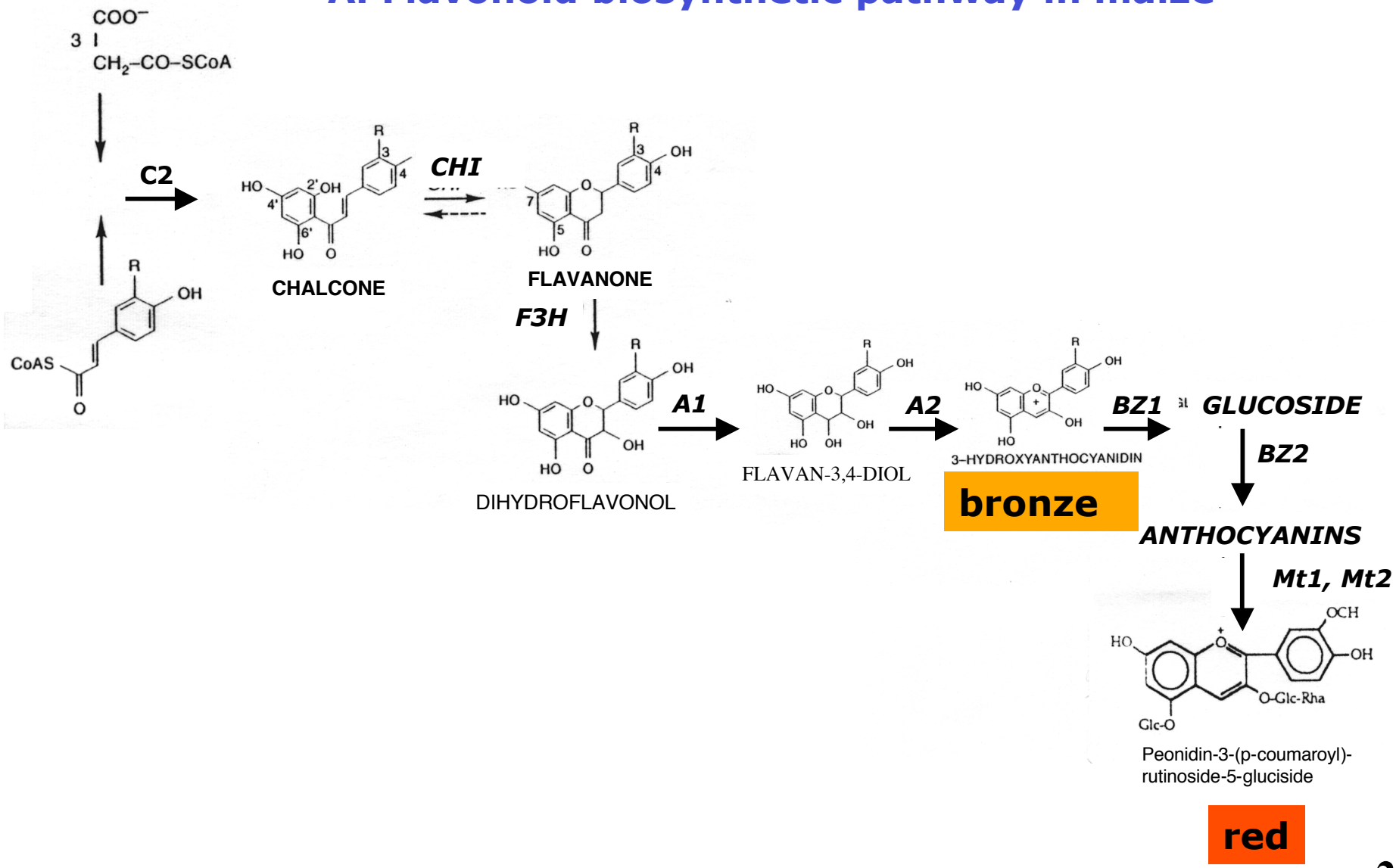
Lecture 5: Genetic interactions and epistasis

A. Epistasis in a biochemical pathway
B. Epistasis in a regulatory pathway
C. Additive interactions
D. Synergistic interactions
E. Suppressions

epistasis analyses (genetic interactions among different mutations)

A. Flavonoid biosynthetic pathway in maize



WT:	**Red**
Mutations in c2, a1, a2:	**Colorless**
Mutations in bz1, bz2:	**bronze**

