

Repetitive elements everywhere?

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Joint work with:

Claudia Fried and Peter F. Stadler

Wien, 21. Februar 2004

There are no repeats in the Hox cluster



“Es gibt keine Transposons im Hox cluster.”
(*Prohaska, Juni 2003*)

“Wenn Sie mir das beweisen können, können Sie ein 'Nature Genetics' paper schreiben.”
(*Schweizer, Juni 2003*)

“Middle repetitive elements are exceedingly **rare** within the HoxA cluster of shark. Only one is detected in the HoxA cluster. Middle repeats are abundant in the regions flanking the HoxA cluster.” (*Kim et al. 2000*)

“There is a **complete absence** of any kind of long repeats between genes of the HoxA cluster in all examined species.” (*Santini et al. 2003*)

Repeat sequences in the human genome

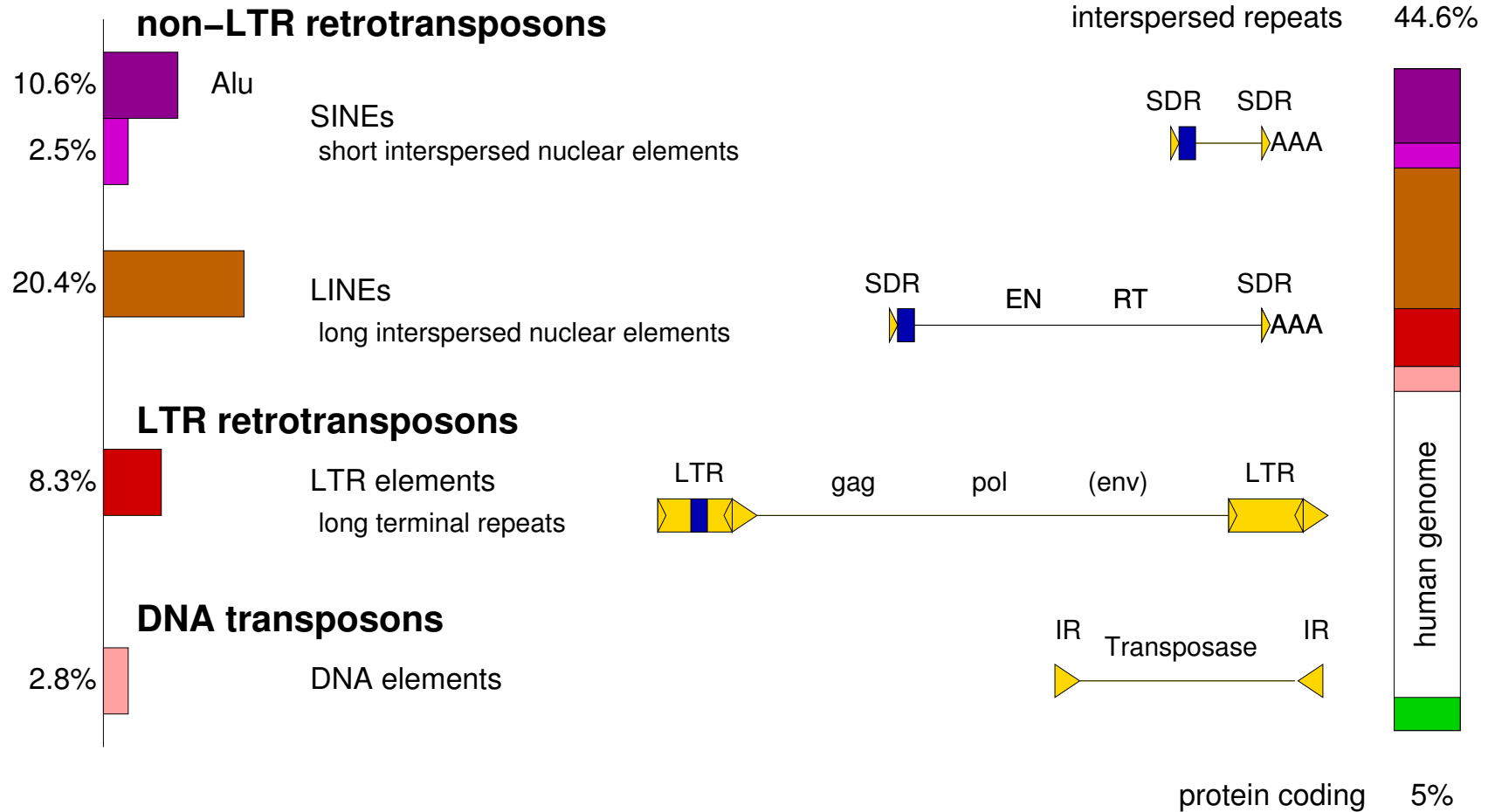
- interspersed repeats (transposon-derived)
- processed pseudogenes
- simple sequence repeats ($(AT)_n$)
- segmental duplications (10-300kb)
- tandem repeats (rDNA, centromeres, telomeres)

