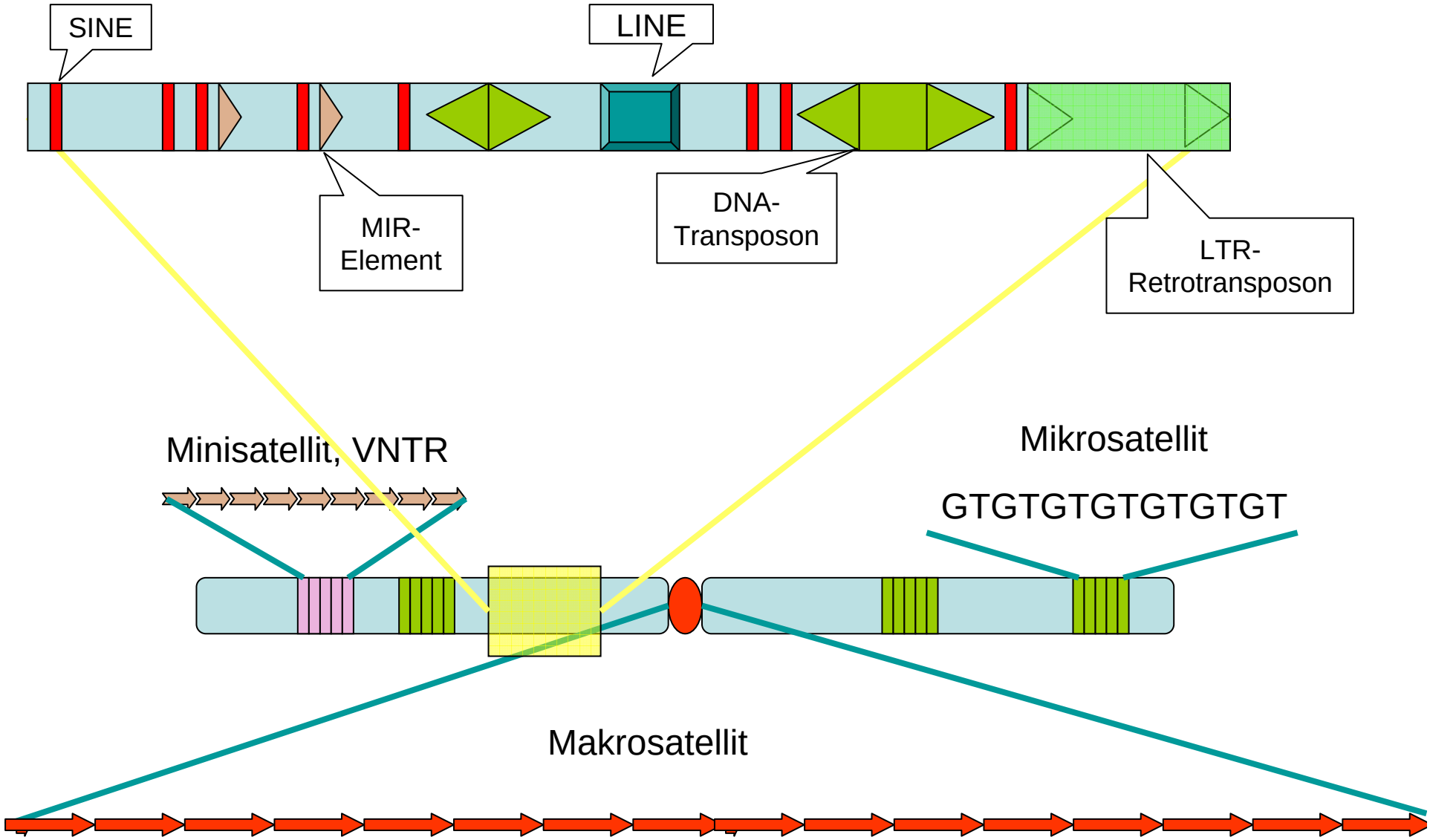
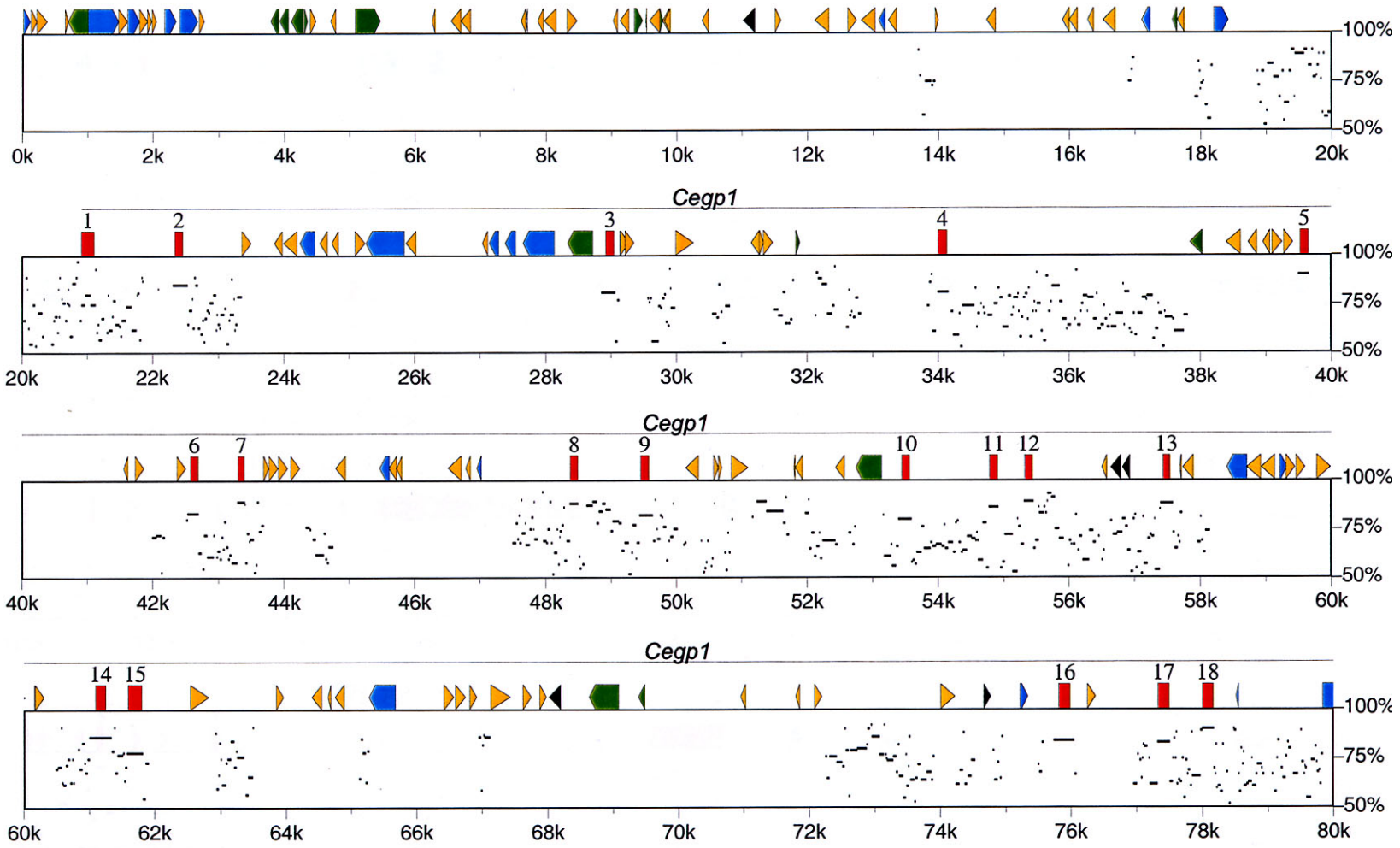
Satelliten-DNAs sind nur ein kleiner Teil
der repetitiven DNA der Eukaryoten,
Interspergierte repetitive DNA-Elemente:

