Phenotypic Analysis: Messages from Biology

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Grateful to:

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Phenotypic Analysis: Messages from Biology

- Phenotype is the message we get that tells us that there has been a change in a gene controlling some biological function. What we learn from the phenotype actually depends on our ability to recognize and properly interpret what we observe. In the simplest case it is the presence or absence of a measurable product (anthocyanin) or structure (ligule). In actual fact there are many functions involved in production of a particular product or activity and these are all a part of a complicated choreography leading to a certain display. Maize is an exceptionally well suited organism for demonstrating this point. We have produced and have access to a unique large collection of several thousand mutants induced by EMS, by transposons, by radiation and of spontaneous unkown origin. All are currently kept in an extensive "Mutants Data Base" with high resolution photo images and pertinent information. A duplicate copy of most of these files with images and data is also kept at MaizeGDB. This presentation will use three mutant gene systems to demonstrate the intricate relationships involved in going to and from a gene and a recognizable phenotype.
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- (1) **clf1(dek1)**; EMS induced recessive mutant; **Ac Ds-1S2,4 Clf1** transposon analysis.
- (2**)** **PgD;** EMS induced dominant chimera case.
- (3**)** **Les; Disease Lesion Mimics**; Programmed Senescence.
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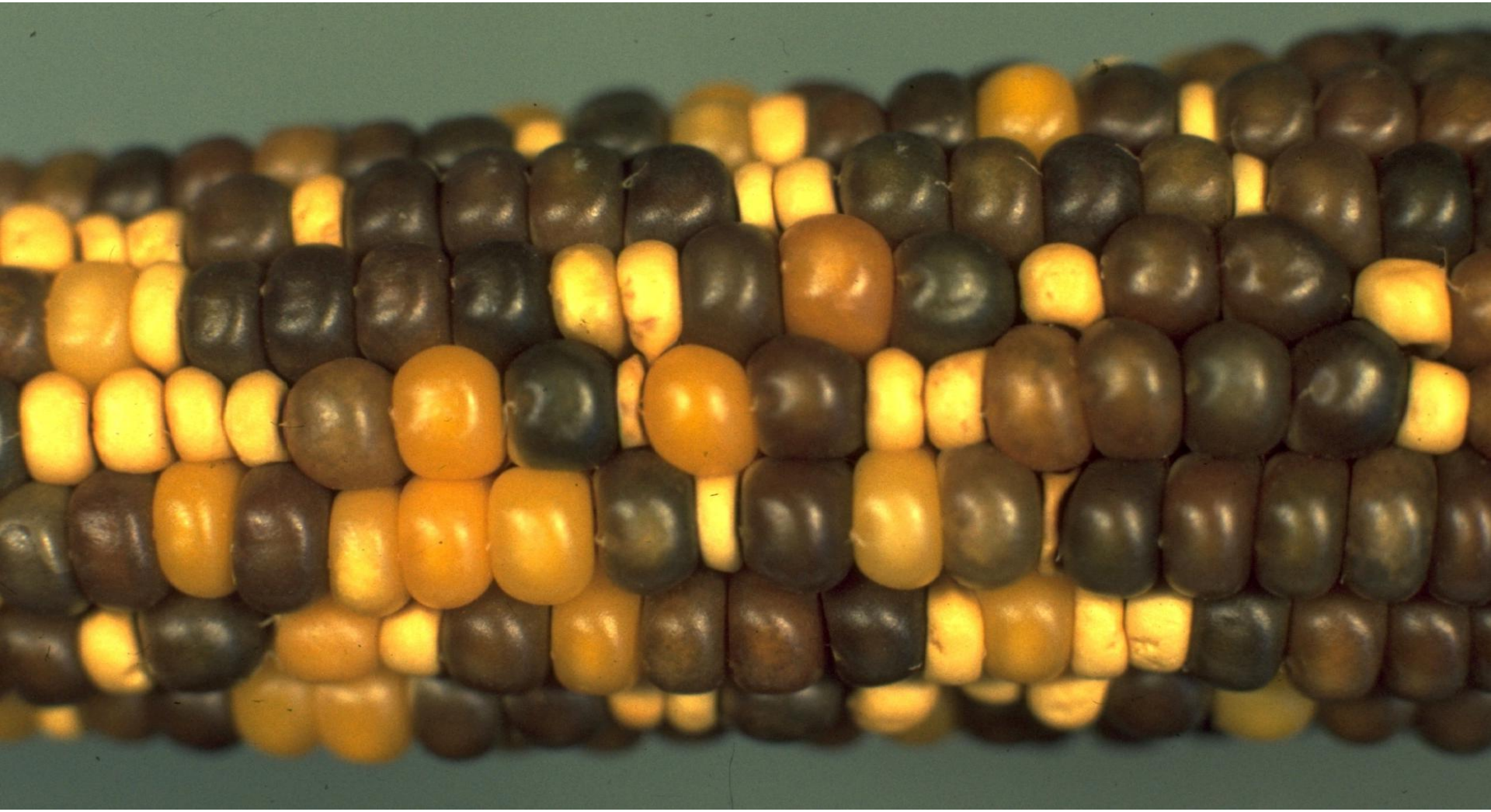
- In reviewing this material it is quite clear that there are many important genetically controlled activities that regulate expression of a phenotype without being in the biochemical pathway that leads to the observed phenotype.