Repeat sequences

in the human genome

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Classes of transposable elements

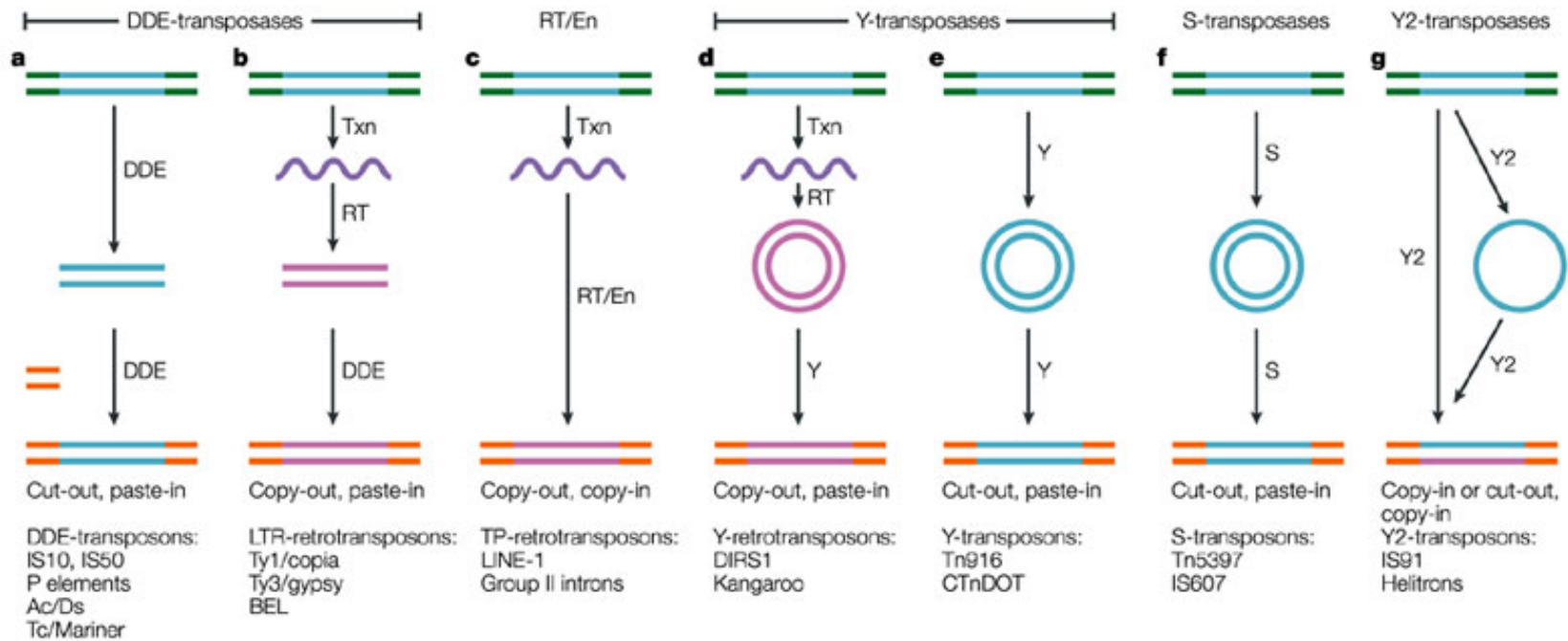


Function of transposable elements

Replicate!

Therefore, transposons are also called 'selfish DNA'
and usually **occur repeatedly** in the genome.

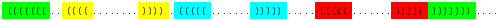
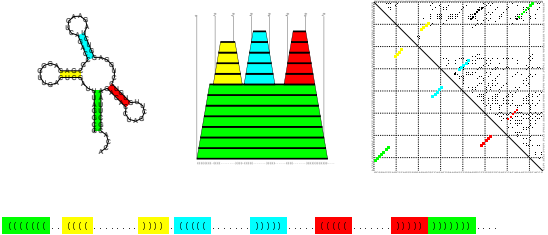
Transposition - replication mechanism



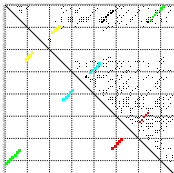
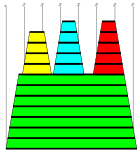
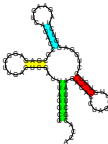
Nature Reviews | Molecular Cell Biology

Therefore, transposons are also called 'mobile elements'.

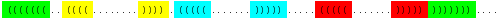
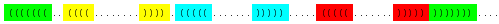
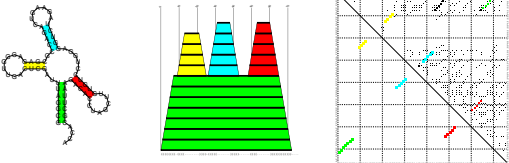
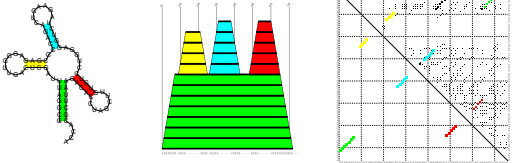
ivo's talk in Bled 2000...



