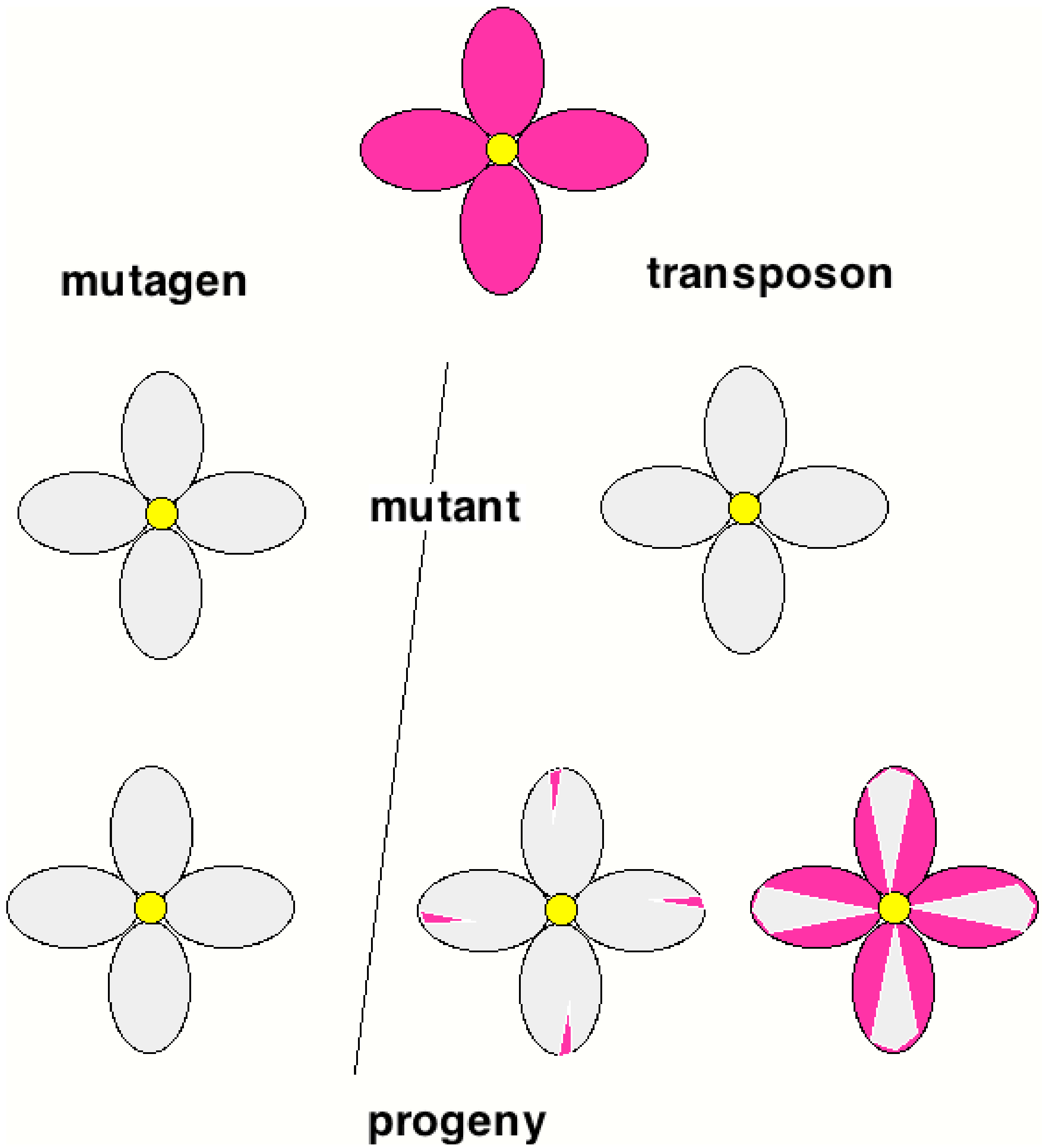

mutagen
transposon
mutant
progeny

Transposable elements

genetic originally known as genetic elements that gave rise to somatically unstable mutant phenotypes (variegation, sectoring, etc.)

were known as such decades before the realization that DNA was the genetic material

exist in all known organisms

many different classes of transposons can exist in the same species (or individual)

the likely source of highly-repetitive sequences in eukaryotic genomes

eukaryotic genomes possess much more than 50% (as a rule) highly-repetitive sequence -> transposons have shaped (and continue to shape) eukaryotic genomes

effects of transposons – may be structural (change protein-coding capacities) or regulatory (changing expression patterns) or both

