Chapter 1: Basic Genetics

Gene & Allele

Phenotype & Genotype

Ploidy & Allelism

Molecular Biology (Molecular Genetics) is simple?

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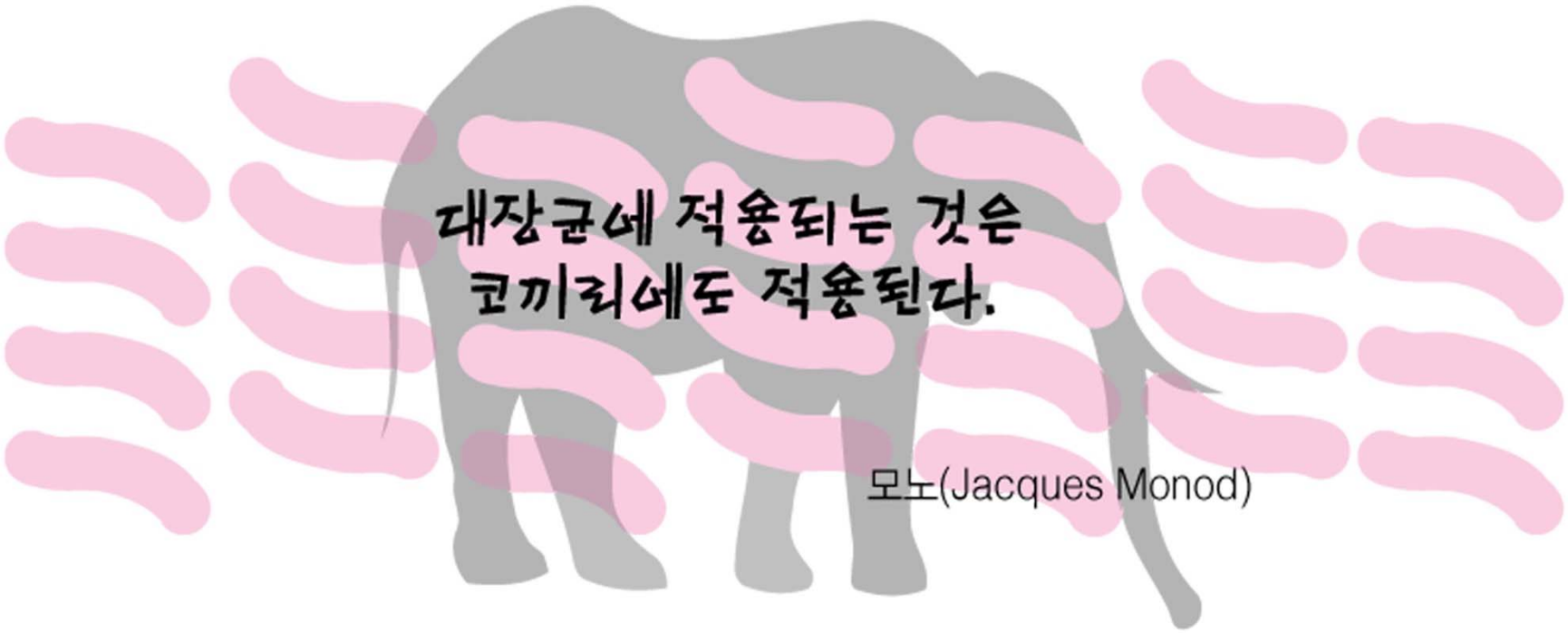
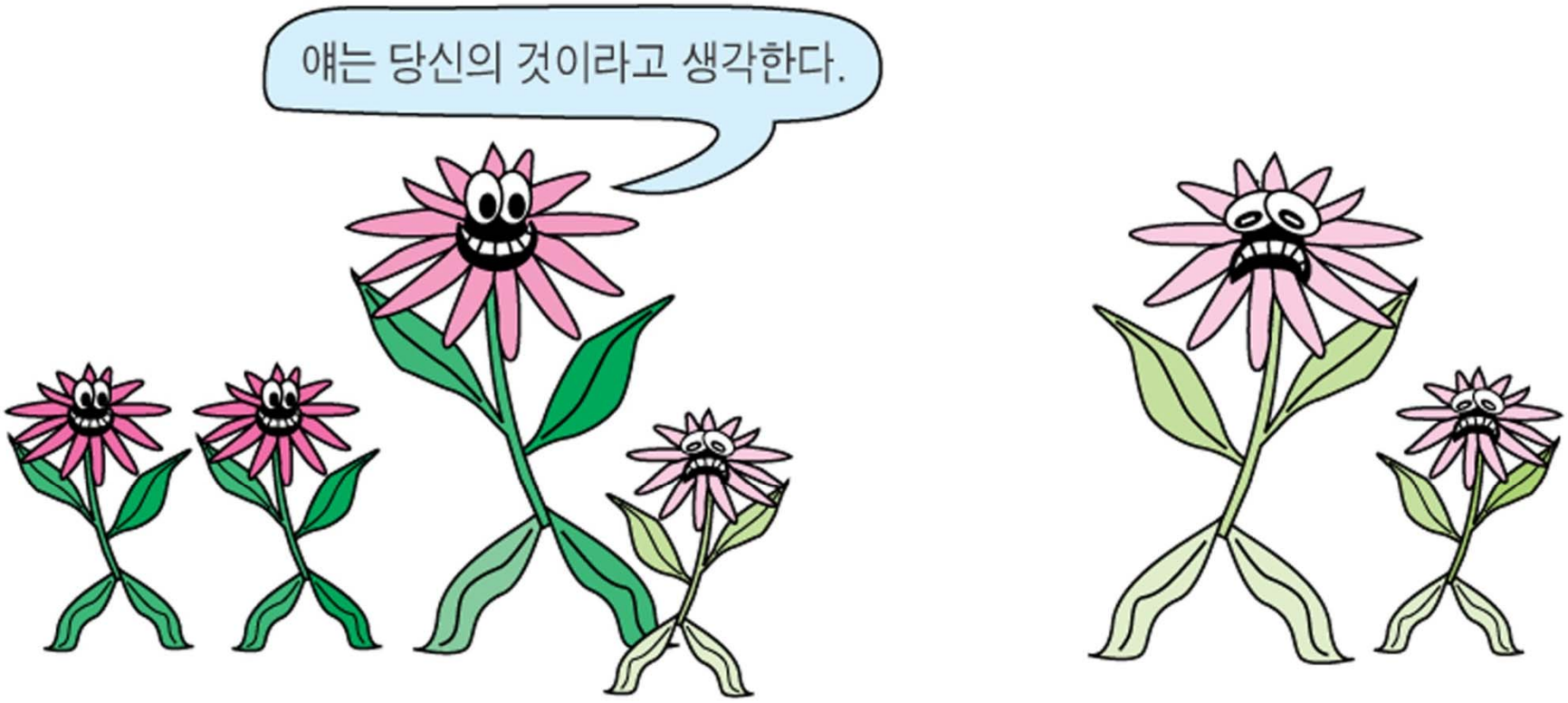