33 series; 1982 M1 CORN FIELD; A632 x Mo17 EMS

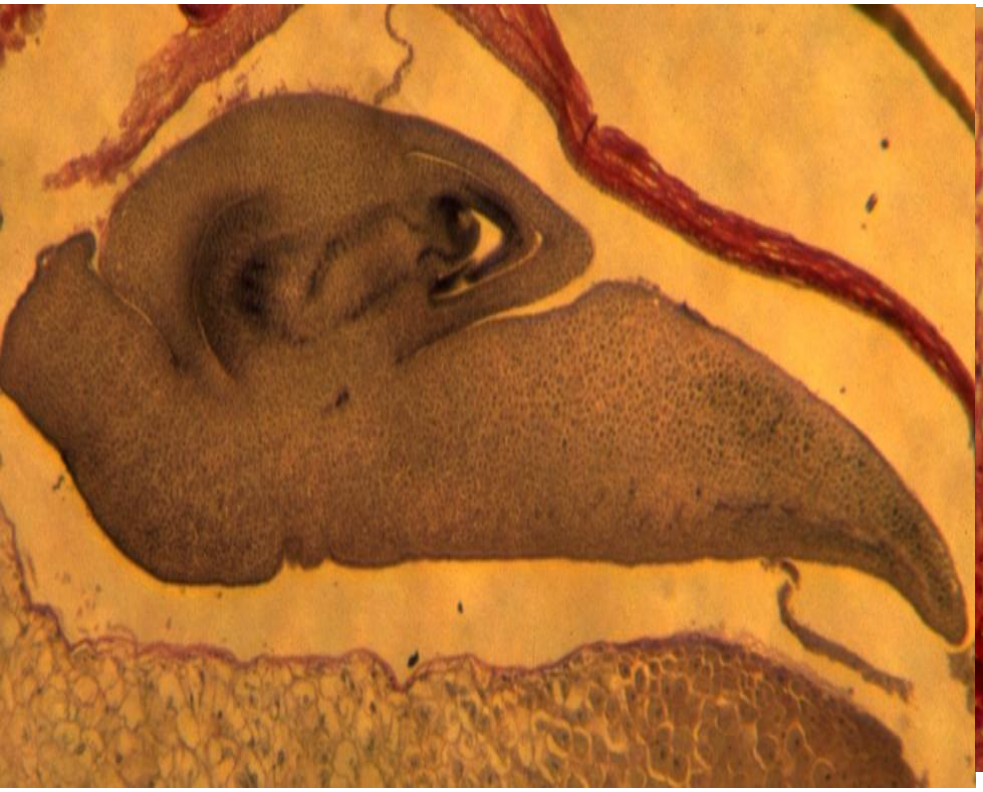
5000 M1 plants; recessive mutation rate; 1mut per locus, per 1000 pollen grains

Clf/clf*-N792, Selfed colored ACR ear segregating for colorless, floury, defective, mutant kernels