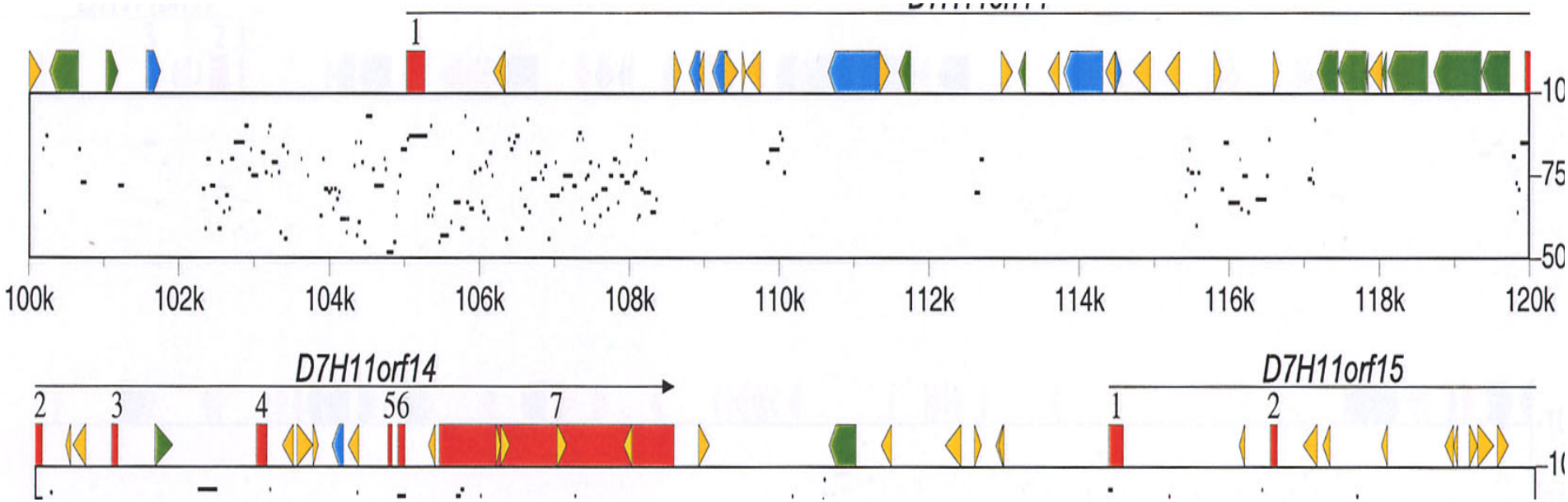


So sieht ein Chromosomenabschnitt in Wirklichkeit aus (Chromosom 11p15.3)



Alle Pfeile/Pfeilspitzen sind interspergiert repetitive Elemente; kleine rote Rechtecke = Exons

Interspergierte Repetitive Elemente können sogar in Exons integriert sein



Ausschnitt aus Humanchromosom 11, Pfeile/Pfeilspitzen = repetitive Elemente; rote Rechtecke = Exons

Interspergierte Repetitive Elemente

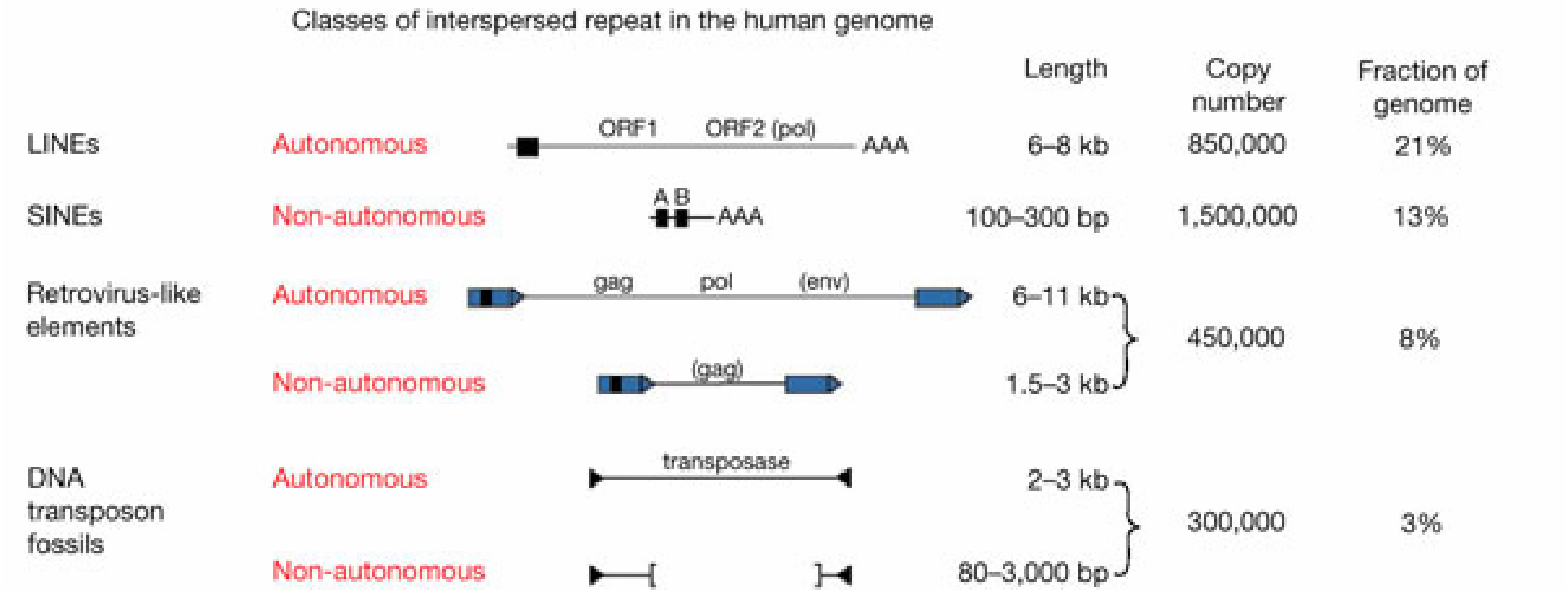
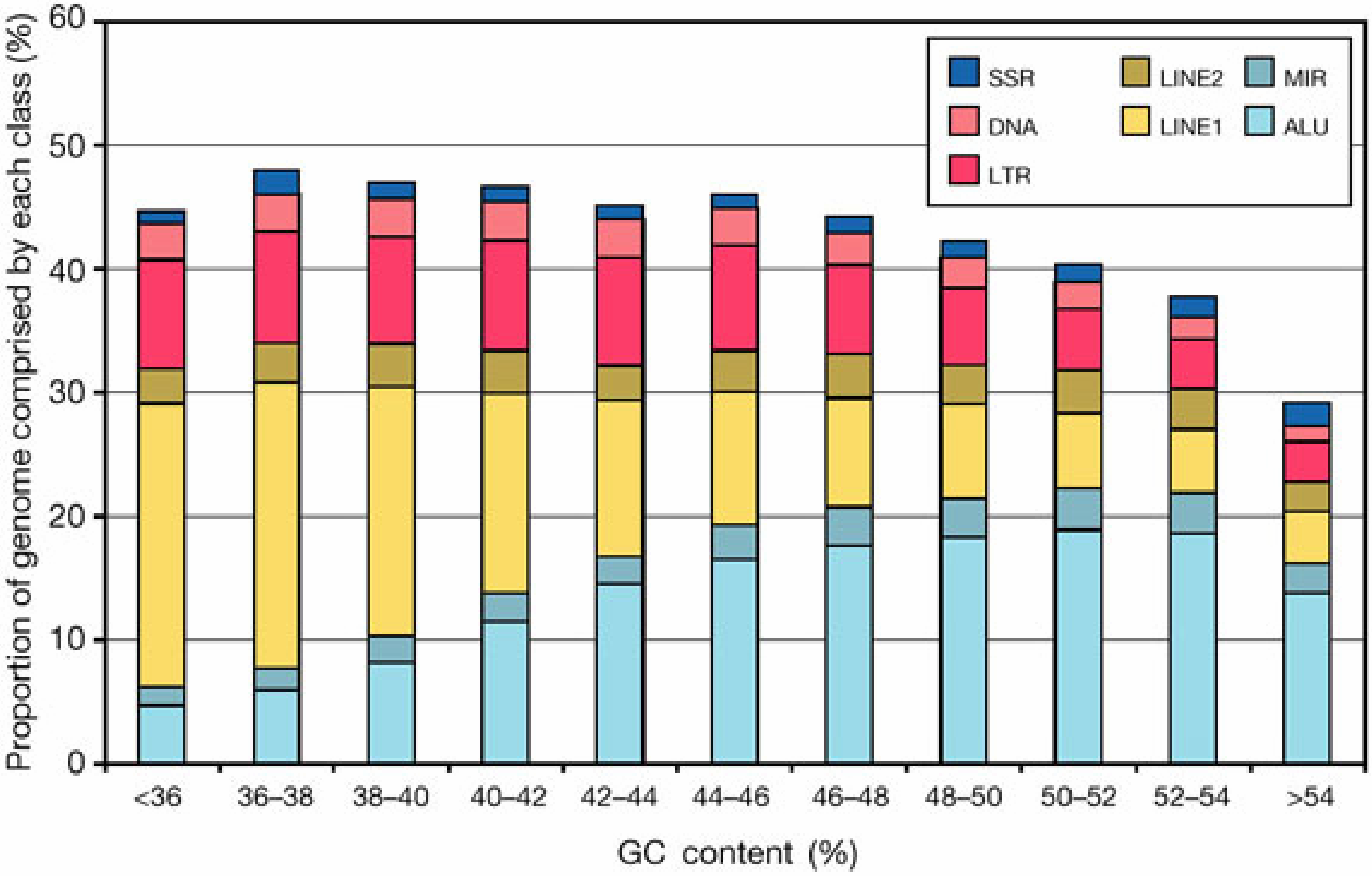