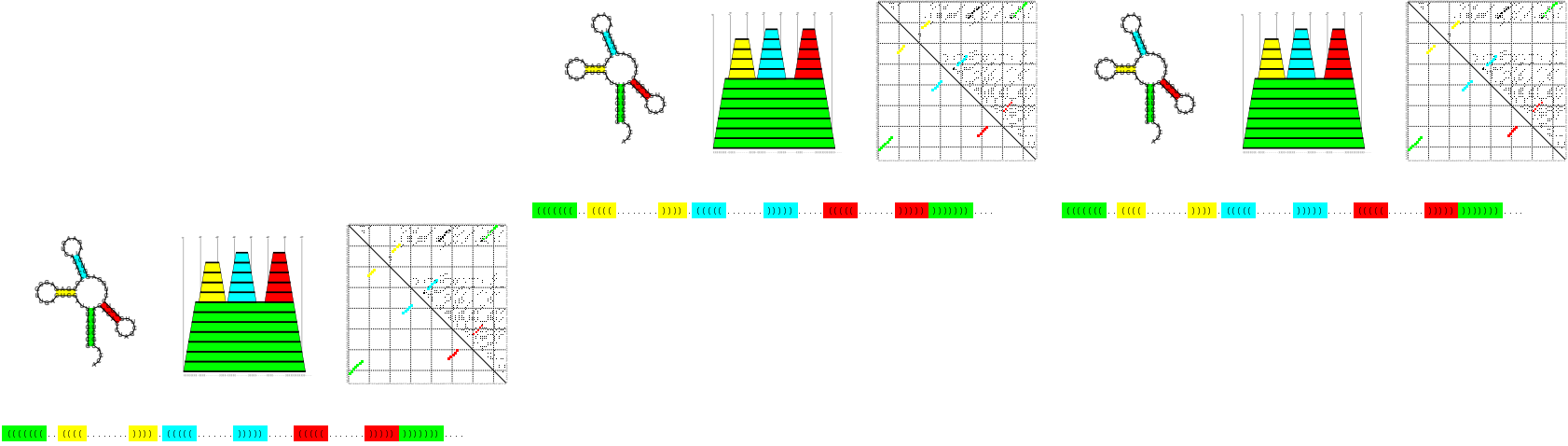
ivo's talk in Bled 2001...



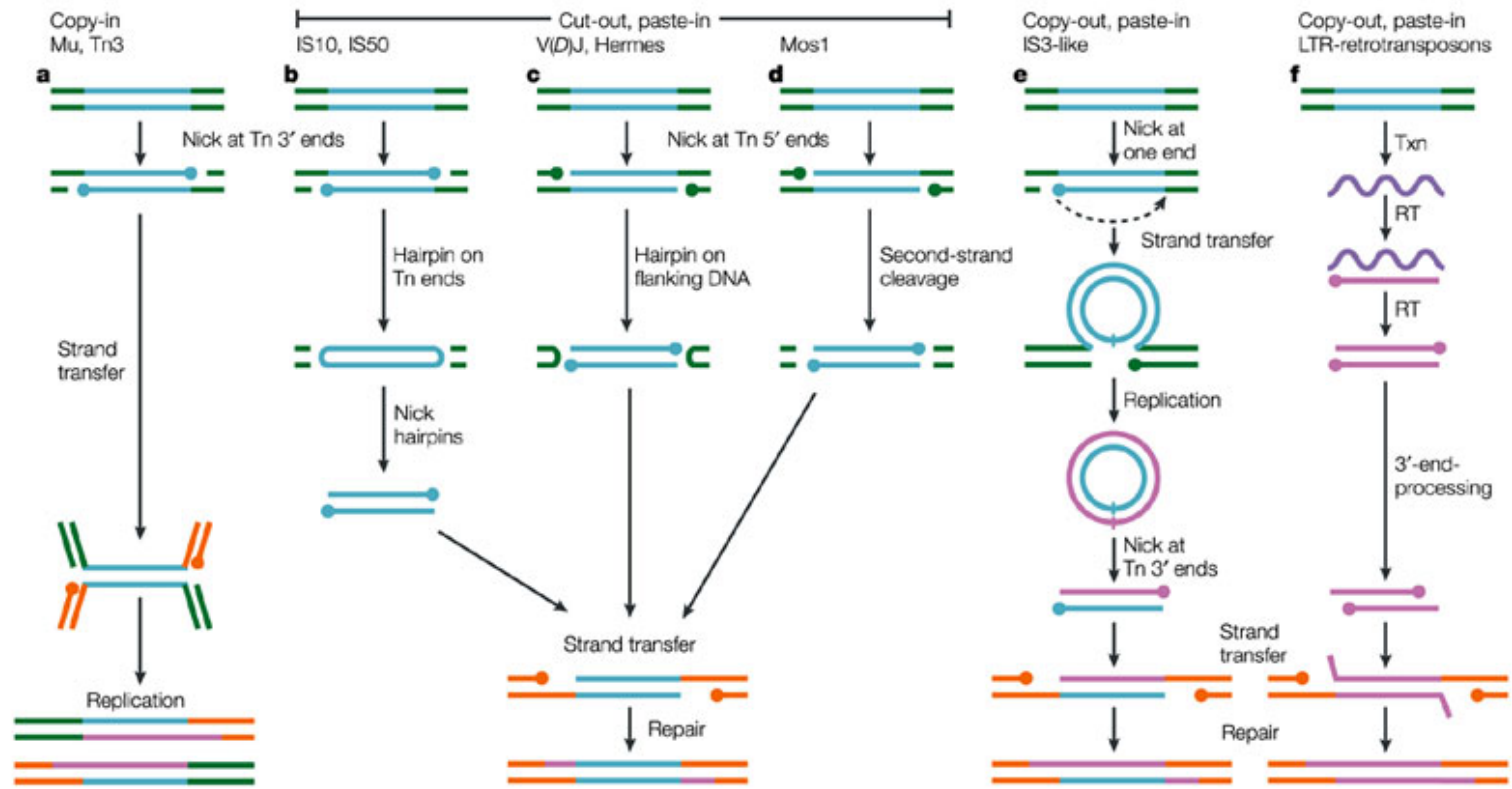
mtw's talk in Bled 2002...