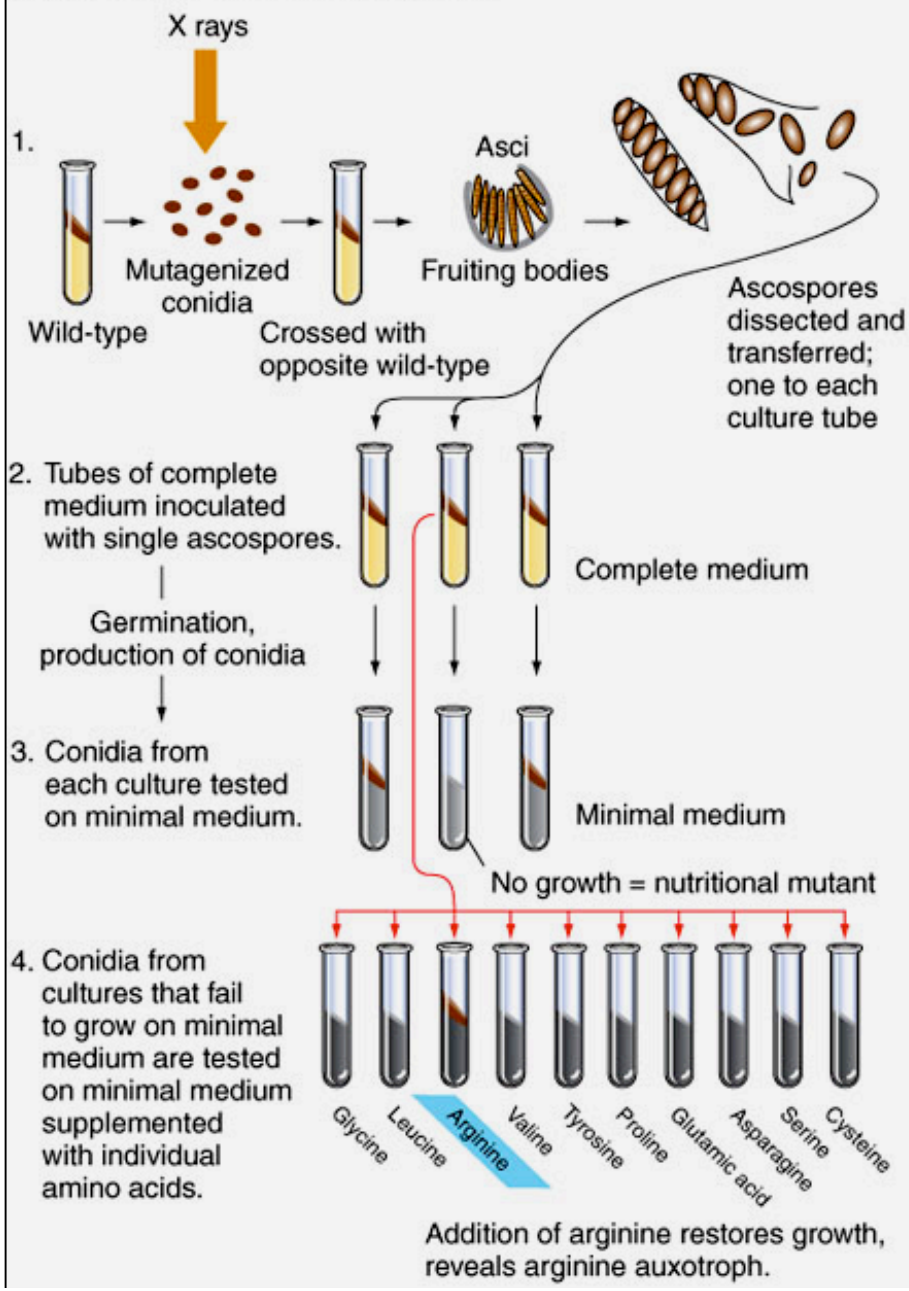
Double mutants

C2/a1: colourless-but uninformative
bz1/a1: colorless-a1 comes before bz1
bz2/a1: colorless-a1 comes before bz2

For biosynthetic pathways, the phenotype of the earlier gene in the pathway shows in the double mutant.
ie. the earlier-step mutant is epistatic to the late-step mutant

Determine relationship between a1 and c2 by feeding experiment:
add flavanone (naringenin): c2+naringenin = red
a1+naringenin = colorless

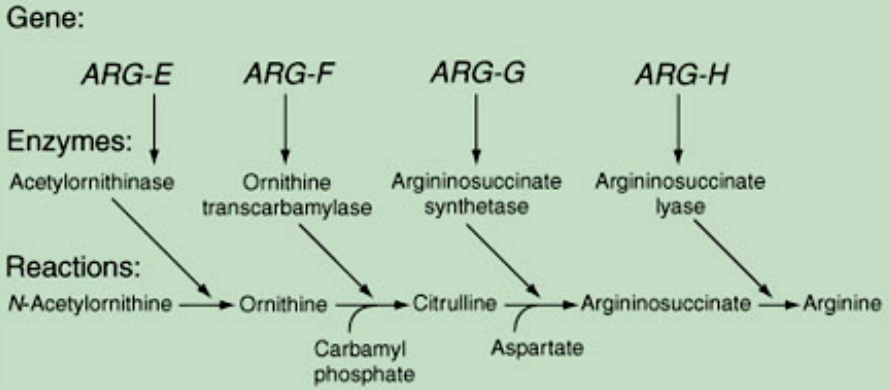
Fig. 7.20 Biochemical Pathways



(b) Growth response if nutrient is added to minimal medium

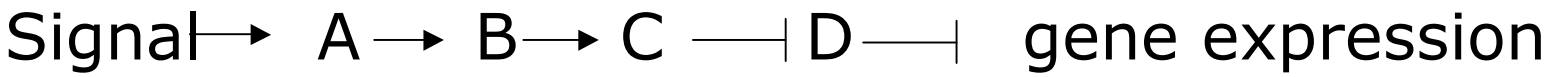
Mutant strain	Supplements: Nothing	Ornithine	Citrulline	Arginino-succinate	Arginine
Wildtype: *Arg*$^+$	+	+	+	+	+
Arg-E$^-$	–	+	+	+	+
Arg-F$^-$	–	–	+	+	+
Arg-G$^-$	–	–	–	+	+
Arg-H$^-$	–	–	–	–	+