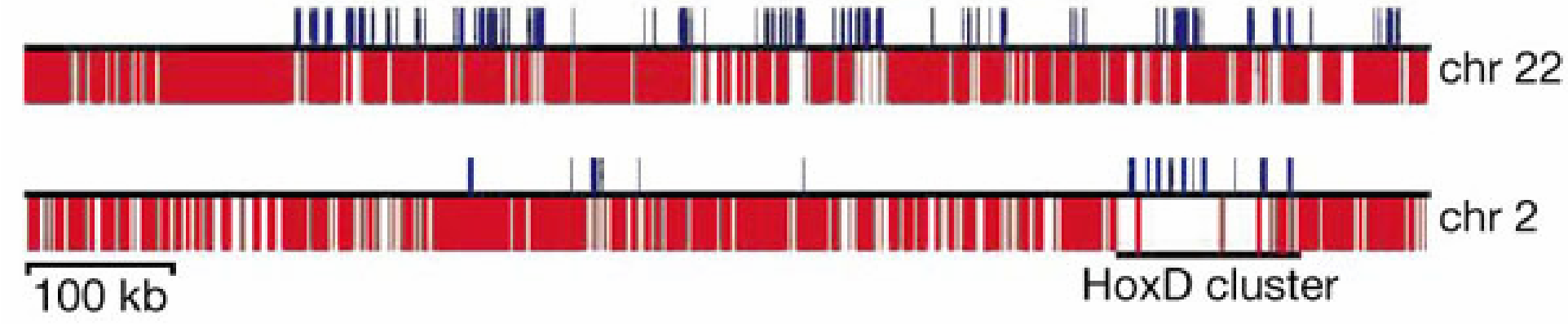
FIGURE 17. Almost all transposable elements in mammals fall into one of four classes.

Interspergierte Repetitive Elemente

- SINEs = short interspersed nuclear elements
- LINES = long interspersed nuclear elements
- MIR = mammalian wide interspersed repeats (Säugerspezifisch?)
- DNA-Transposons
- Retroelemente
- Pro-/Retroviren



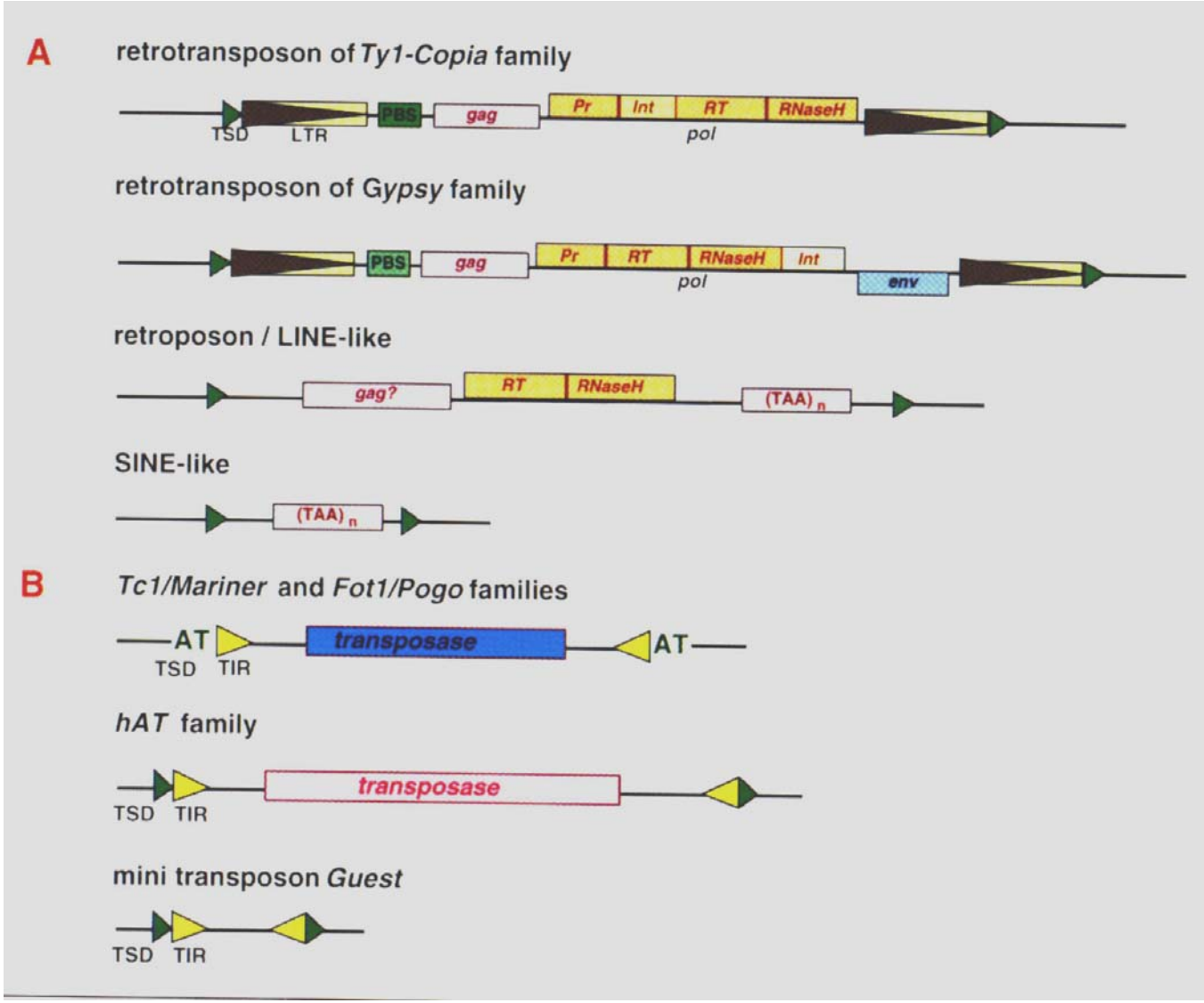
Nature **409**, 860-921 (15 February 2001)