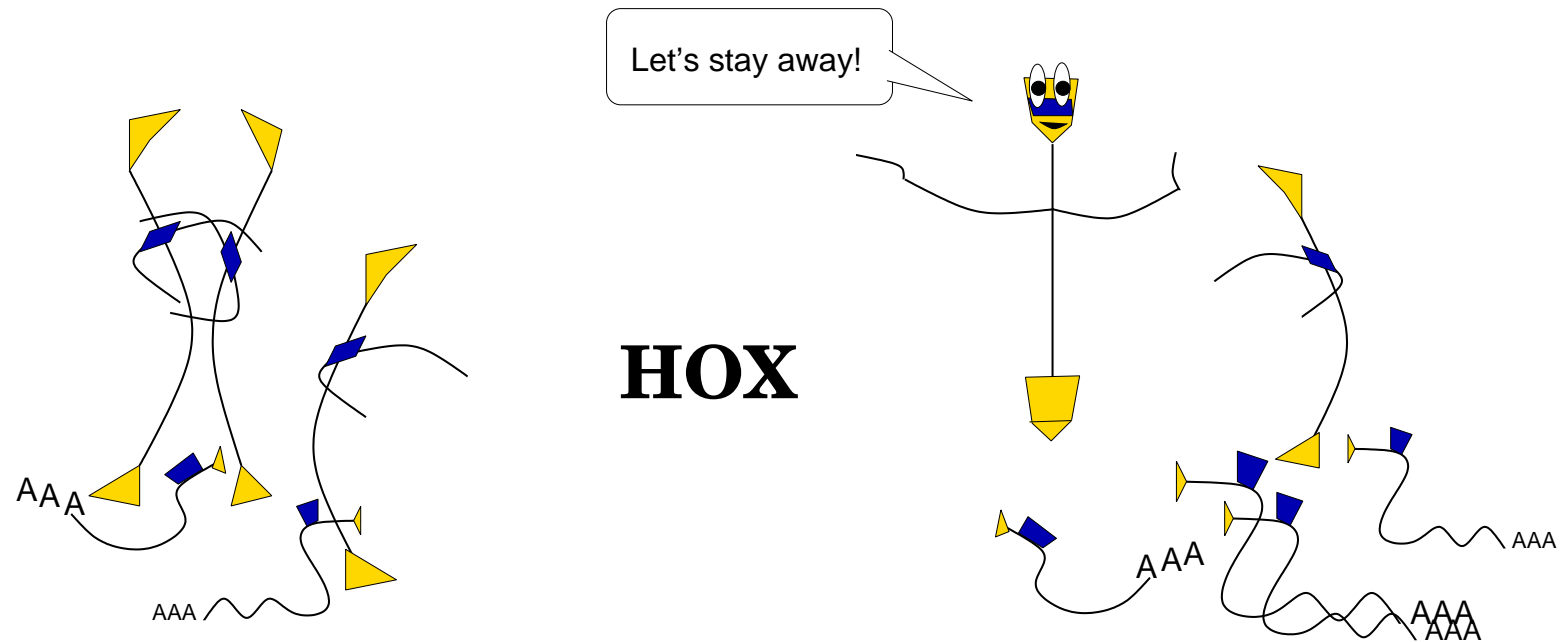
ivo's talk in Leipzig 2002...