What does determine the similarity and difference?



Gene & Allele

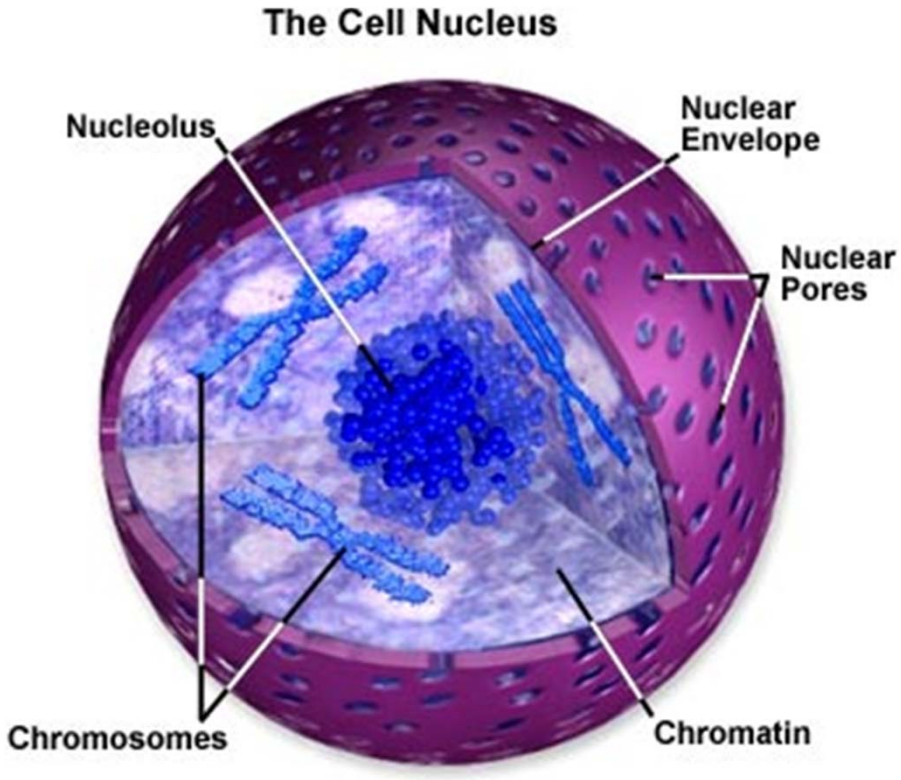
- The birth of modern genetics was due to the discoveries of **Gregor Mendel**
- Each of the characteristics examined by Mendel is determined by a **single gene** (gene = a unit of genetic information)
- Each gene may exist in alternative forms, called **alleles** (allele = one particular version of a gene)

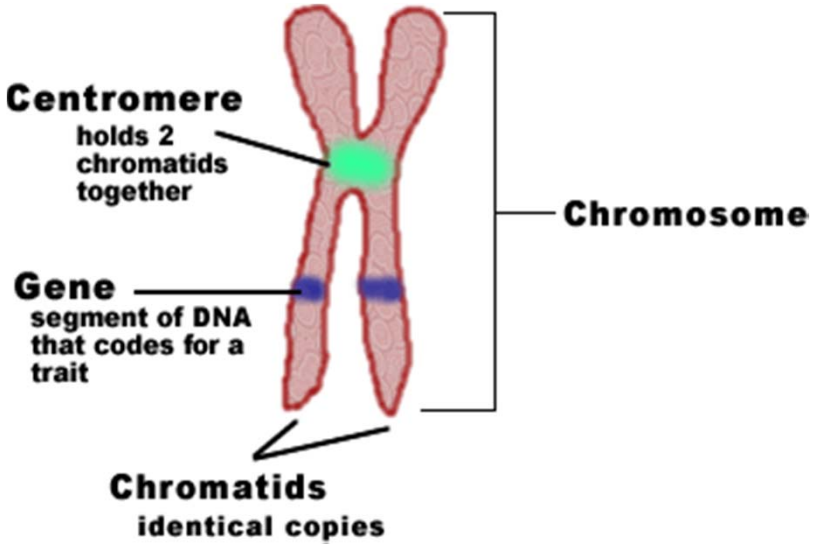
Questions!