Red bars, interspersed repeats; blue bars, exons of known genes. Note the deficit of repeats in the HoxD cluster, which contains a collection of genes with complex, interrelated regulation.

Nature **409**, 860-921 (15 February 2001)

SINEs, LINEs, Retroelemente, DNA-Transposons

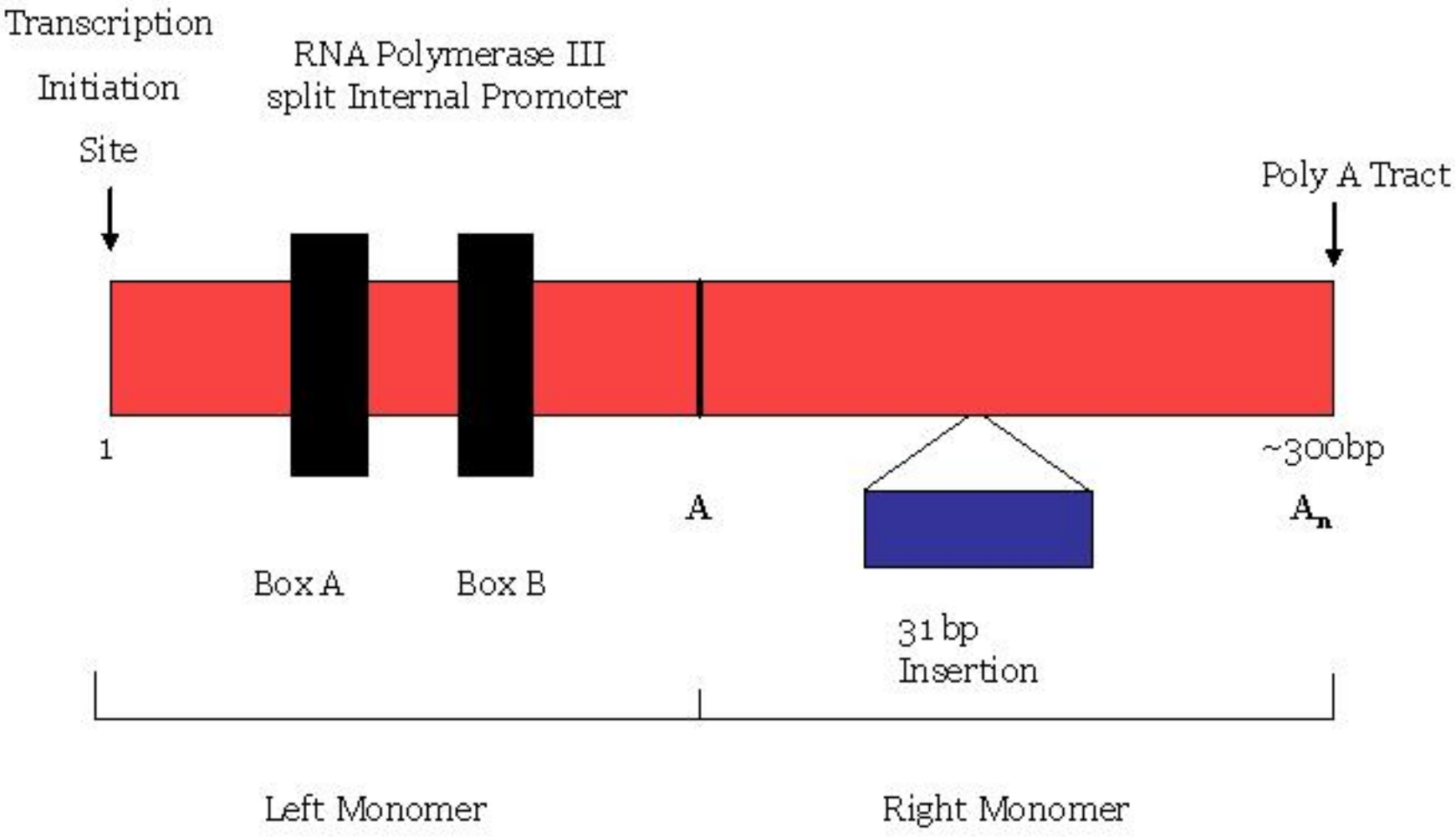


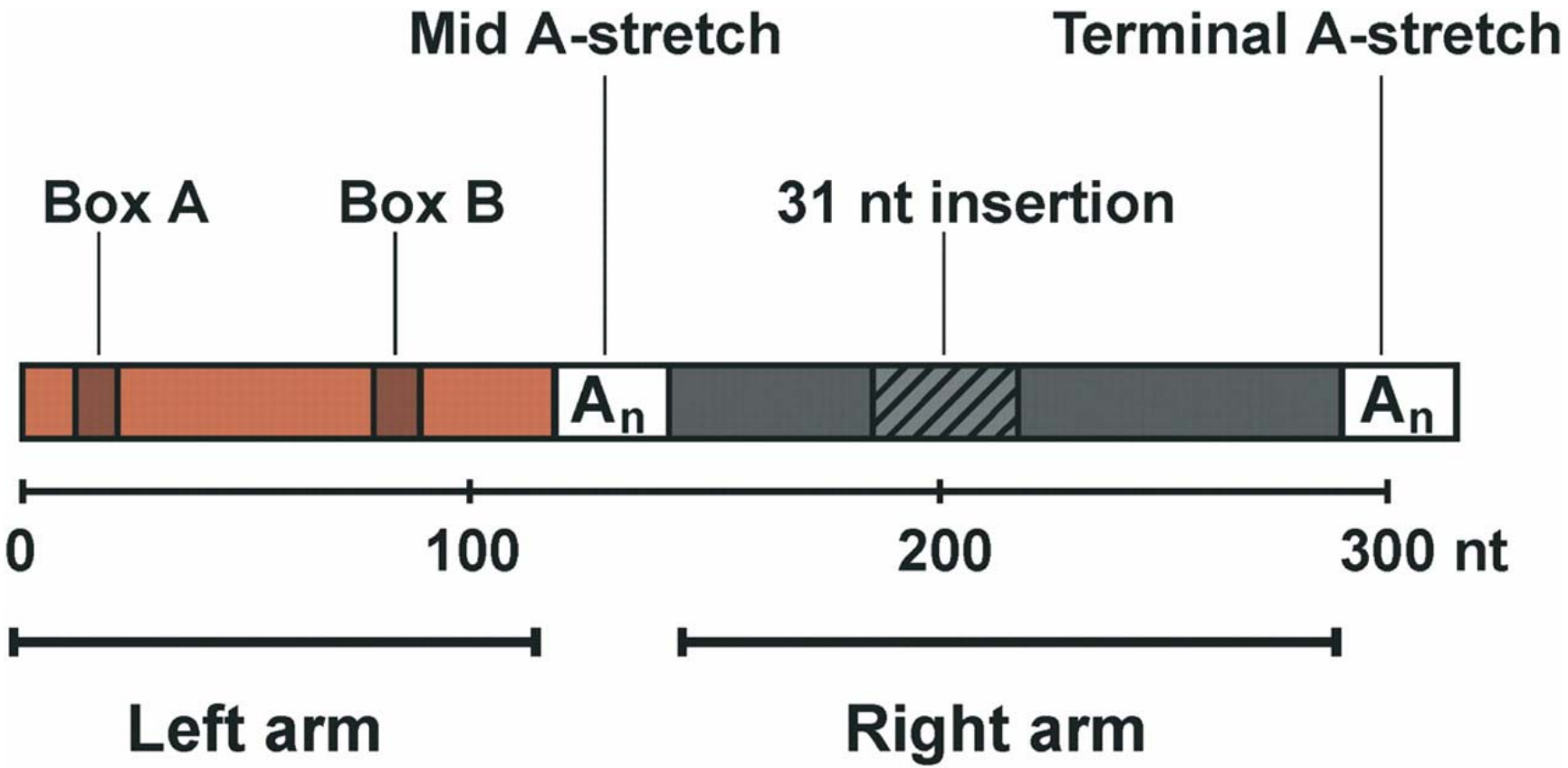
SINEs:
Alu-Elemente