Transposition - replication mechanism

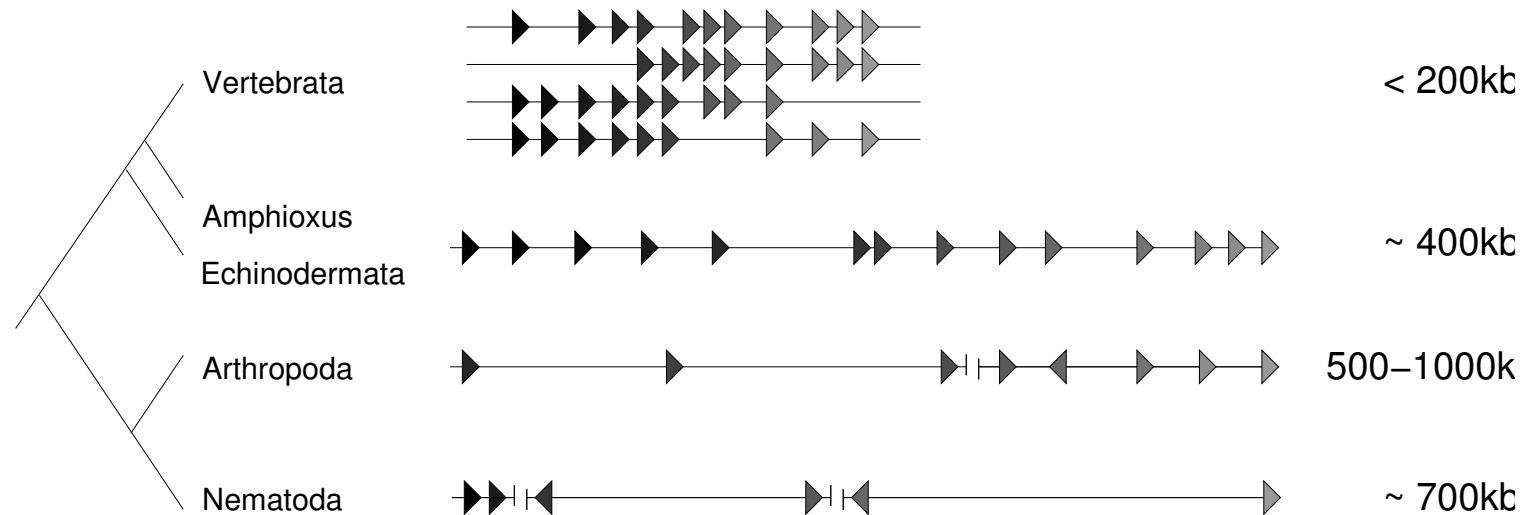


What's so special with the Hox cluster?



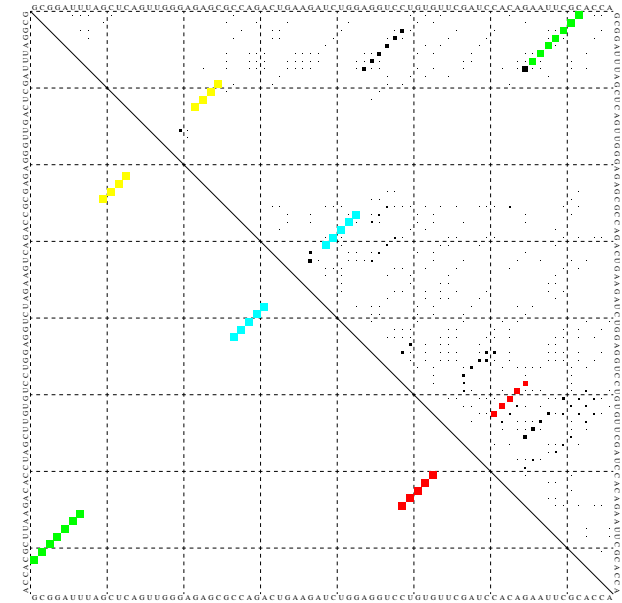
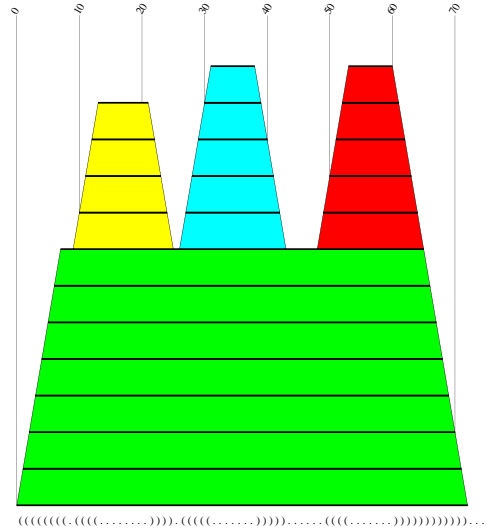
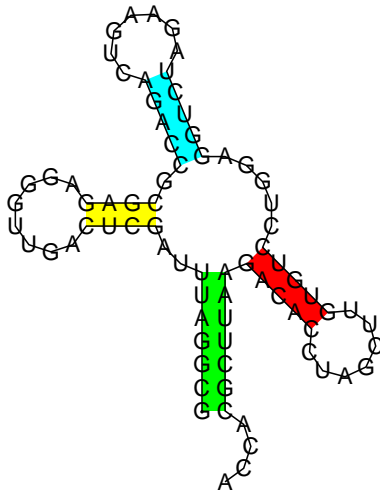
Why should evolution keep repeats away from the Hox clusters?