Features of individual classes of transposons

members of a system may be active or inactive, autonomous or non-autonomous

individual elements may be active or inactive

historically, transposons were identifiable because they cause mutations

transposon-induced mutations may yield null phenotypes, active phenotypes, or intermediate phenotypes

transposon-induced mutations themselves change at a high rate: one may see revertants, pseudo-revertants, stable nulls, and other variants

<u>Classes of transposable elements</u>

Terminal Inverted Repeat, or TIR, transposons

viral retrotransposons

non-viral retrotransposons

<u>TIR transposable elements</u>

Examples: Ac/Ds, En/Spm

transpose in a non-replicative manner

usually move to nearby regions in the genome

classical example: Ac/Ds in maize

Ac/Ds - first transposon system described

Ac - "Activator", Ds - "Dissociator"

Ac - the active, autonomous member of the system

Ds - many different Ds elements have been identified; all are inactive (they do not transpose) in the absence of Ac (and are thus non-autonomous)

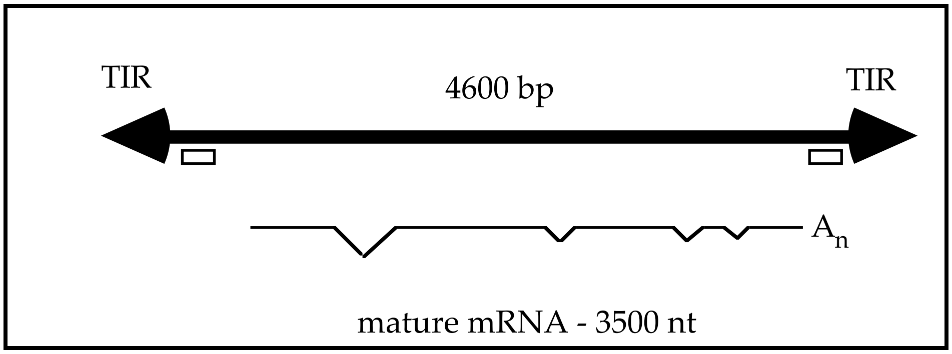
present in most, if not all, maize

transposition activity is inversely related to Ac copy number

developmental timing of transposition is also affected by Ac copy number

transposition activity, negative dosage effect, and timing are all encoded by Ac: mutations that affect these parameters can be mapped to Ac

Properties of Ac



element is 4.6 kbp

element encodes a single, extremely low abundance 3500 nt mRNA

mRNA encodes a single 807 amino acid protein (the putative transposase)

mRNA has a 650 nt 5'-UTR

element has an imperfect 11 bp terminal inverted repeat

mRNA abundance - 0.00001% of polyA+ RNA

[mRNA]   Ac copy number -> dosage effect is not a transcriptional effect

transposition   1/[transposase] -> increasing transposase levels inhibit transposition, inhibitory levels may change during development and differ in different plant species