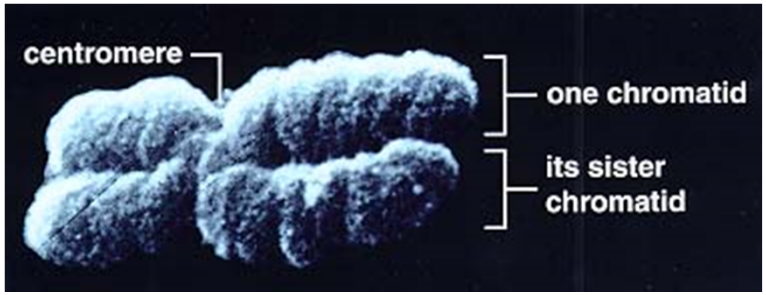
- **How did Mendel describe the genes and their alleles?**
- **How many alleles for a gene exist in our classroom?**
- **How many different alleles for a gene exist in our classroom?**

A, a, A1, A2, A3……

Chromosomal basis of genetic inheritance







Genes are also found in cytoplasm organelles

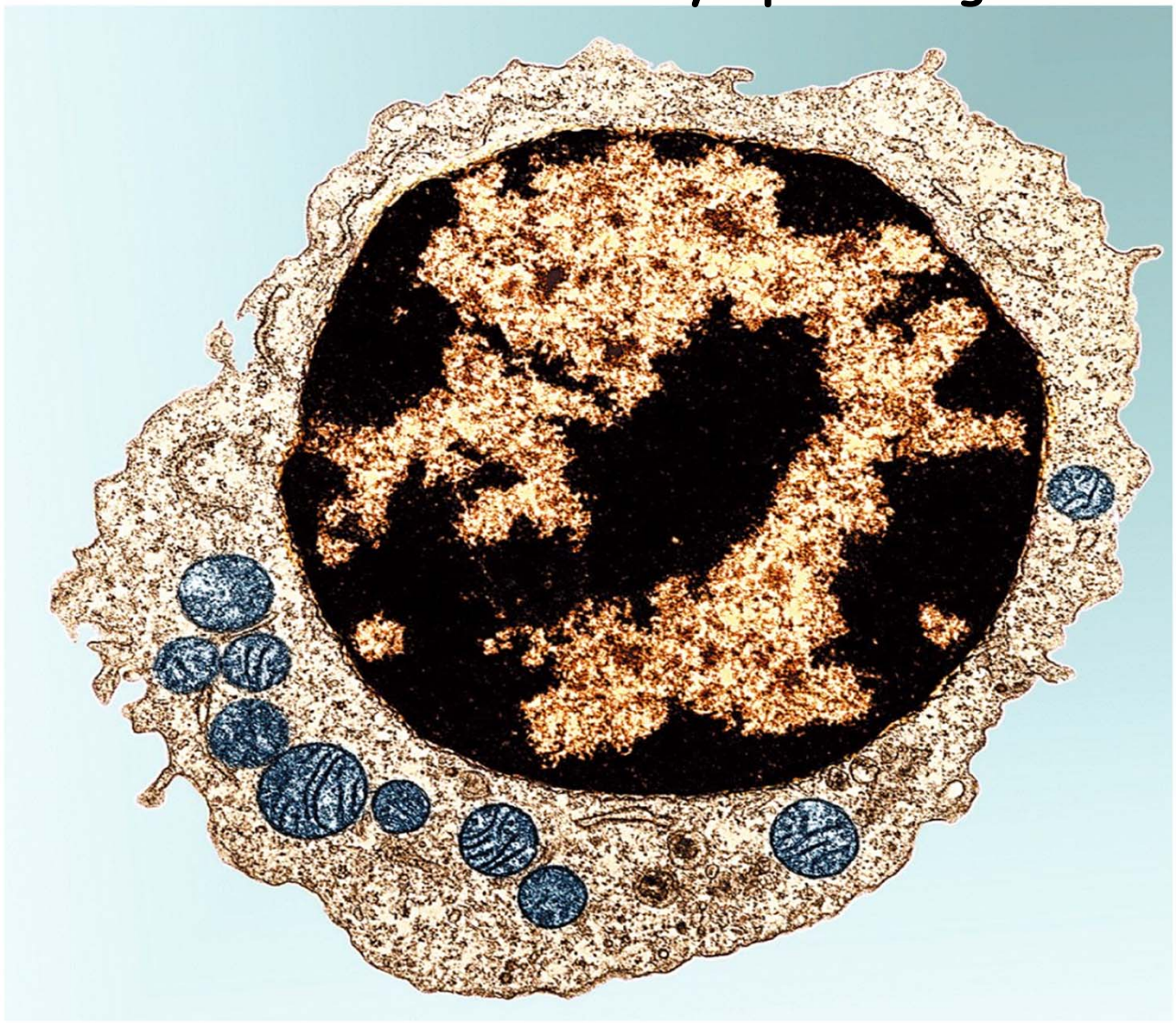
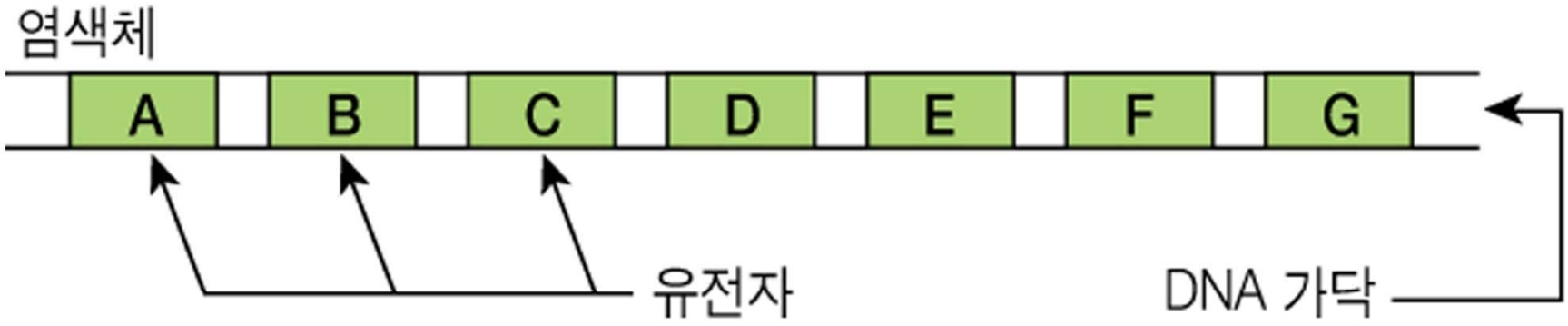