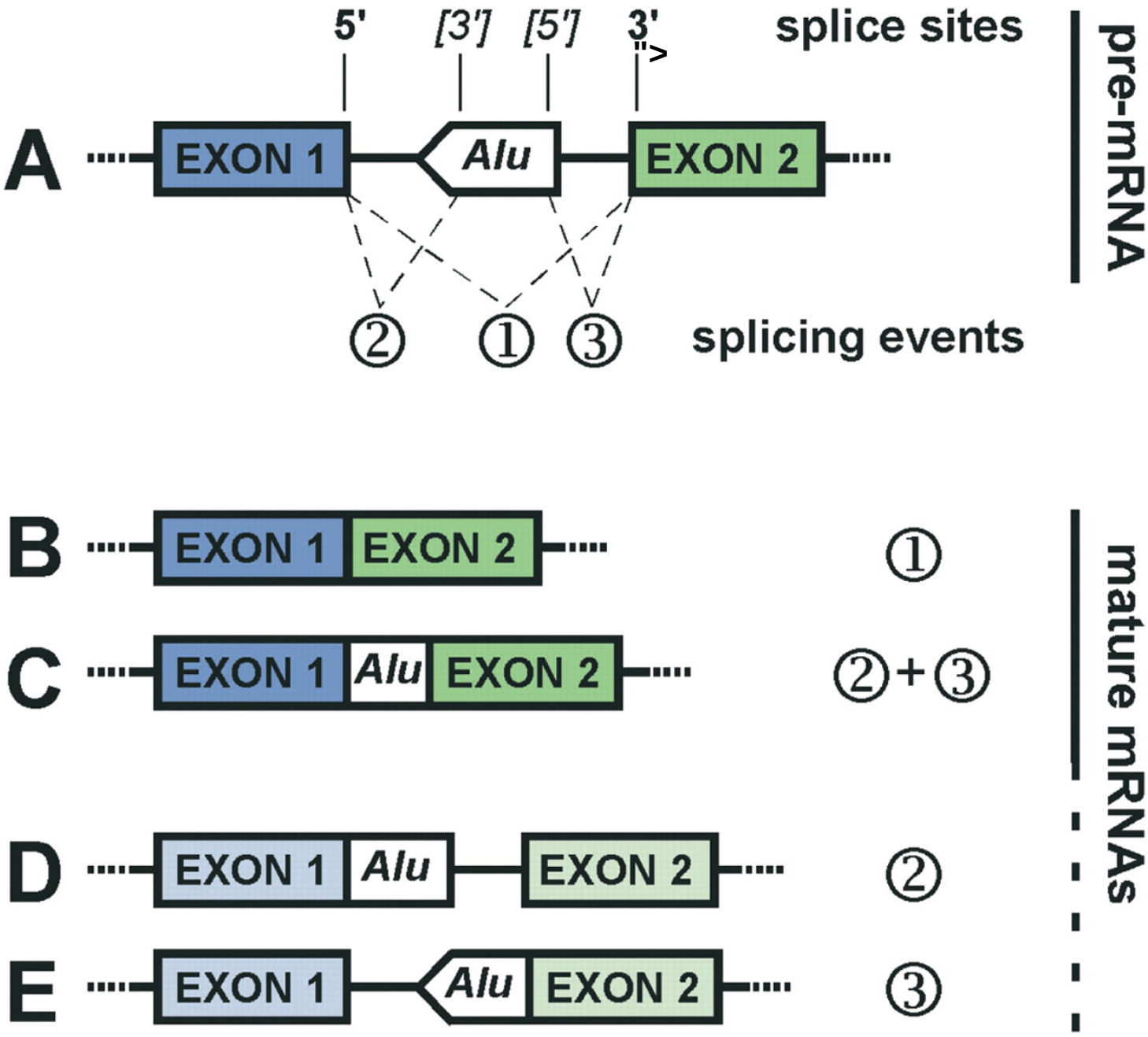
Table 11 Number of copies and fraction of genome for classes of interspersed repeat

	Number of copies (× 1,000)	Total number of bases in the draft genome sequence (Mb)	Fraction of the draft genome sequence (%)	Number of families (subfamilies)
SINEs	1,558	359.6	13.14	
Alu	1,090	290.1	10.60	1 (~20)
MIR	393	60.1	2.20	1 (1)
MIR3	75	9.3	0.34	1 (1)