(c) Inferred biochemical pathway



See p214-215 of the textbook

B. Regulatory pathways



→ Positive action-stimulate next step.
Null mutation makes insensitive to signal

⊣ Negative action-represses next step.
Null mutation makes the gene turned on at all time (constitutively)

b^-: gene expression never turned on
even in the presence of the signal

d^-: gene expression constitutively on
even in the absence of signal

$b^-d^- = d^-$: constitutively on

For regulatory pathways, the phenotype of the later-acting genes shows in the double mutant.
ie. the later-acting mutant is epistatic to the earlier-acting mutant

wt wt *ctr1 ein2*

ethylene air air ethylene

ctr ein2 :?

Ethylene ⊣ CTR1 (Kinase) ⊣ EIN2 → triple response

For regulatory pathways, the phenotype of the later-acting genes shows in the double mutant.

ie. the later-acting mutant is epistatic to the earlier-acting mutant

C. Additive pathways

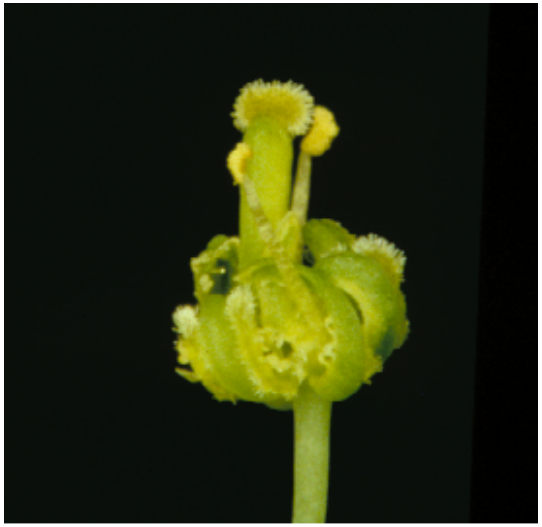
Double mutants of dissimilar phenotypes produce a combination of both phenotypes

Indicate that the two mutations are in genes acting in separate pathways

ap2-2 (flower abnormal) X *gl* (no trichome)

ap2-2 gl double mutant
abnormal flower and no trichome

ap2-2



gl1

D. Synergistic interactions (enhancement)

Two genes may act at the same step of a pathway

Or in parallel or (redundant) pathways

E. Suppression

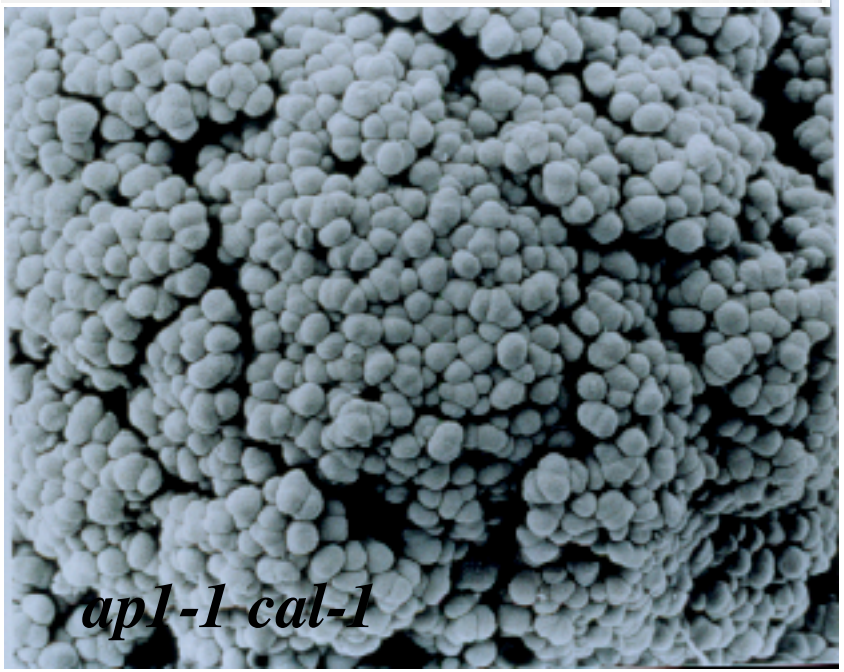
Intrgenic suppressors

Extragenic suppressors


ap1-1


WT


ap1-1 cal-1


ap1-1 cal-1

E. Suppression

Intrgenic suppressors

Extragenic suppressors

Allele-specific suppression