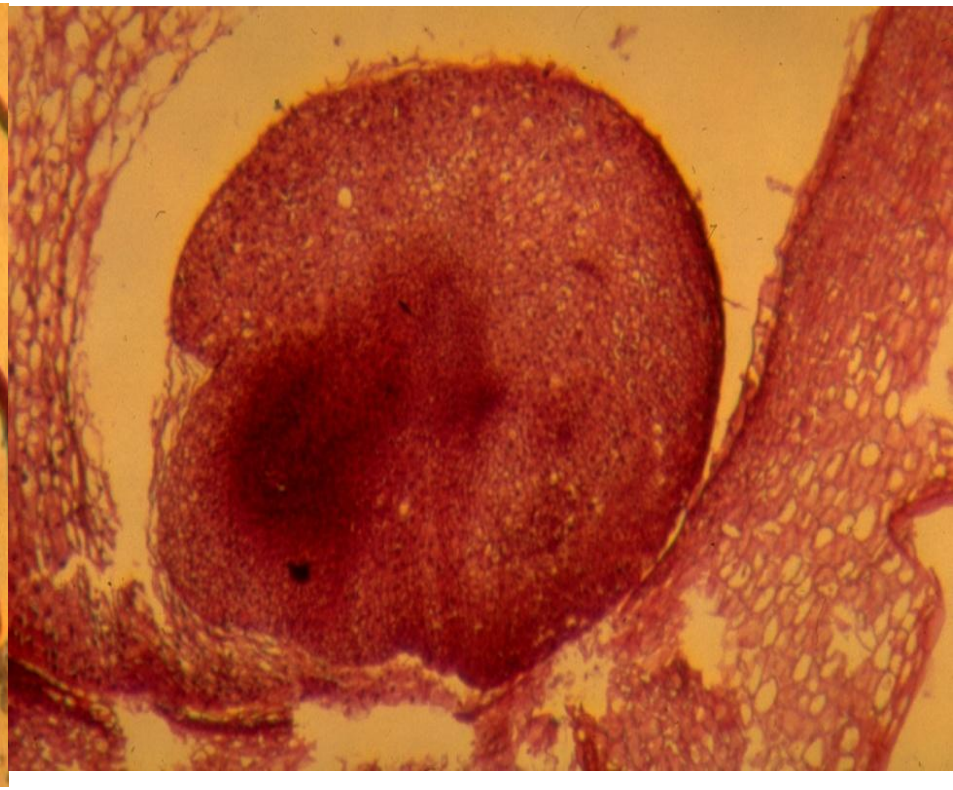
Selfed Clf/clf, C1/c1 purple A1R1Y1 ear segregating for colorless, floury and flinty kernels.



Stained kernel cross-section of a normal embryo, for comparison. Photo courtesy W.F. Sheridan.



stained section of a 16-day clf1-N971, defective colorless floury nonviable kernel showing a round embryo with root but no leaf primordia (photo courtesy W. F. Sheridan).



Anthocyanin color and starch texture expression in chimeric loss of Clf in Clf clf clf kernels due to Ac-Ds induced chromosome breakage.

Original Ac Ds-1S2 Clf/clf/clf mutant kernel with two large attached colorless areas and many smaller lace like colorless sectors (repeated later losses of Clf). Why no floury statch?

Clf/*clf* x *Ac* Clf *Ds-1S4/clf* ears with full colored normal, colored colorless mosaic, collapsed *dek* kernels. The mosaic kernels have large patches of colorless tissue set off from smaller areas of colored cells that are often arranged in chains of dots and isolated islands of intensely colored tissue of one or more dots . The largest colorless areas have an interior of white floury tissue bordered by a yellowish flinty tissue between the floury and the colored areas. Non-aotonomous expression.

Top leaf surface of Ac Ds-1S2 Clf/clf plant from original mosaic kernel. Showing small long narrow sectors of indented tissue. Largest one, lower left, is white indicating absence of chlorophyll (albino).

Underneath surface of same leaf section (left), showing sectors which are indented on the top, protrude on the undersurface conforming to and confirming the aberrant aleurone and leaf shoot morphology. The largest sectors is also white.

Pleotropic Phenotyes of clf1

1. Failure to produce purple aleurone due to absence of aleurone layer(Cone 1989), not involved in anthocyanin pathway. Message is cell autonomous,

2. Failure to produce normal scutelum and shoot primordia but allows poor minimal root growth.

3. Failure to produce hard vitreous starch; message is non-cell autonomous

4. Failure to produce normal leaf tissue in chimeral loss sectors but instead show a morphological indentation of top leaf surface and a concordant protrusion of bottom leaf surface of the same sector

5. Large sectors are seen to be white indicating failure to produce chlorophyll

Original PgD*-N2542/+ mutant chimeric plant showing broad pale-pale green sector covering 1/2 of the leaf blade, sheath and tassel. This sector is physically supported and nutritionally sustained by normal plant tissue. Note necrosis in cross bands in pale green chimeral tissue only on older leaves.l

A vigorous, pale green dwarf PgD*-N2542/+ mutant plant from the 1st outcross on + + normal.

Progeny from PgD*-N2542/+ mutant crossed on normal showing small pale green lazy dwarf plants. Under different field conditions.