Hox cluster organisation



- spatial and temporal collinearity
- tightly linked cluster organisation in vertebrates
- large intergenic distances in invertebrates

Representation of Secondary Structures

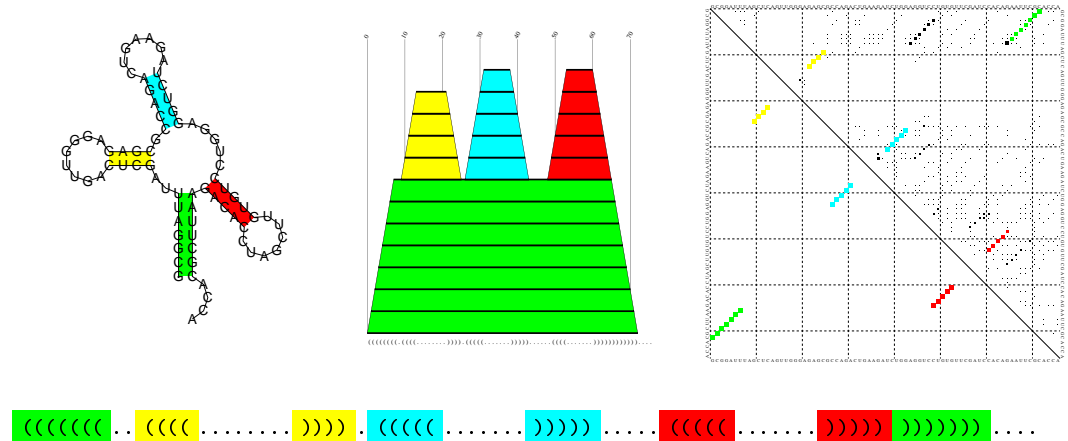


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Repetitive elements - putative effects on cluster organisation

accept repeats:

- increasing intergenetic distance
- enhanced frequency of DNA rearrangements (inversions, translocations)
- cluster fragmentation
- crossacting regulatory activity



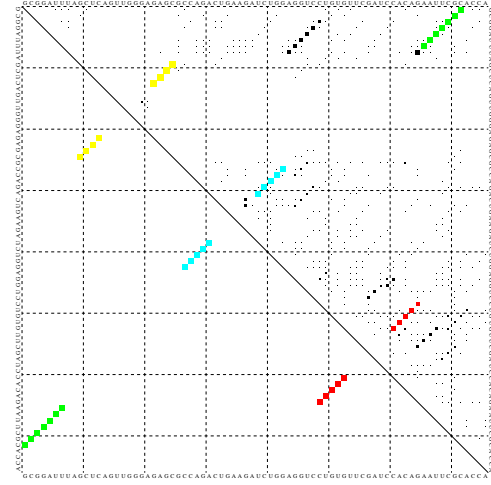
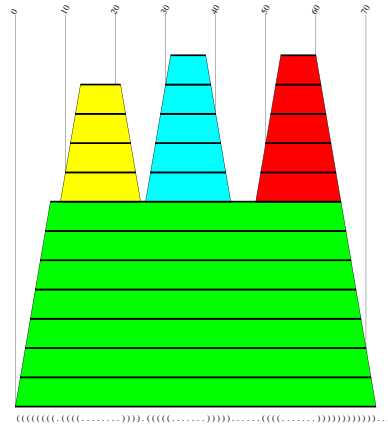
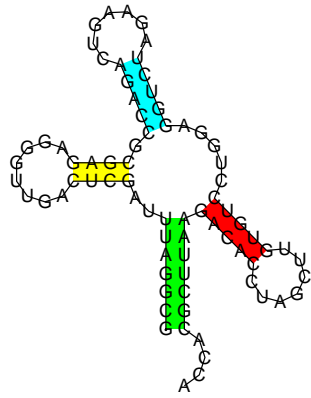
Repetitive elements - putative effects on cluster organisation

accept repeats:

