced exon of *ST7*.

Table 4. Ancestral repeats overlapping detected MCSs.

MCS Position[a]	AR Family	Identity Relative to Consensus[b]	Location
994407-994484	SINE MIR3	78.20%	7 kb upstream of *ST7* exon 8
994487-994603	LINE L2	68.20%	7 kb upstream of *ST7* exon 8
961929-962042	LINE L3b	71.20%	400 bp downstream of *ST7* exon 6, in an alternatively spliced intron
962047-962151	LINE L3	73.30%	500 bp downstream of *ST7* exon 6, in an alternatively spliced intron
962280-962946	LINE L3	69.00%	800 bp downstream of *ST7* exon 6, in an alternatively spliced intron
501569-502046	LINE L1M2	69.80%	Upstream of and overlapping with *MET* 5' UTR.

[a]Coordinates within the human reference sequence of the targeted region, as displayed in the customized UCSC Genome Browser (see http://genome.ucsc.edu).
[b]Percentage identity between the MCS sequence overlapping an AR and the consensus for that AR family.

References

Makalowski, W. 1995. Sines as a genomic scrapyard: an essay on genomic evolution. In *The impact of short interspersed elements (SINEs) on the host genome* (ed. R.J. Maraia), pp. 81-104. R.G. Landes Co., Austin.