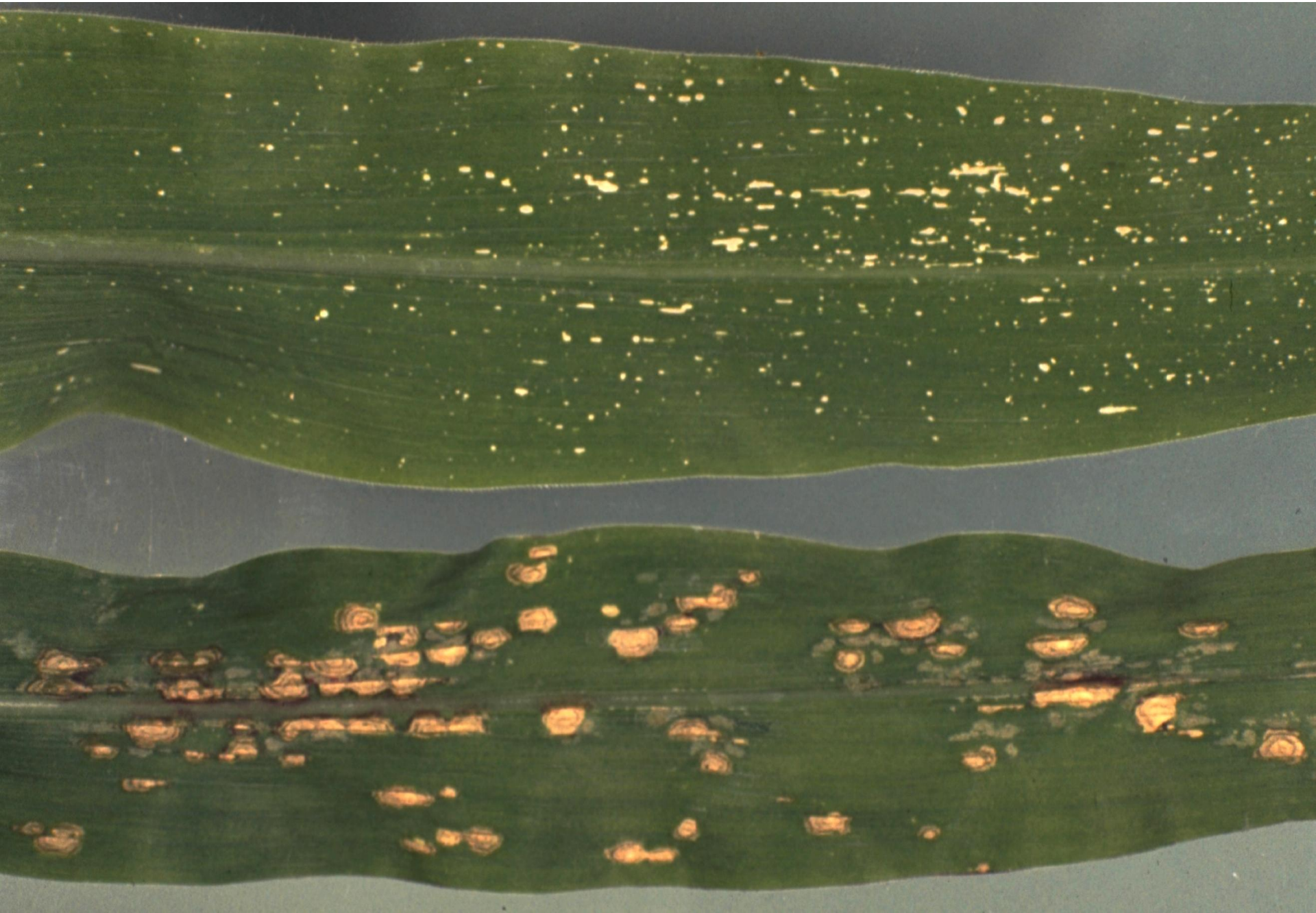
Progeny from PgD*-N2542/+ mutant crossed on normal showing a small pale green lazy dwarf plant.

A PgD*-N2542/+ mutant plant from a cross on normal showing small pale green lazy dwarf phenotype Supported so as to produce pollen.

