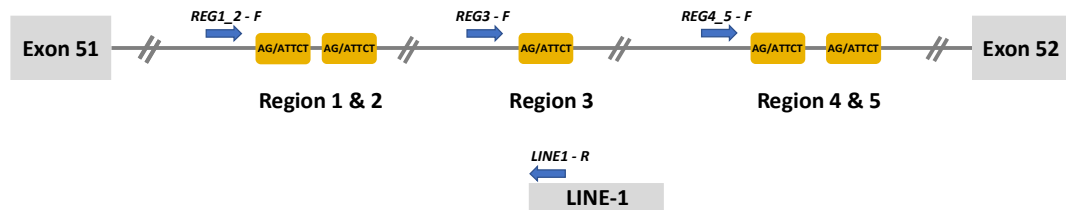


## Data S1. Additional material and methods for the LINE-1 characterization.

### I. Primers used to identify the LINE-1 5' insertion site in *DMD*.

a) Schematic showing primers' location:



b) Primers sequences:

REG1\_2-F: TCAAGCAAACTGATCCAGG

REG3-F: GCAGTCATGGCCACAAAAG

REG4\_5-F: ACAAATCTATGGAGGCACAGG

LINE1-R: TGGAAATGCAGAAATCACCC

### II. Sequences of custom probes used for isolating and enriching target region by hybridization capture of DNA libraries for (PacBio) single molecule real time sequencing.

> Probe nr1 (NM\_004006.2:c.7542+8207\_7542+8516)

AGATGAGGCAGTGATCACACATCACTATTAGTAAAAGTGTTCCTGTACCTGTAT  
CCACACTTTTATGTATATGGTTACTTATGTTAAAGTGATACATATTATATAAAAT  
TAACGTATACATTAAGTAGATATTTTAATAGTCTGTAATTAAATACTACTAGTAT  
TTTCTTTCTCCTTCAAGTGCTTACTTTTGATACCTCGAGTTACAGTGTCATAAAG  
ATTCTTTAGAAATATATTGACTGTCTTTTAAGAGCTTTTGATACAATACTGAGTTT  
ACATTCATCTGTTATTTATTGAACACTTGCTGGTGAAAGGCATC

Primers: F: AGATGAGGCAGTGATCACACA R: GATGCCTTTCACCAGCAAGT

> Probe nr2 (NM\_004006.2:c.7542+9241\_7542+9589)

```
AATGTAACCCCTCCCCCATATCAAGTTAATCTATGTTCAACTCCAGAATTATTTT
TGAACACTCAAACCTAGAAATTAATAAATAATCCATGAAGACGATTTTGTG
CCAAAAGCATATAGATAAATTGAGTTGATTCTATACTTAAGAAAGTGGAGAGG
AGAGAGTAATTTGGAGAGAGTAATTTACTCTTAATCCCATATTTTTTCCCTAAAT
GTGAAAGAAGTAGATTGTAGTGAGAGGGAAAATAACCTGTAGCAACTTCATTG
AGGCTAAGCTTTCTGTCATGTTATATTATACGAAAGTAATGAAATGCTTCCACA
GATAGAATCAGAAGTCCCCTCTGAGA
```

PRIMERS: F: AATGTAACCCCTCCCCCATA R: TCTCAGAGGGGACTTCTGATTC

### III. Custom bioinformatic pipeline for single molecule real-time sequencing analysis

- a) FASTA/Q files were mapped against a LINE-1 reference sequence (ID: L1HS from <http://www.girinst.org/replib/>) using BLASR alignment software (“-clipping soft -minMatch 7 -minPctSimilarity 90”) (Chaisson et al., 2012).
- b) BAM files were generated from SAM files with Samtools (v0.1.19; “view -b -S”) (Li et al., 2009).
- c) Consensus sequence was called from BAM files using Samtools mpileup command (“-uf” option), bcftools (“view -cg” option) and vcfutils.pl (vcf2fq command).

#### References:

Chaisson MJ, Tesler G. Mapping single molecule sequencing reads using basic local alignment with successive refinement (BLASR): application and theory. BMC Bioinformatics. 2012;13:238.

Li H, Handsaker B, Wysoker A, et al. The Sequence Alignment/Map format and SAMtools. Bioinformatics. 2009;25:2078-2079.