SINEs: Alu-Elemente

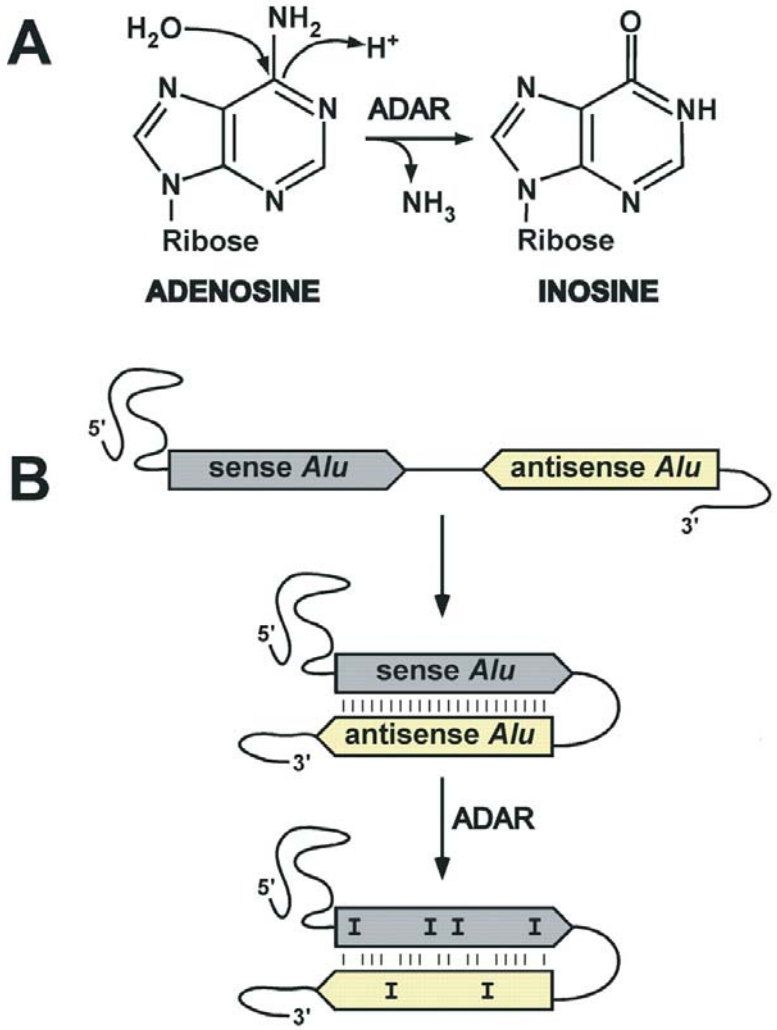



Mid A-stretch
Terminal A-stretch
Box A
Box B
31 nt insertion
An
An
0
100
200
300 nt
Left arm
Right arm



Hasler, J. et al. Nucl. Acids Res. 2006 34:5491-5497; doi:10.1093/nar/gkl706

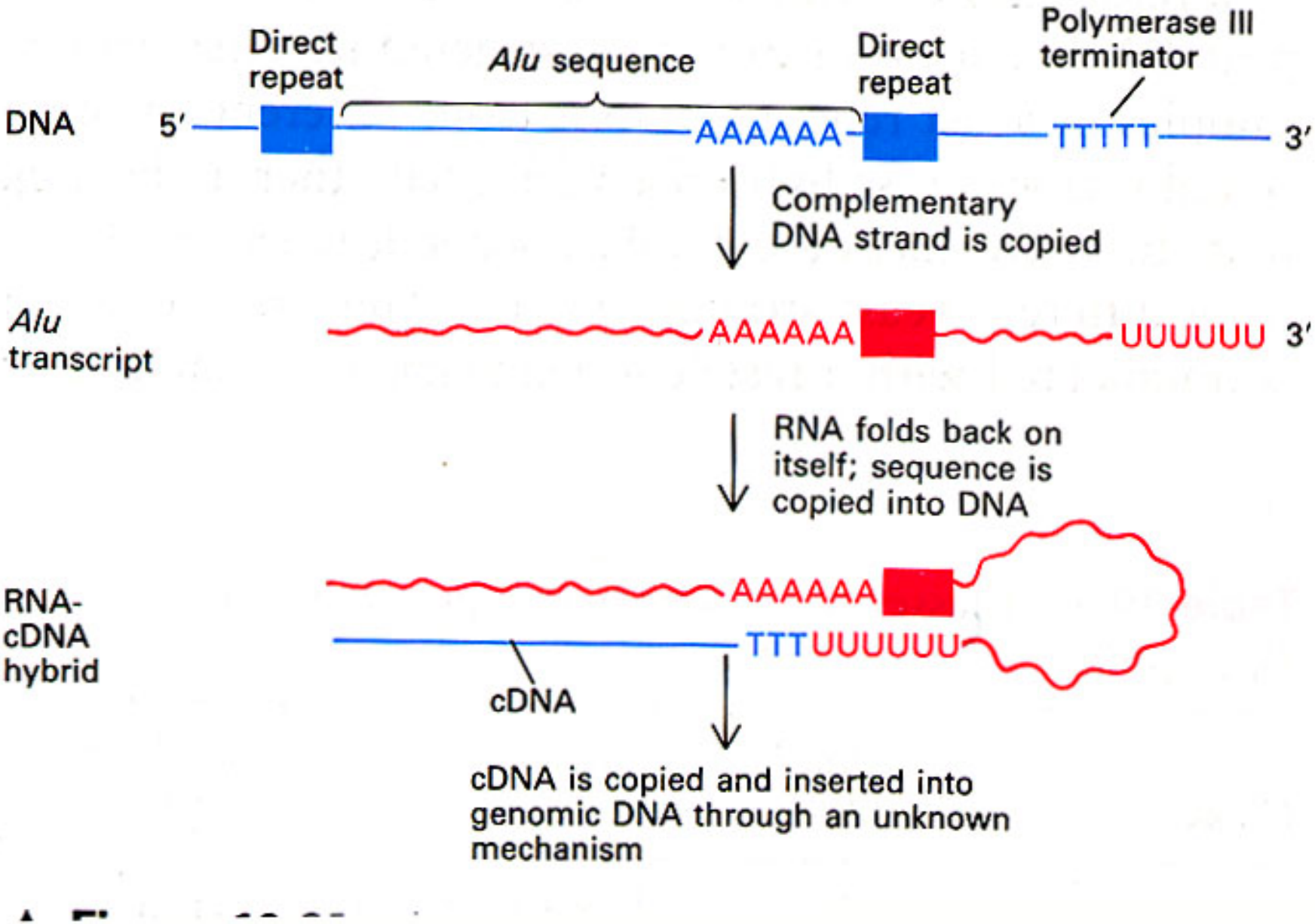
">



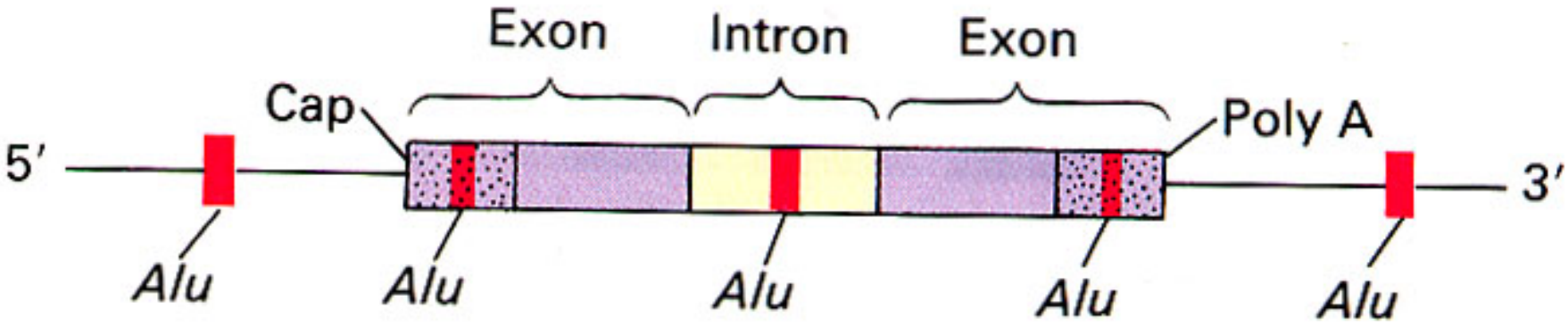
Hasler, J. et al. Nucl. Acids Res. 2006 34:5491-5497; doi:10.1093/nar/gkl706