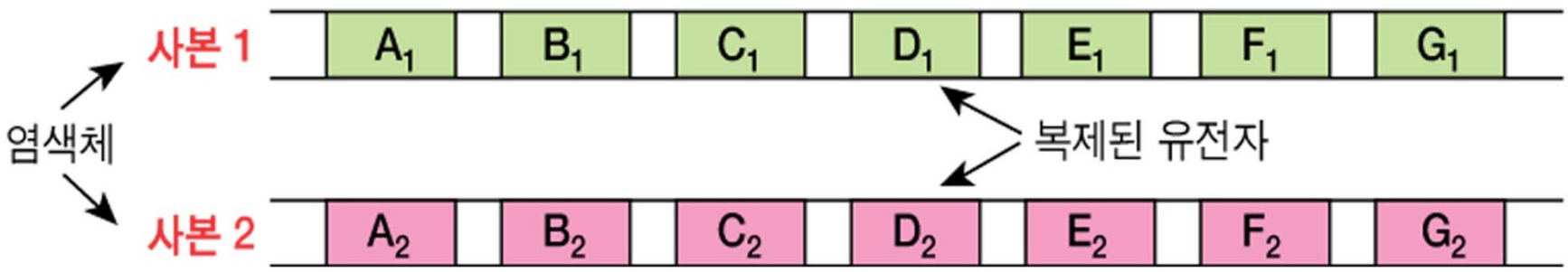
Figure 2-1b Principles of Genetics, 4/e

eukaryotic cells typically contain one or more **mitochondria**
Light colored material: chromosomes