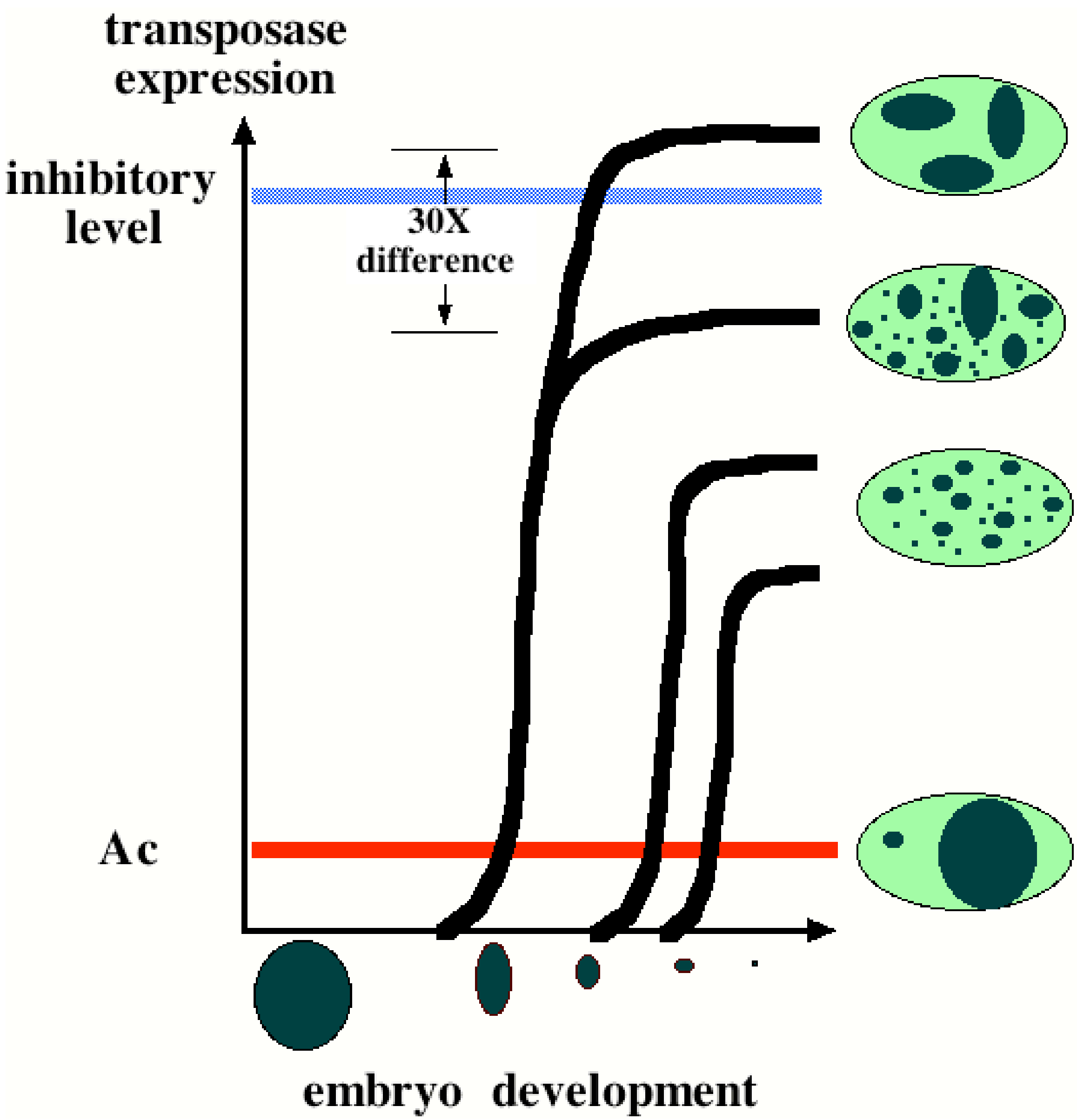
transposase - binds to "subterminal" sequences, but not the TIRs (subterminal regions are required for transposition -> all known Ds elements have these regions)