Nucleic Acids Research

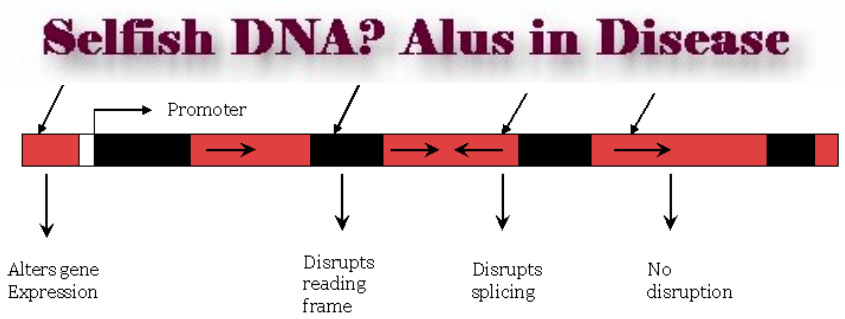
Alu-Elemente verbreiten sich durch Retroposition

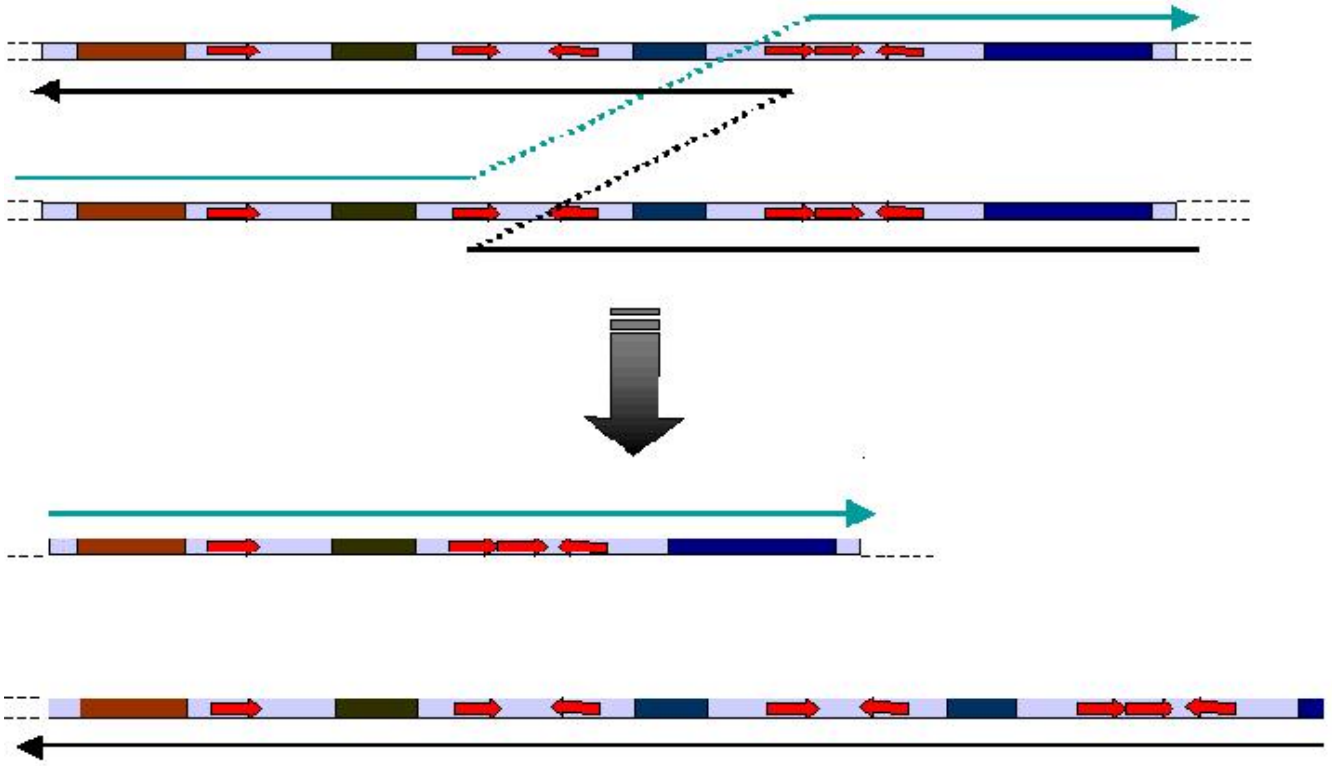


SINEs:
Alu-Elemente



SINEs.


Selfish DNA? Alus in Disease
Promoter
Alters gene Expression
Disrupts reading frame
Disrupts splicing
No disruption



LINEs z. B. L1

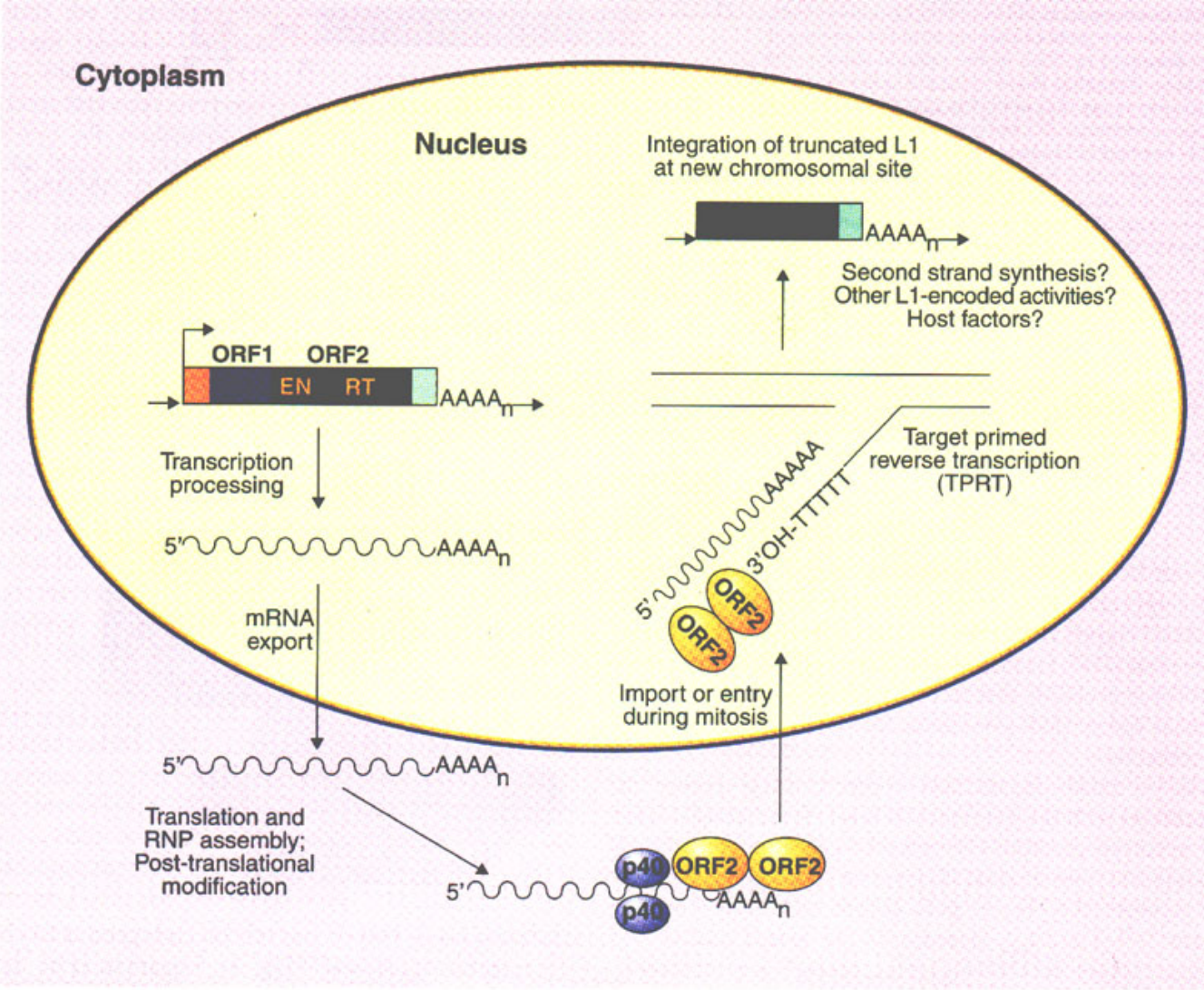
Table 11 Number of copies and fraction of genome for classes of interspersed repeat