Genes in a chromosome



Genes(alleles) in homologous chromosomes of diploid



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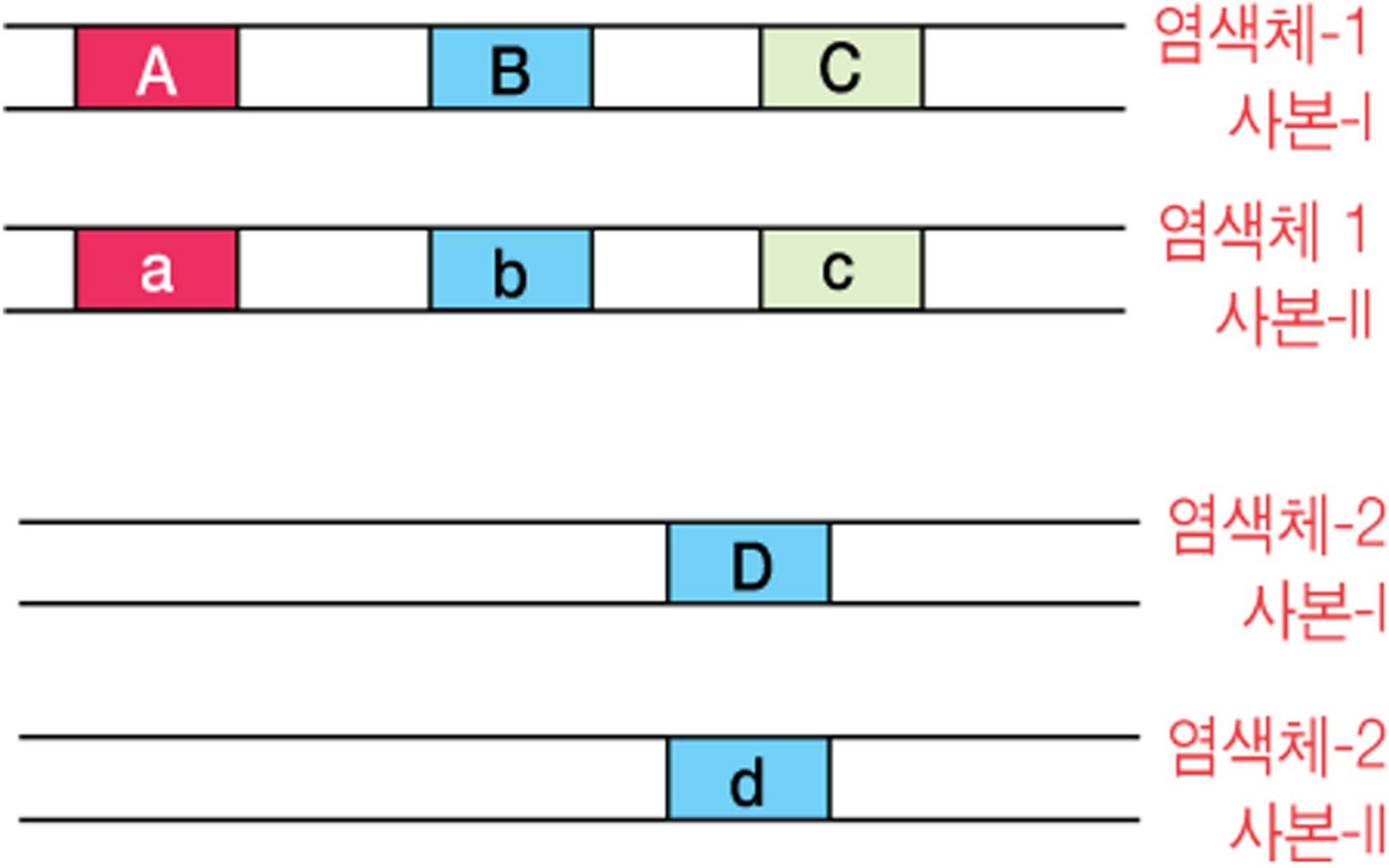
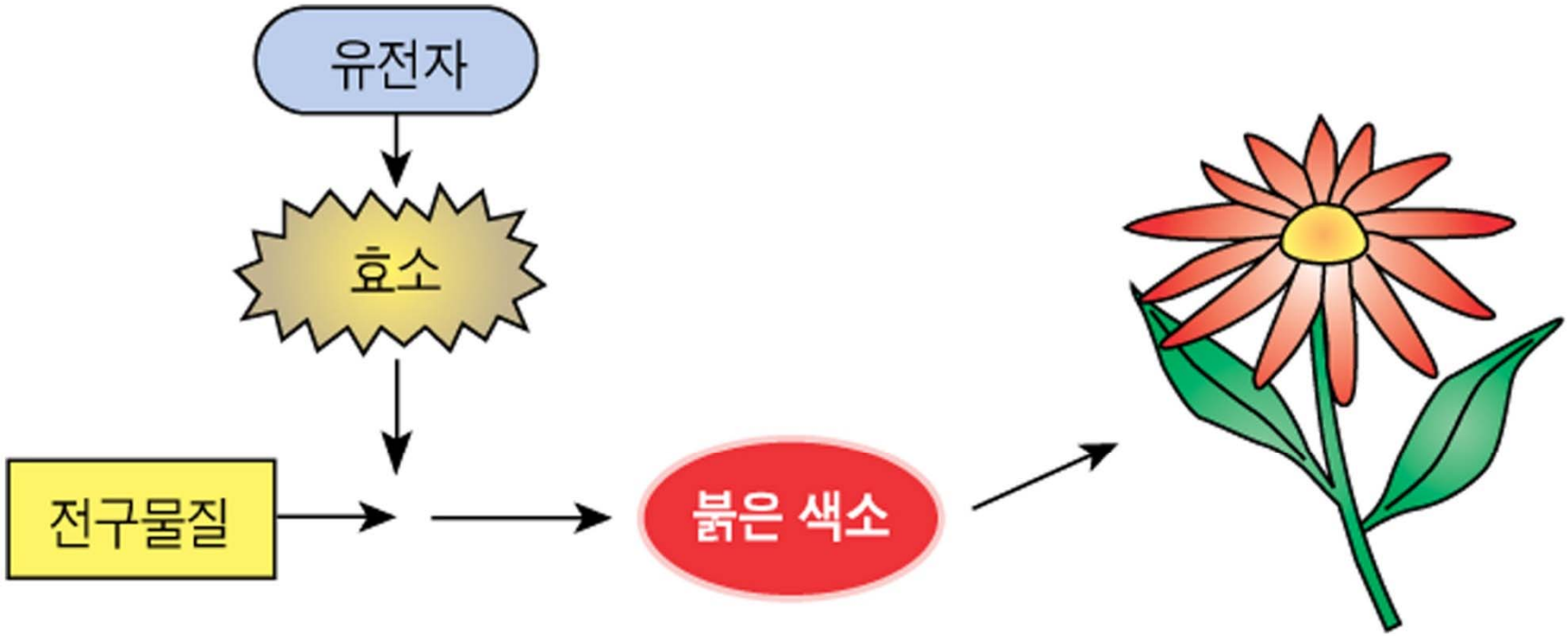


What is the function of genes?

