BIS101 F2013 Lecture 15: Transposable Elements

Genome Size

C-value paradox

Genome size not correlate strongly with complexity (eukaryotes): C-value paradox

Varies 2000-fold in plants, from 62Mb in Genlisea to 120GB in Fritilleria (Humans are ~3Gb) Arabidopsis 120Mb, Rice, 400Mb, Pines 25Gb, Maize ~2.5Gb

No correlation in eukaryotes with number of genes (but pro vs. eu)

- So what is rest of genome? "Junk DNA"
- a. coli ~1% or less, Nematode ~5-10%, drosophila ~15%, humans ~50%, maize 85%
- Some is simple repeats: telomere, centromere. Most of it is TE

Genome size and repeat % in plants.

Majority of plant DNA in world is TEs

Transposable elements are pieces of DNA that can replicate themselves in the genome independent of the host. Essentially DNA parasites.

Transposable Elements

RNA elements Class I

- transcribe RNA, use an enzyme called reverse transcriptase (what does it do?)transcribe into DNA
- DNA moved to nucleus, integrase inserts into genome.
- copy and paste
- small to very large (20kb)
- **LTR long terminal repeat** (8% human genome)
    - thought to derive from virus (gag/pol/env)
    - LTR identical on insertions & used to date (5' LTR promoter, 3' LTR polyA signal, then each copied) -> -> in same orientation
- LINE: just a reverse transcriptase and RNA pol promoter
- **TSD** target site duplication b/c of sticky end cuts and replication to insert

- Most abdunant gene sequence in the world is reverse transcriptase

DNA elements Class II

- cut and paste using transposase
- recognizes TIR
- usually small(ish)

How do they replicate ?

- jump ahead of rep. fork
- DS break repair by homologous recombination mechanism w/ TE on other copy (or chromatid) as template
- when they jump out, leave draw in TSD in old spot in genome

Most DNA & RNA elements leave TSD because of staggered cuts.

- e.g. target site is

  AGG*TAAGG TAG

  TCC ATTCC*ATC

Other weirdos (helitrons)

- rolling circle replication
- monstrous (20kb or larger)
- pick up other genes, no target-site duplication/TIR

Autonomous & nonautonomous of each

- autonomous: code for own proteins to transpose
- nonautonomous? can use proteins but not make
- nonautonomous can vary in size, incl. host genes
- SINE = short interspersed nuclear element
    - Alu SINEs in humans ~ 11% genome

Which class will be more common in genome ?

- class I b/c copy and paste
- but turns out small nonautonomous class II called MITEs are also quite common.
    - maybe because avoid some of silencing machinery

    - maybe because insertion preferences

TE Impacts on the Genome

Impacts 1

Insertions in genes, regulatory seq.

- Different TEs have different preferences for where they insert.
- insertion obv. has big impact, often selected against.
- Where would **safe** place to insert be ❓

Retrotransposition

- new position & enhancer effects
- loss of introns, creation of **pseudogenes**
- insertion of new exons **exon shuffling**
- Exon shuffling by reverse transcriptase

Impacts 2

-New regulatory sequences (hopscotch tb1) -Rearrangement due to excision (recombination etc.) moves regulatory element

Impacts

Recombination between TEs leads to genomic rearrangements - gene loss - translocations Recombination within TEs mechanism for TE removal -- solo LTR

Impacts 4

epigenetic silencing -> DICER & RISC readthrough transcription of TEs -> silence other genes spread of methylation.

Exaptation

Exapted transposase **VDJ example**

Examples

Kernels in maize

Nonautonomous insertion

Example of sectoring

- C purple; c no purple
- Ds = dissaciator; Ds+ = no TE; on same chromosome as C
- Ac activator elswhere in genome; Ac+ not present