Studying transposase dosage effects

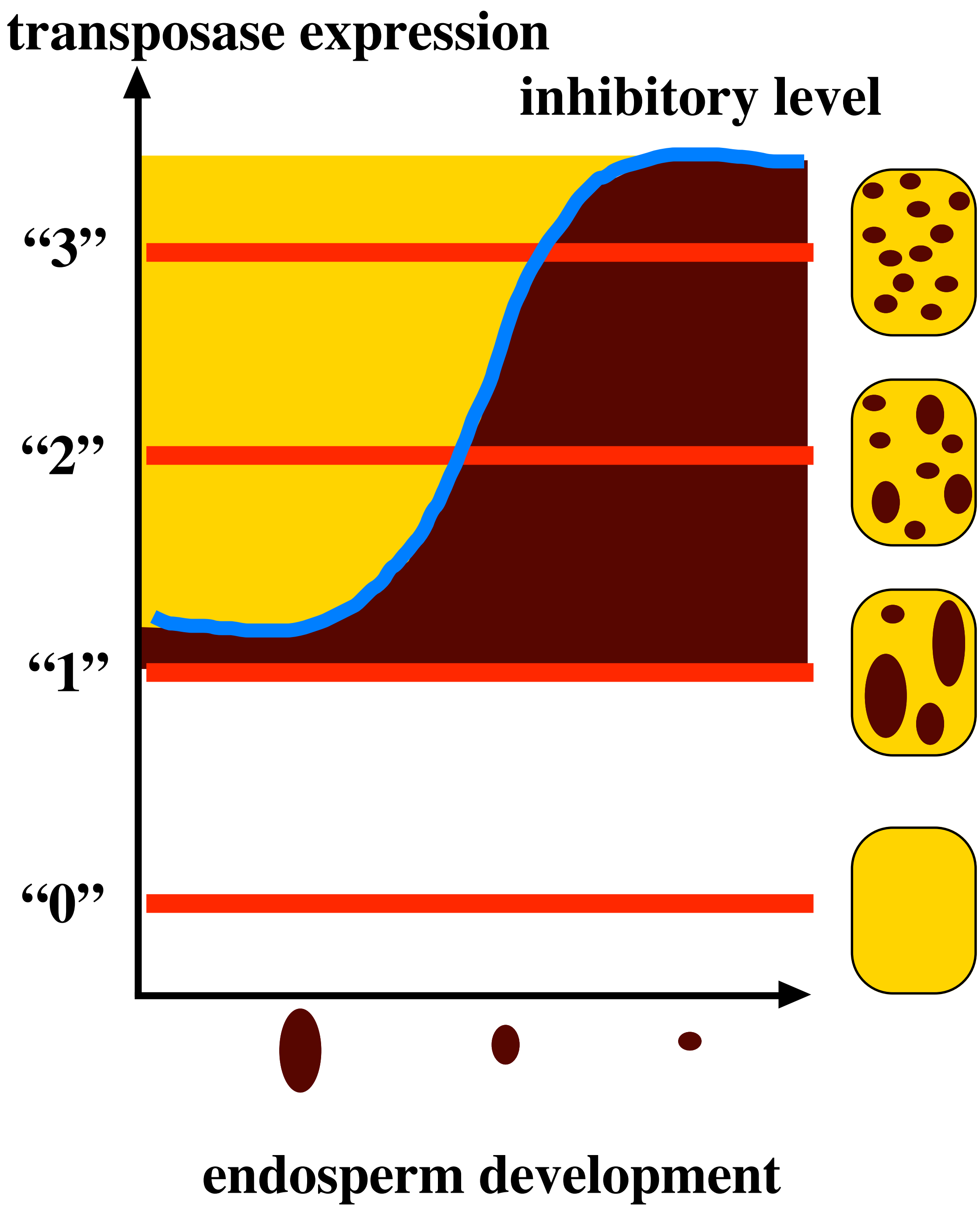
Generate transgenic tobacco with a GUS gene interrupted by a Ds element -> excision required to obtain GUS activity

Introduce transposase genes (without repeats, under control of various promoters) into these plants, monitor GUS activity in cotyledons (which are embryonic tissues)