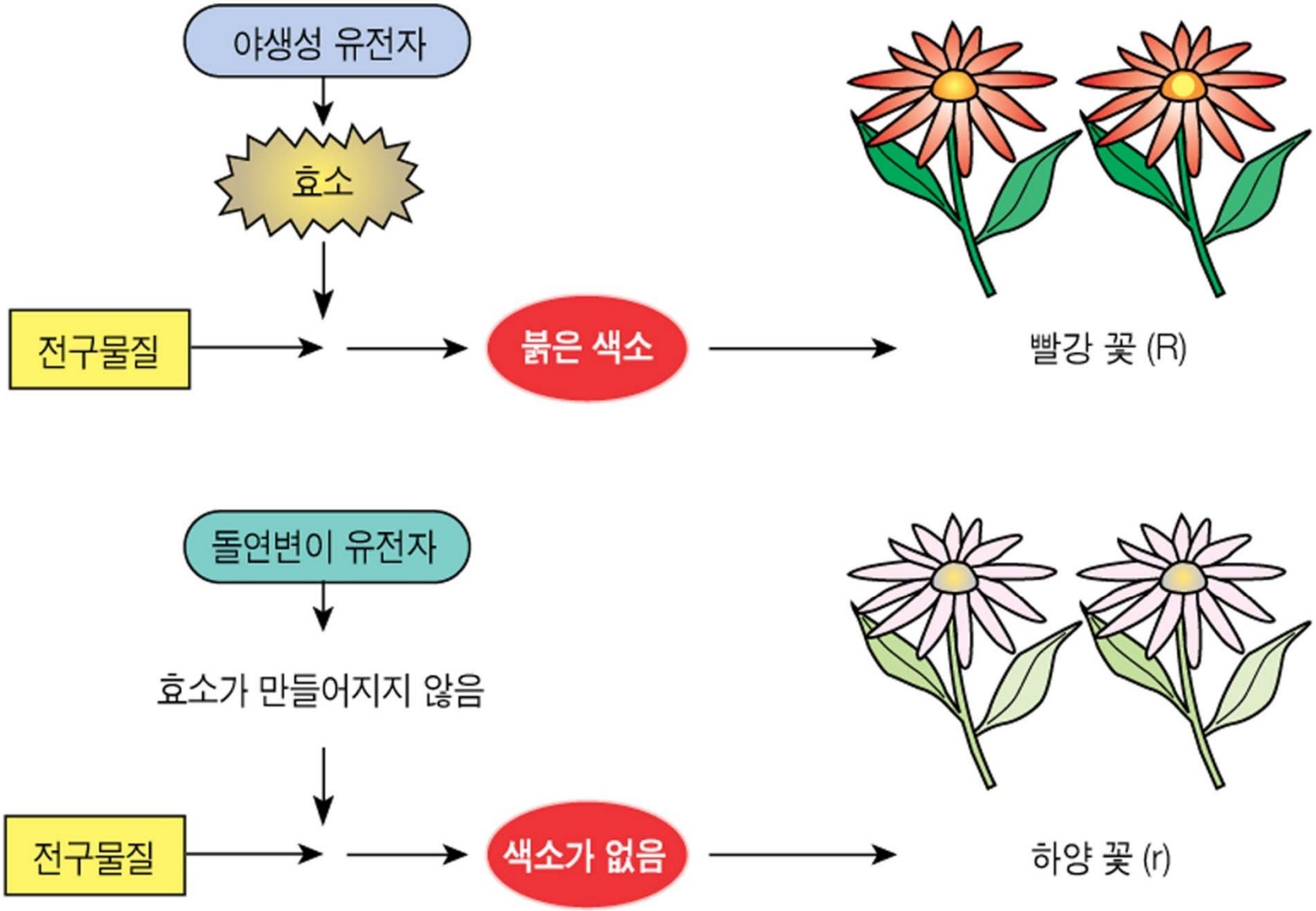
- Each step of a biosynthetic pathway is controlled by a single gene
- Each biosynthetic reaction is carried out by a special protein known as enzyme, produced by a single gene (*one gene – one enzyme* model)
- The properly functioning gene is referred to as the wild-type allele, and defective gene as the mutant allele (genetic alteration)
- A mutant allele that results in the complete absence of an enzyme is known as a null allele