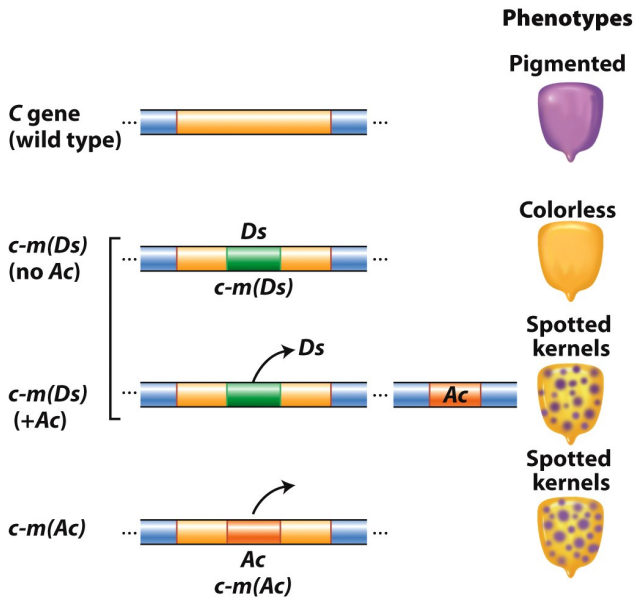


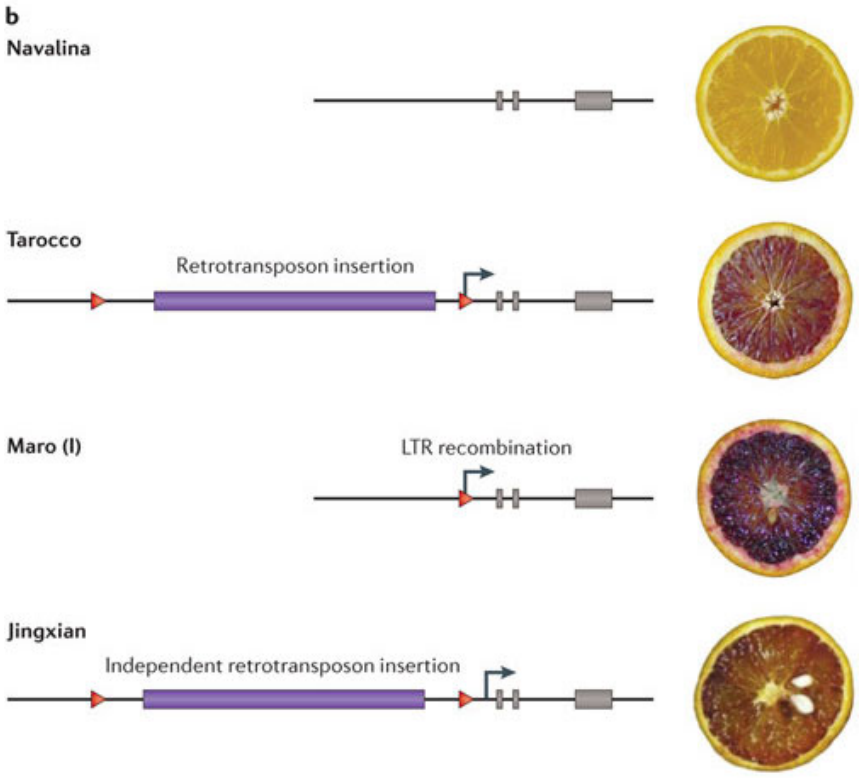
Figure 15-4
Introduction to Genetic Analysis, Tenth Edition

Ex. cross:

C/c-Ds Ac / Ac+ x c/c Ac+/Ac+

1/4 C/c; Ac/Ac+ solid
1/4 C/c; Ac/Ac+ solid
1/4 c-DS/c; Ac/Ac+ yellow with purple spots
1/4 c-DS/c; Ac+/Ac+ colorless
-really rare C/c; Ac/Ac+ purple with yellow spots

Blood oranges



Epigenetic effects

transgene for red color in white fly

DRAW density of TEs. DRAW insertions results of variegated, not. * why variegated when inserted into TE region of chromosome? (epigenetic silencing of heterochromatin)

agouti color in mice

- TE causes leads to odd bidirectional transcription that reads into agouti gene
- level of color depends on epigenetic state of TE (less sielenced, more color)

morning glory

- spread of methlyation from nonautonomous MuLe can turn off color gene

Drosophila P

not found in wild in early 20th century, now in 100% of Drosophila in wild

causes hybrid sterility in one direction of cross.

male P x female M = death or sterility.

female P x male M = no problemo

how explain ❓ P's generally silenced. Ovule cytoplasm has RNA or other mechanism needed for silencing. male does not. so if male P x female M -> P goes wild and death

P-element insertions provide mutational diversity for selection/evolution! DRAW selection experiment

V()D)J

immunoglobulin and T cell receptors production of the immune system.

V(D)J recombination takes place in the primary lymphoid tissue (the bone marrow for B cells, and Thymus for T cells)

Responsible for immense diversity of antibodies B and C lymphocytes use to recognize foreign material and prime immune response

3 loci, with V (D on heavy chain) and J regions with different numbers/types of three gene segments. Variable/Diverse/Joining regions

Each exon has an Recombinational Signal Sequence, and this RSS = TIR

Each exon is equivalent of Nonautonomous transposon!

(Recombination activating gene) RAG proteins are co-opted immobile versions of a TE w/ transposase but no TIR. So cannot move itself!

RAG proteins cause recombination of the genes

Different RSS interact w/ RAG in diff. cells for ~3×10^11 possibilities of antibodies to match bacteria, viruses, pollen