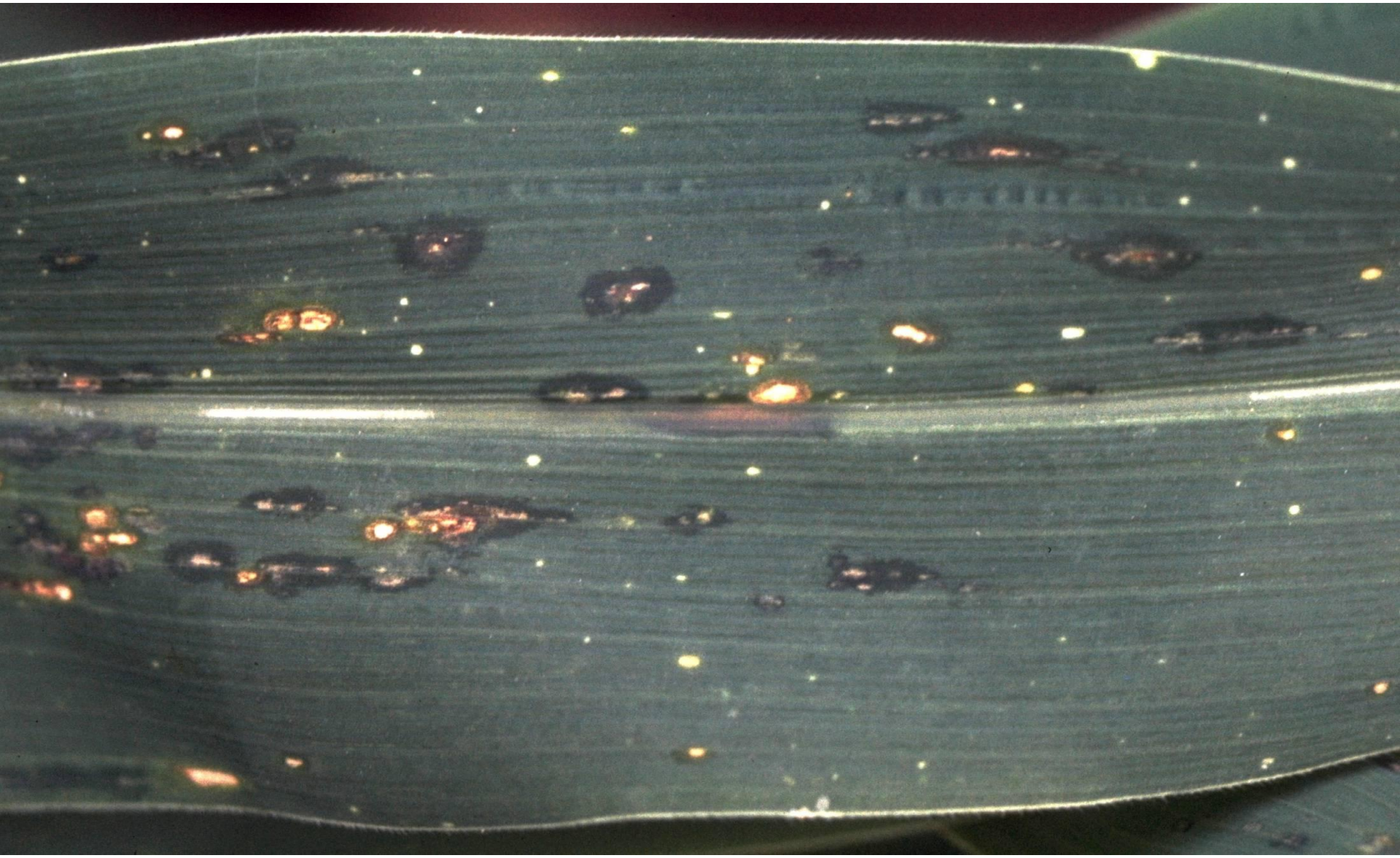
Pleotropic Phenotyes of PgD

- **Dominant pale-pale green lethal mutant chimera mechanically supported by and nutritionally sustained through pollen shedding by healthy normal tissue. "light green lethal phenotype"**
- **First out cross(using pollen from chimeric plant) to genetic stock gave dwarf pale green mutant plants that tended to fall over so that normal appearing pollen was hard use. "The dwarf and lazy phenotype"**
- **Difficult Cross of this pollen on W23 produced "healthy pale green" mutant plants that were almost normal in vigor while that on Mo 20W were weak & fragile like the first out cross indicating a strong complementary modifier in the former**

Leaves of two Les mutant plants. Top, Les2-845A/+, extreme expression resulting from favorable modifiers and conditions. Bottom, Les1-843/+, standard expression showing differences in lesion characteristics.



Les1 & Les2 interaction; Leaf of a Les1-843/+, Les2-845A/+ plant, grown under winter conditions in the greenhouse, showing large dark Les1 lesions with light centers and dark watery halos and small round white necrotic Les2 lesions. Some Les 2 lesions appear within the halo of a Les1 lesion.