A genes encodes an enzyme playing a role in a biochemical pathway

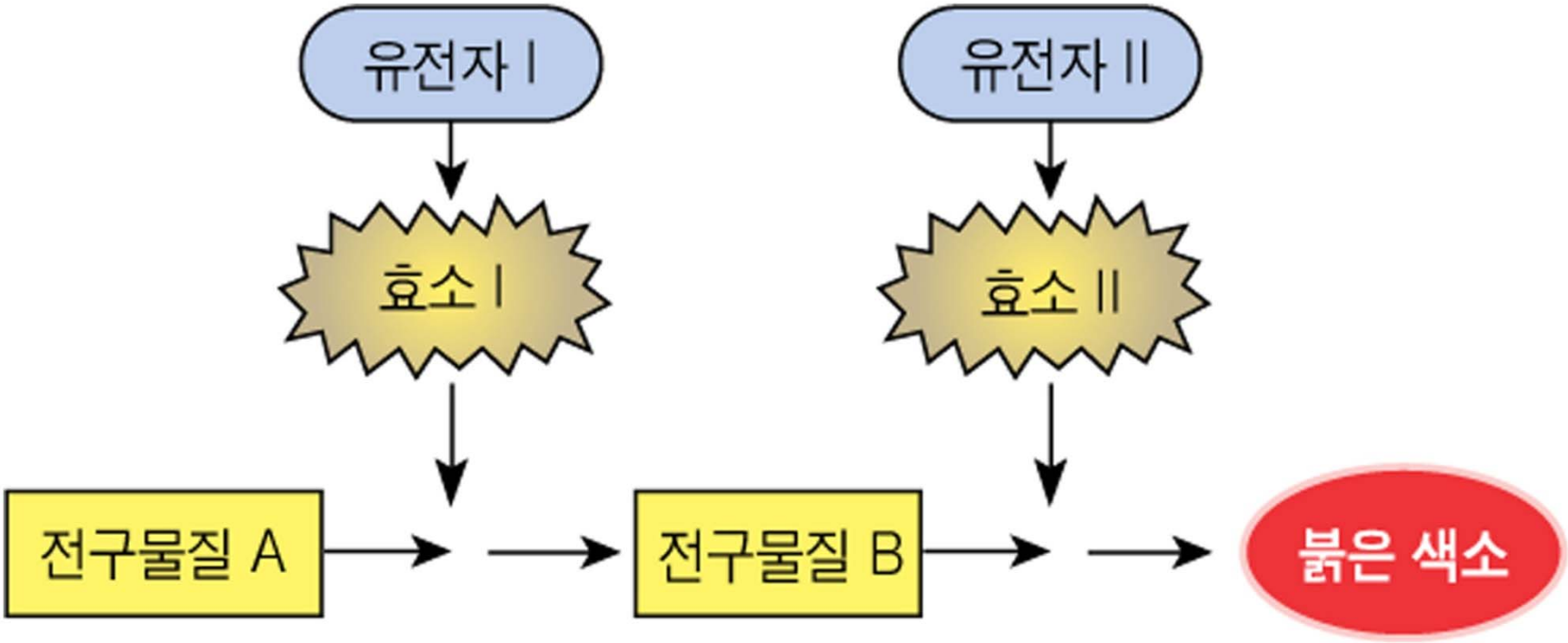


Example of a pathway for red pigment synthesis

Wild-type allele & mutant allele