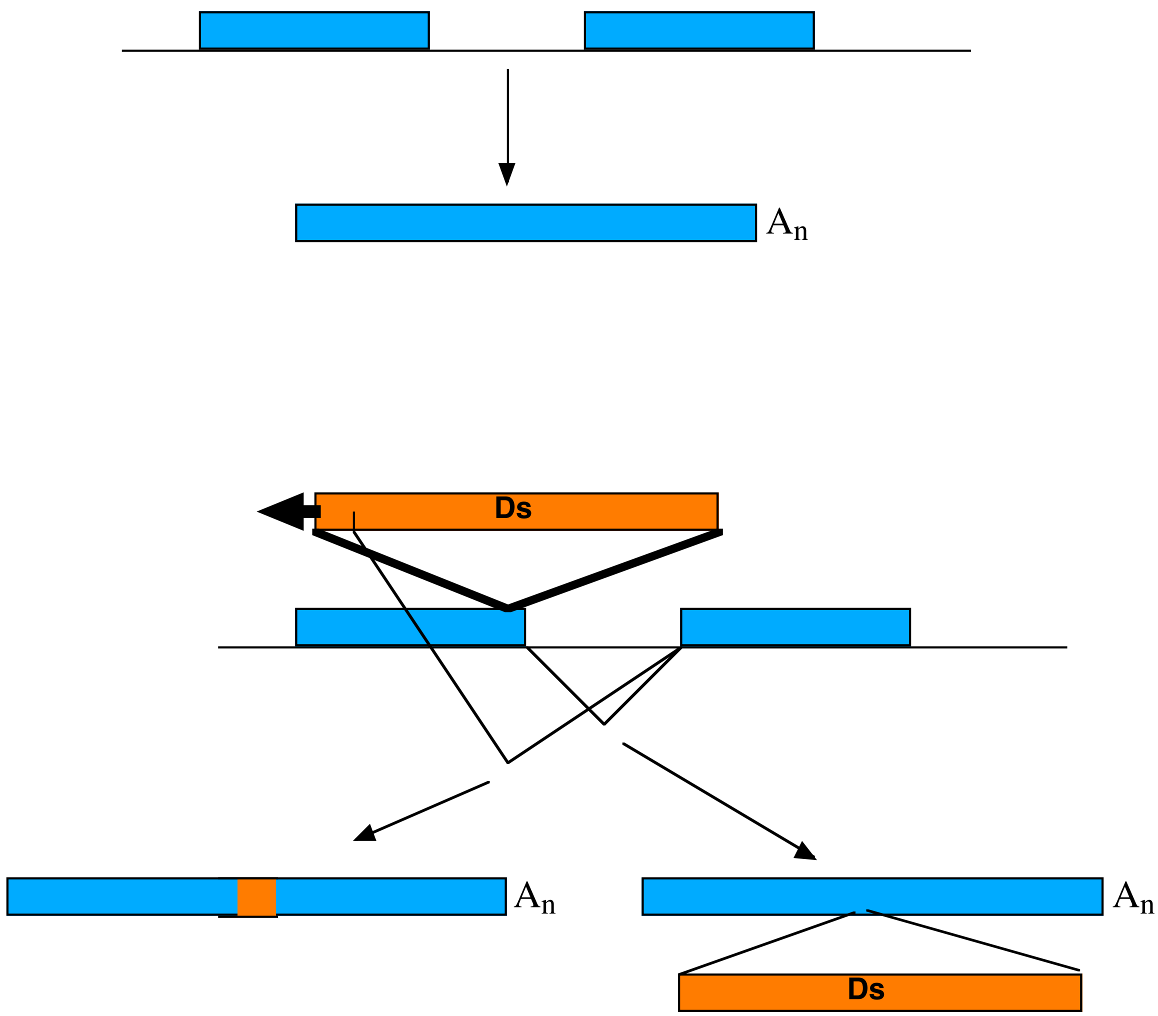
Inferences can be drawn based on size and number of GUS sectors



A model for the regulation by dosage and during development of Ac activity in maize

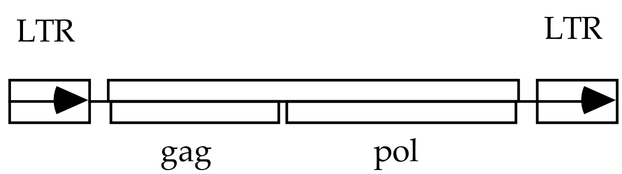


Donation of 5' splice sites by the 3' ends of Ac and Ds elements



Viral Retrotransposons

general structure:



LTR - terminal direct repeats

100-1000 bp

contain transcription control elements

rest of element - encodes 1 or two open reading frames

orf(s) have a gag-pol arrangement

gag - NTP binding

pol - protease, RNAse H, reverse transcriptase modules

these features resemble, in sequence and organization, genes found in retroviruses (and pararetroviruses)

The origins of much highly-repetitive DNA

reversion - low frequency (compared with Ac); presumably via recombination at LTRs

insertion generates 4-6 bp duplications

high copy number (>100/genome)

expression characteristics - not well understood

transposition properties - not well studied

incomplete elements - analogous to non-autonomous TIR elements (incomplete orfs)

transposition mechanism - presumed to be via reverse transcription:

mRNA

↓

DNA

↓

recombine

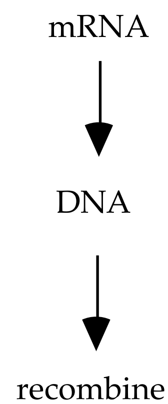
Non-viral retro-elements

Derived from reverse transcription of endogenous RNAs in the cell

Source of many highly-repetitive DNA classes (Alu, LINEs, …)

> **[polIII transcripts will accumulate as repetitive DNA because promoters are within the RNA coding region for these genes]**

transposition mechanism - presumed to be via reverse transcription:



Transposable elements and genomes

- **transposons may accumulate over evolutionary time -> repetitive DNA**
- **too much transposition = mutations! (disrupting genes or altering their expression) -> transposons are targets of silencing mechanisms (endogenous siRNAs, etc.)**
- **transposons may change genes in non-debilitating ways – protein and/or regulatory evolution**
- **transposons may be lost – recombination, selection, drift**
- **transposable elements are part of a larger dynamic structuring and evolution of the eukaryotic genome**