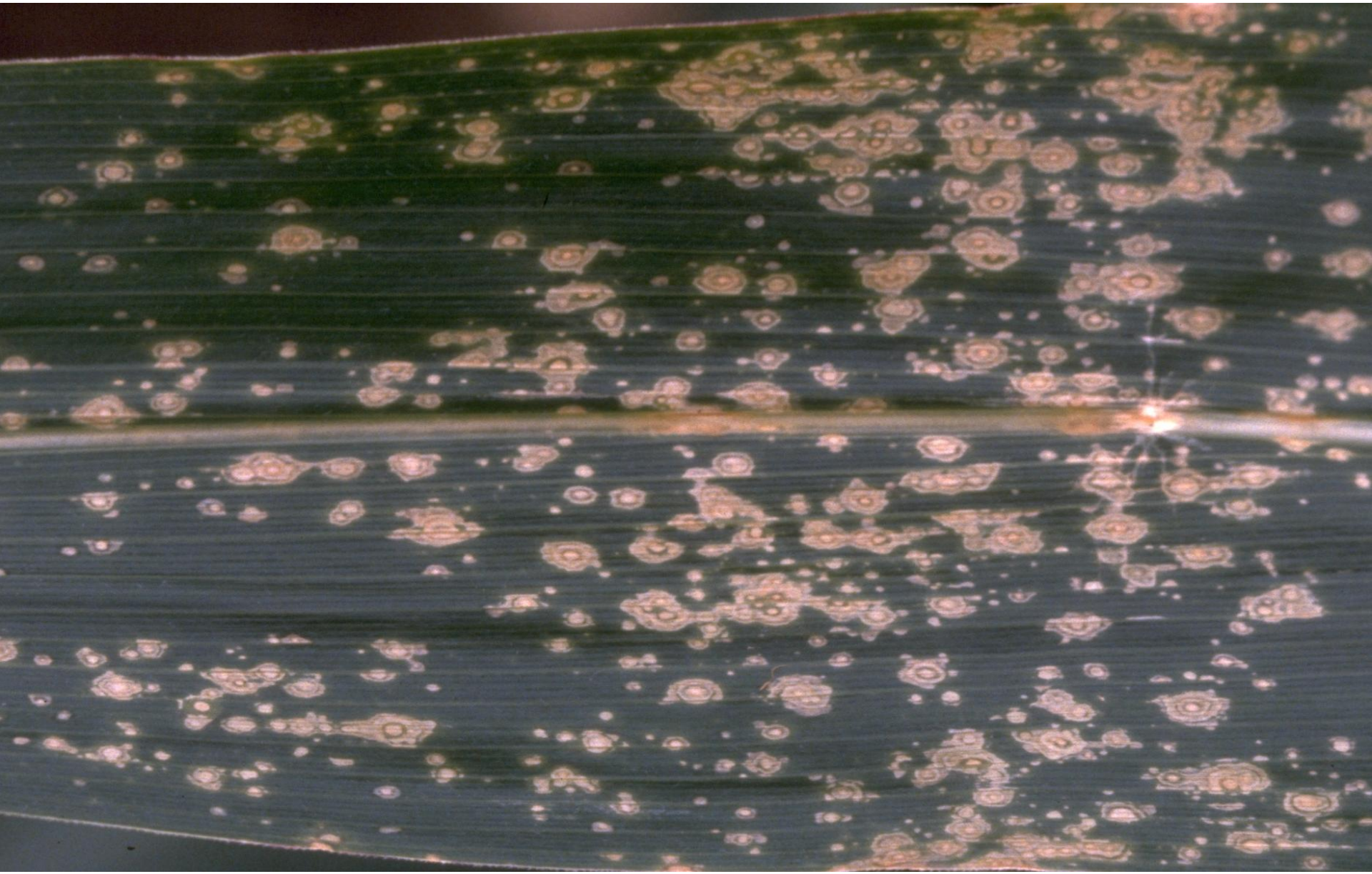
Leaf of a Les1-843/+ mutant plant showing dramatic lesion distribution in rings and target spots, resulting from growth in winter greenhouse conditions

Les2-845A/+ leaf, under favorable conditions, showing unusual clustering of small round white necrotic lesions