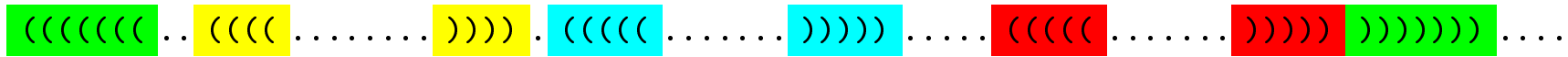
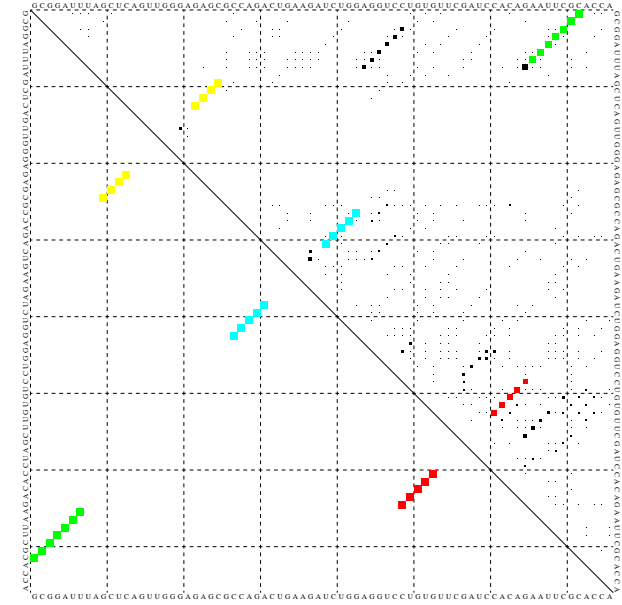
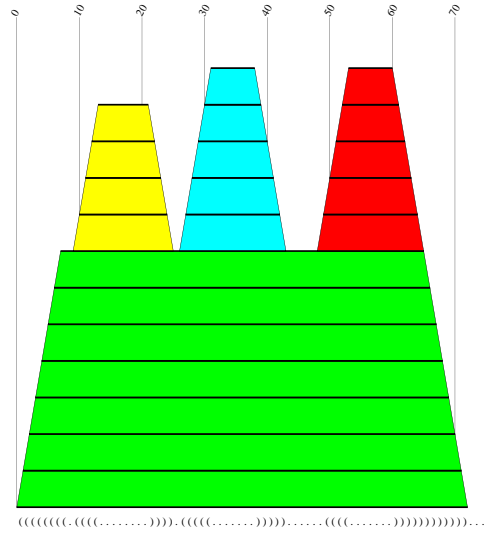
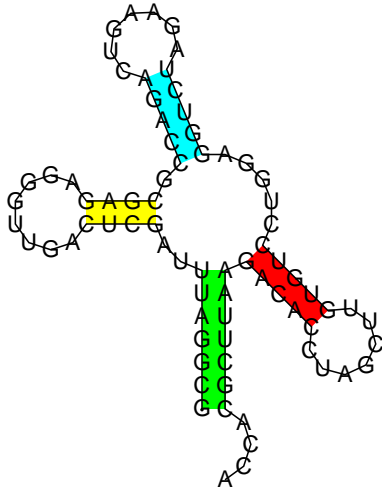
- increasing intergenetic distance
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exclude repeats:

- selection pressure against invasion of repeats
- maintenance of cluster integrity
- maintenance of intergenic distances
- maintenance of a tight, complex regulatory network



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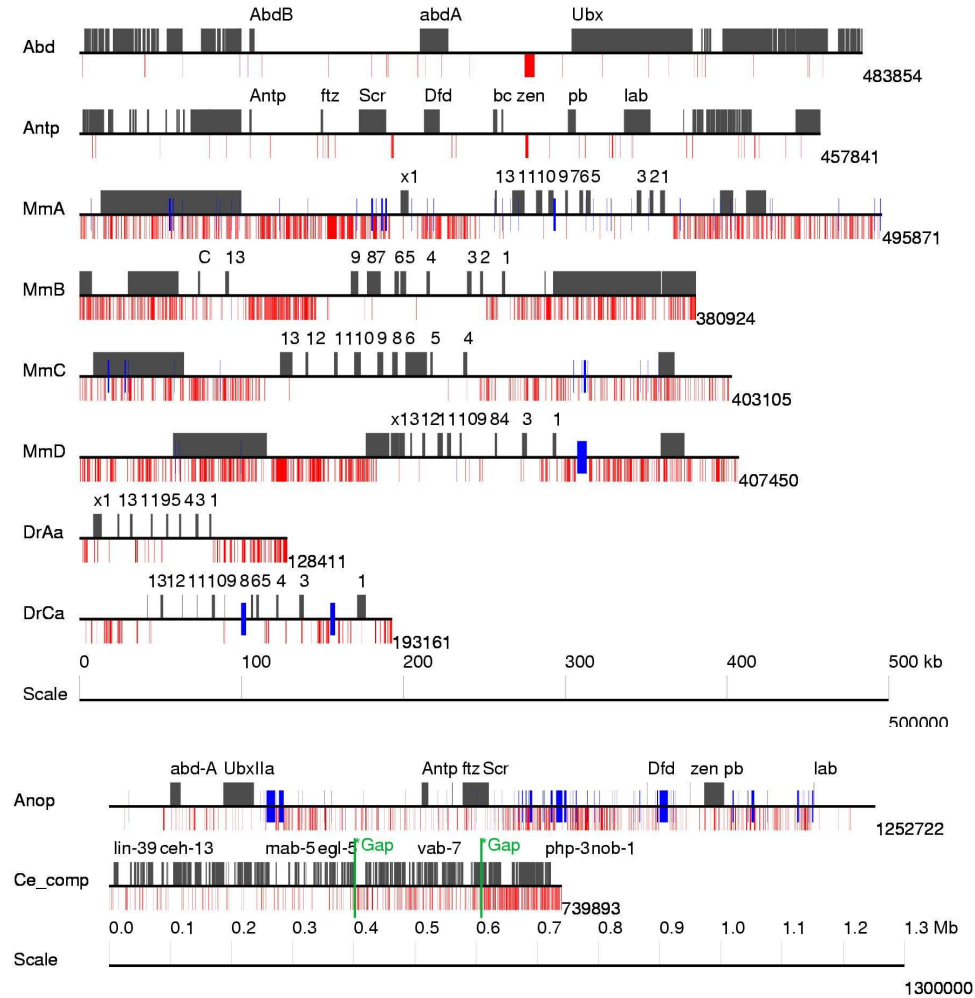


