Barbara McClintock and transposable elements in maize

Reading: pp508-514; posted lecture notes.
Problem Set 4 for this and the next lecture.

McClintock did her work on transposable elements in the 1940s and 50s, but wasn’t accepted until the 1970s when transposable elements were discovered in bacteria.

WHY?

Proposed that the genome was not a static structure, but dynamic a changing, a revolutionary idea at the time.

Proposal came during the molecular biology revolution.

McClintock won the Nobel Prize in 1983.

Transposable elements

• 20% of the human genome contains DNA derived from mobile elements.
• Cause mutations and rearrangements
• Tools for molecular genetics
• First defined genetically by Barbara McClintock.

Activator/Dissociation

Activator (Ac)  IR  transposase  IR

Dissociation (Ds)  Derived from Ac and heterogeneous in size.  IR

Ac and Ds can move. Both require IR in cis and transposase provided by Ac.

Three kinds of events mediated by Ac/Ds.

1. Ac promotes breakage at Ds.

\[
\text{Ac} \rightarrow \text{Ds} \rightarrow \downarrow
\]

2. Ac promotes transposition of Ds

\[
\text{Ac} \rightarrow \text{Ds} \rightarrow \downarrow
\]
3. Ac can act on itself to promote breakage and transposition.

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Maize Biology and Genetics

gametogenesis

Pollen precursor cell → meiosis → 4 cells each cell divides several times mitotically to produce a pollen grain. Two sperm nuclei are genetically identical.

Ovule precursor cell → meiosis → 4 cells (three degenerate. Remaining cell undergoes several mitotic divisions → 8 cells in embryo sac: one egg nucleus and 2 central cells. All cells genetically identical.

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The fertilization product

Diagram of a kernel. The pericarp is of maternal origin. The endosperm and the aleurone layer (purple) are fleshy and are derived from the fusion of the haploid sperm cell and the diploid central cell. The embryo is diploid and develops into the plant when the kernel is germinated.

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The short arm of chromosome 9

cytological marker +/ knob C Bz Wx centromere

Colorless (C) gene encodes
bronze pigment

Bz* Purple pigment

C*/C' or C'/C - purple kernel
C-I/C-I, C-I/C', C-I/c or c/C - colorless kernel

Bz'/Bz* or Bz'/Bz - purple kernel
bz/bz - bronze kernel

C-I/C-I or C-I/C' or C-I/c or c/C and bz/bz - colorless kernel

Wx'/Wx* or Wx'/wx - starchy endosperm
wx/wx - waxy endosperm

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Ds could cause breakage.

\[
\begin{array}{c}
\text{C-I} \\
\text{Bz+} \\
\text{Wx+} \\
\text{m} \\
\text{X} \\
\text{C+} \\
\text{bx} \\
\text{wx} \\
\text{f}
\end{array}
\]

Some kernels were variegated. Bronze sectors on a colorless background. Waxy sectors in the endosperm
All three dominant traits lost

\[
\begin{array}{c}
C- & Bz^+ & Wx^+ & Ds \\
C^+ & bz & wx & Ds^+
\end{array}
\]

*In mapping experiments element causing the loss of all three dominant traits mapped one map unit to the right of Wx. Referred to this element as Dissociation.

*When she looked cytologically at tissue from kernel, could see the end of the chromosome containing the knob was lost.

Ds could move!

\[
\begin{array}{c}
C- & Bz^+ & Wx^+ \\
C^+ & bx & wx
\end{array}
\]

Bronze sectors on a colorless background. Waxy sectors in the endosperm

But occasionally found purple sectors on a colorless background, uniformly starchy.

Ds now mapped between C and Bz

Ac controlled Ds.

\[
c \text{ Ds}+/c \text{ Ds}^+; \text{ Ac}+/\text{Ac}^+ f \times C^+ \text{ Ds}/C^+ \text{ Ds}; \text{ Ac}/\text{Ac}^+ m
\]

50% c Ds+/C+ Ds; Ac+/Ac+ purple

50% c Ds+/C+ Ds; Ac/Ac+ Purple with colorless sectors

Ac acts dominantly from an unlinked site.

Ds can move into a gene!

\[
c \text{ wx Ds}+/c \text{ wx Ds}^+; \text{ Ac}+/\text{Ac}^+ f \\
X C^+ \text{ Wx}^+ \text{ Ds}/C^+ \text{ Wx}^+ \text{ Ds}; \text{ Ac}/\text{Ac}^+ m
\]

50% c wx Ds+/C+ Wx+ Ds; Ac+/Ac+ Uniformly purple and starchy

50% c wx Ds+/C+ Wx+ Ds; Ac/Ac+ Purple with colorless sectors

Starchy with waxy sectors

BUT!!

1/4000 kernels

Purple sectors in a colorless background

Uniformly starchy

\[
\begin{array}{c}
C^+ & Wx^+ & Ds
\end{array}
\]
Ds had inserted into C+ to generate an unstable c allele

Ac could also generate unstable alleles.

Isolation of the Transposable Maize Controlling Elements Ac and Ds

N. Fedoroff, S. Wessler, and M. Shure
Department of Entomology
Carnegie Institution of Washington
1165 West University Parkway
Baltimore, Maryland 21210