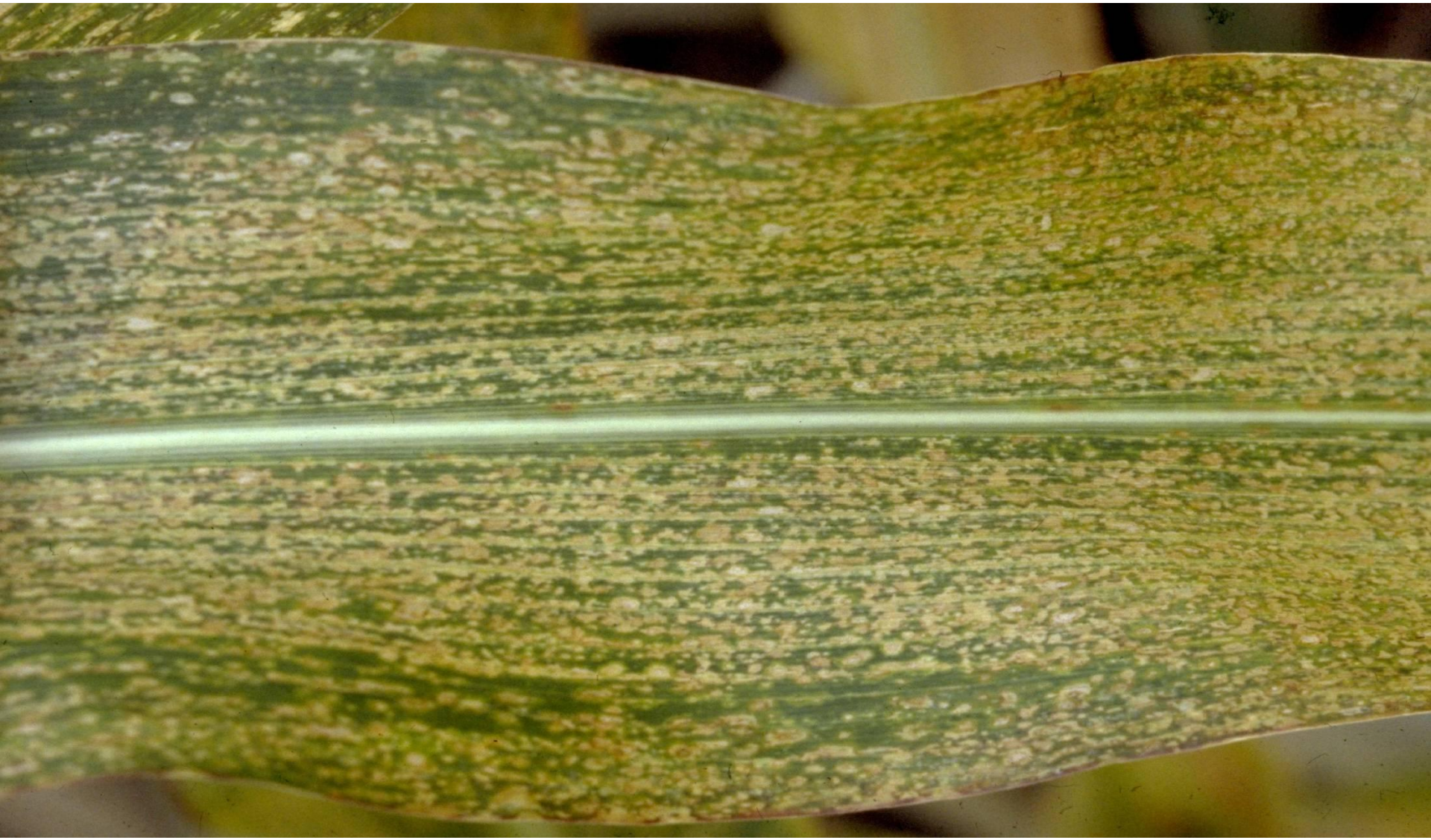
8th leaf of Les10-A607 heterozygous plant, (Mo20W/W23 background) showing profuse tiny to small necrotic lesions

8th leaf of a Les3-A781 homozygous plant, Mo20W background, showing large round white necrotic lesions on dark green leaves (photo courtesy D. Hoisington)

8th leaf of Les*-N1378/+ plant, showing tiny to medium-sized necrotic lesions with white center and brown and tan concentric rings.