Distribution of repeats

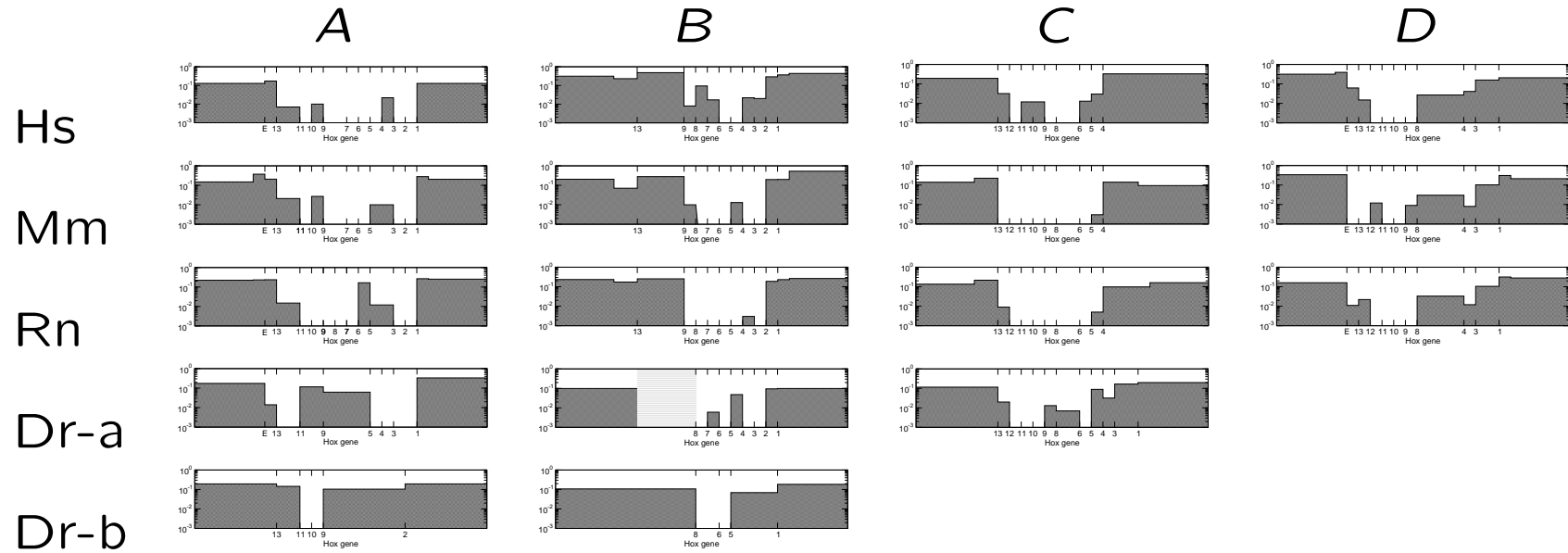
black boxes:
genes (genbank)

blue lines:
gaps (Ns in the sequence)

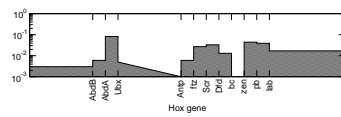
red lines:
repeats (censor)



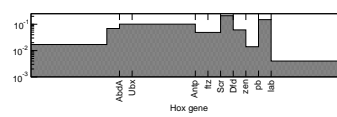
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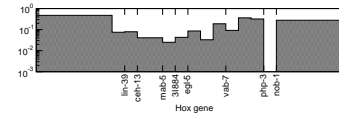
Protostomes



Dm



Ag



Ce

Conclusion and Outlook

- exculsion of repeats is a gnathostome innovation
- organisational constraints on the Hox clusters developed/tightened in early chordate evolution
- about 530 million years ago

- Why did new organisational constraints develop?
- Why is a tight organisation important - conserved for 530Ma?