	Number of copies (× 1,000)	Total number of bases in the draft genome	Fraction of the draft genome sequence (%)	Number of families (subfamilies)
LINEs	868	558.8	20.42	
LINE1	516	462.1	16.89	1 (~55)
LINE2	315	88.2	3.22	1 (2)
LINE3	37	8.4	0.31	1 (2)

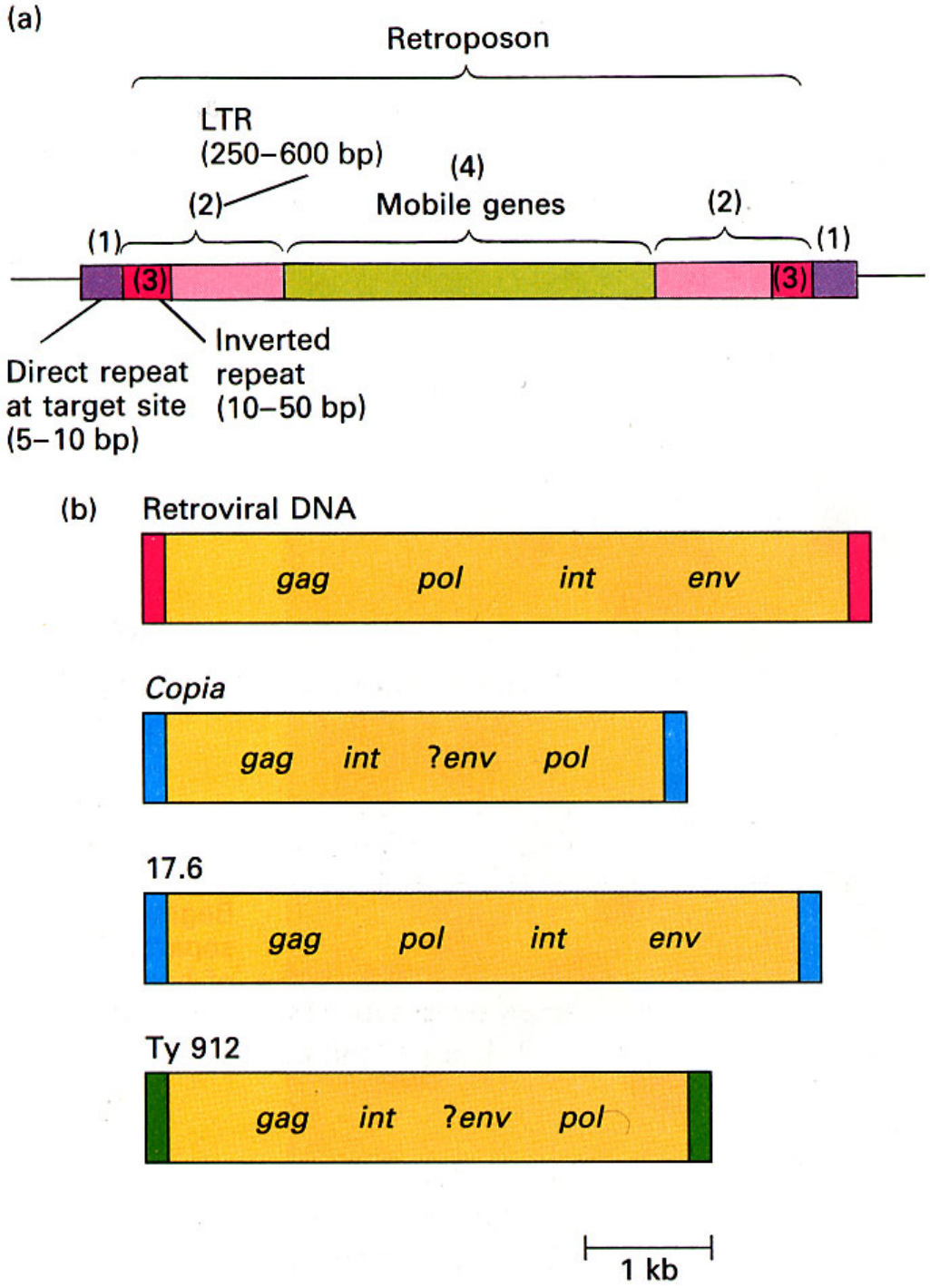
Alu elements as regulators of gene expression

Alu elements are the most abundant repetitive elements in the human genome; they emerged 65 million years ago from a 5' to 3' fusion of the 7SL RNA gene and amplified throughout the human genome by retrotransposition to reach the present number of more than one million copies.

LINEs z. B. L1


Cytoplasm
Nucleus
Integration of truncated L1
at new chromosomal site
AAAA$_n$
Second strand synthesis?
Other L1-encoded activities?
Host factors?
ORF1
ORF2
EN
RT
AAAA$_n$
Transcription
processing
5'
AAAA$_n$
mRNA
export
Target primed
reverse transcription
(TPRT)
AAAAA
3'OH-TTTTT
5'
ORF2
ORF2
Import or entry
during mitosis
5'
AAAA$_n$
Translation and
RNP assembly;
Post-translational
modification
p40
ORF2
ORF2
p40
5'
AAAA$_n$

Retrotransposons



Retrotransposons

Table 19.1 Retroposons can be divided into the viral or nonviral superfamilies.

	Viral Superfamily	Nonviral Superfamily
Common types	*Ty (S. cerevisiae)* *copia (D. melanogaster)* LINES L1 (mammals)	SINES B1/Alu (mammals) Processed pseudogenes of pol III transcripts
Termini	Long terminal repeats	No repeats
Target repeats	4-6 bp	7-21 bp
Reading frames	Reverse transcriptase and/or integrase	None (or none coding for transposon products)
Organization	May contain introns (removed in subgenomic mRNA)	No introns

Beweis für Retro-transposition