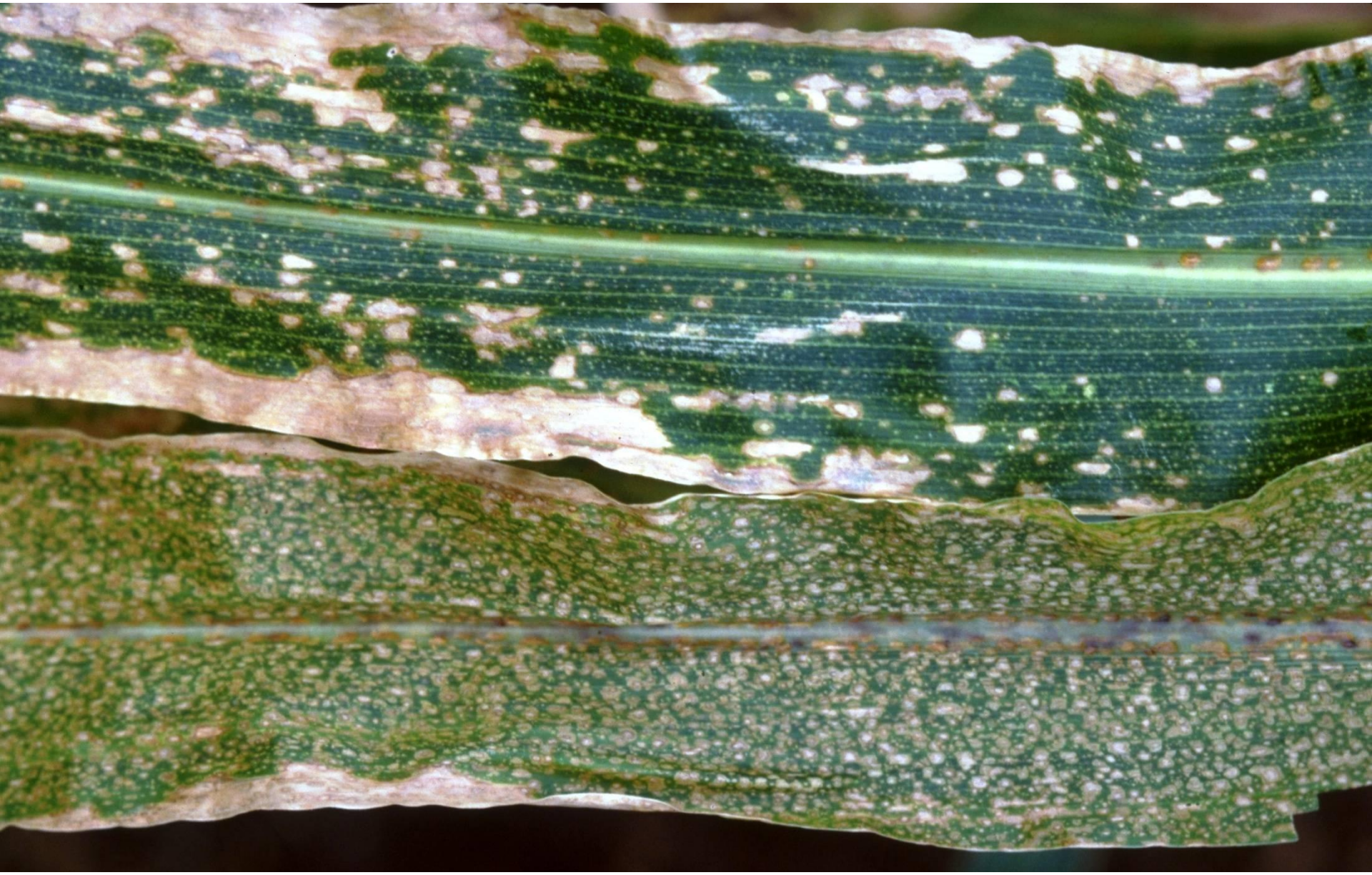


section of the eighth leaf of a Les11/+ plant showing many small chlorotic and necrotic lesions.



Eigth leaf orginal LesGr*-N2639/+ mutant plant showing profuse tiny chlorotic lesions especially along veins giving a grainy striped appearance.

8th leaf section of Les10-A607/+ heterozygous plants, Mo20W (top) and M14/W23 (bottom) backgrounds, for comparison of respective genetic modifiers



Les Mutant Phenotypes

- **Les mutants: total of 51 dominant mutants from approximately 50,000 M1's screened. One possible allelic pair among 22 tested leads to an estimated 200+ loci. Must be important in resistance.**
  - **Phenotypic variables to consider: size shape color texture and lesion profile.**
  - **frequency timing distribution and developmental patterns.**
  - **response to genetic and environment modification**
  - **relationship to actual disease symptoms and resistance**

Attention drawn to possible developmental relationships exhibited by certain "patterns of lesions" and the implication of "signaling between cell groups".

Propheria in humans and Les22 have same DNA Sequence.

- **Les1, Les2,Les3,Les10,Les-*1378, etc lesion types**
- **Les1 target spot: model for cell death signal in Maize (slide 18)?**
- **Cell death signal in mice**
- **Ring of brown aging skin spots on forehead of 90 year old human**

Ring of Brown Aging Skin Spots on Human Forehead