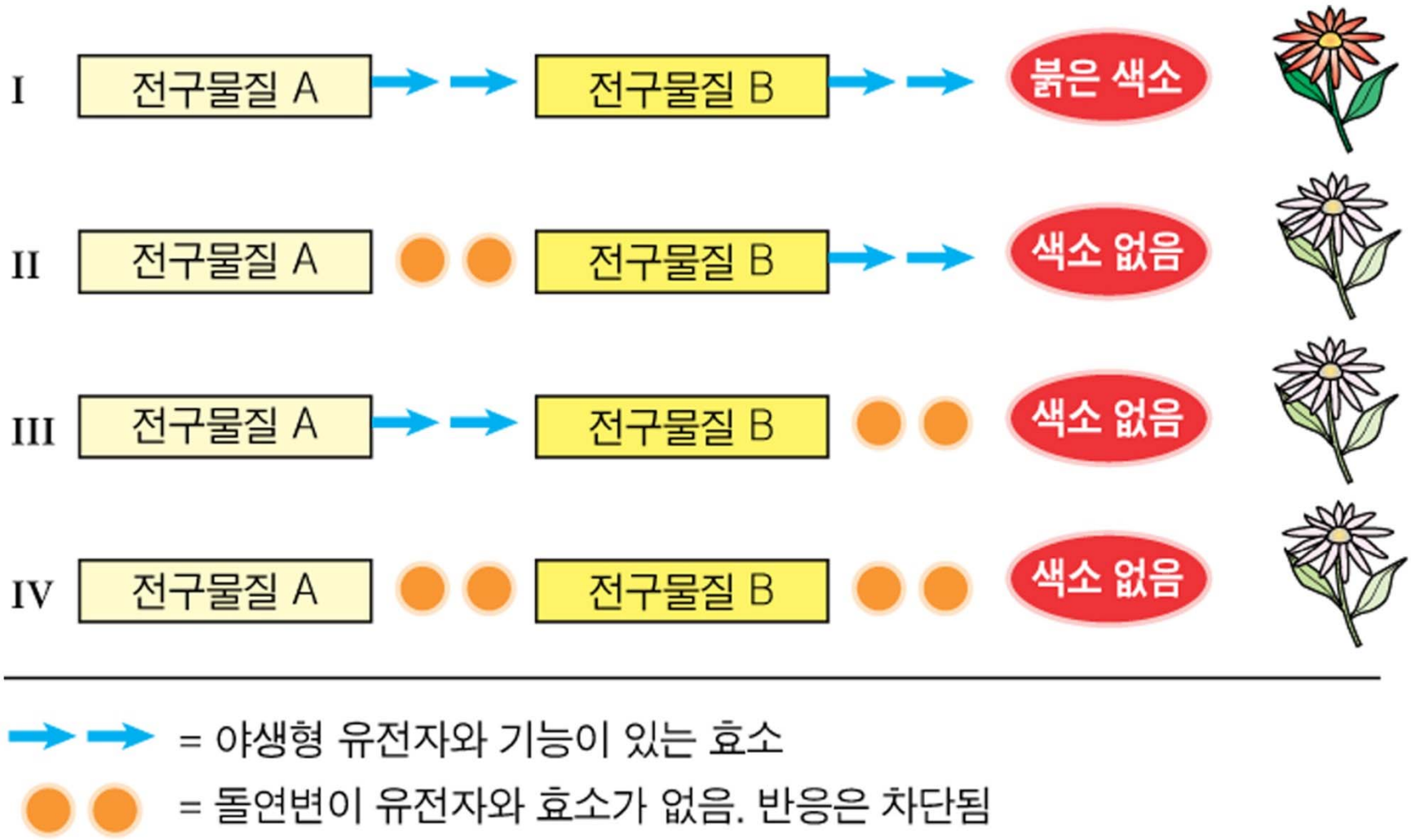


One gene – One enzyme model



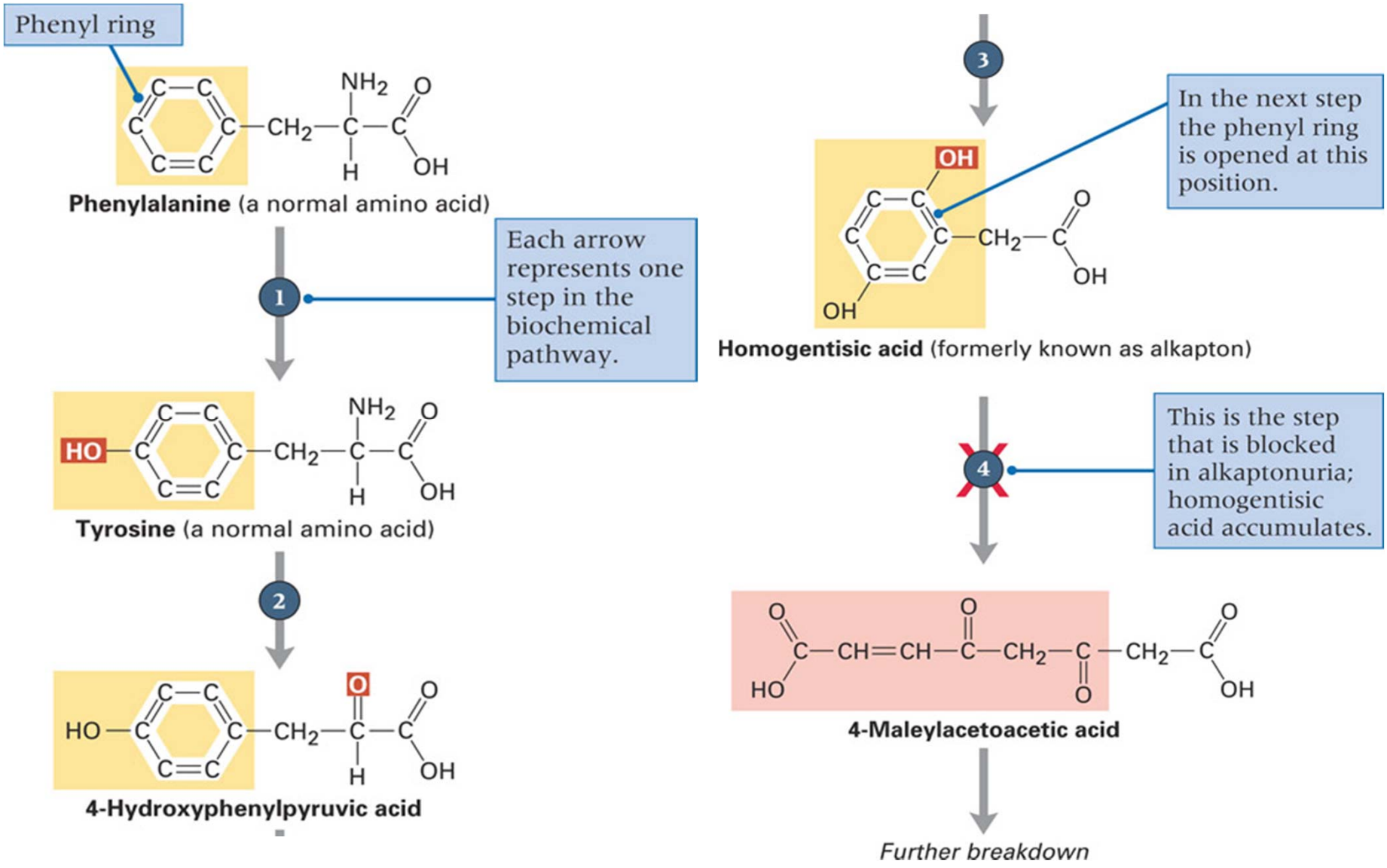
Each step of a metabolic pathway requires an enzyme encoded by a gene

One gene – One enzyme model and epistasis

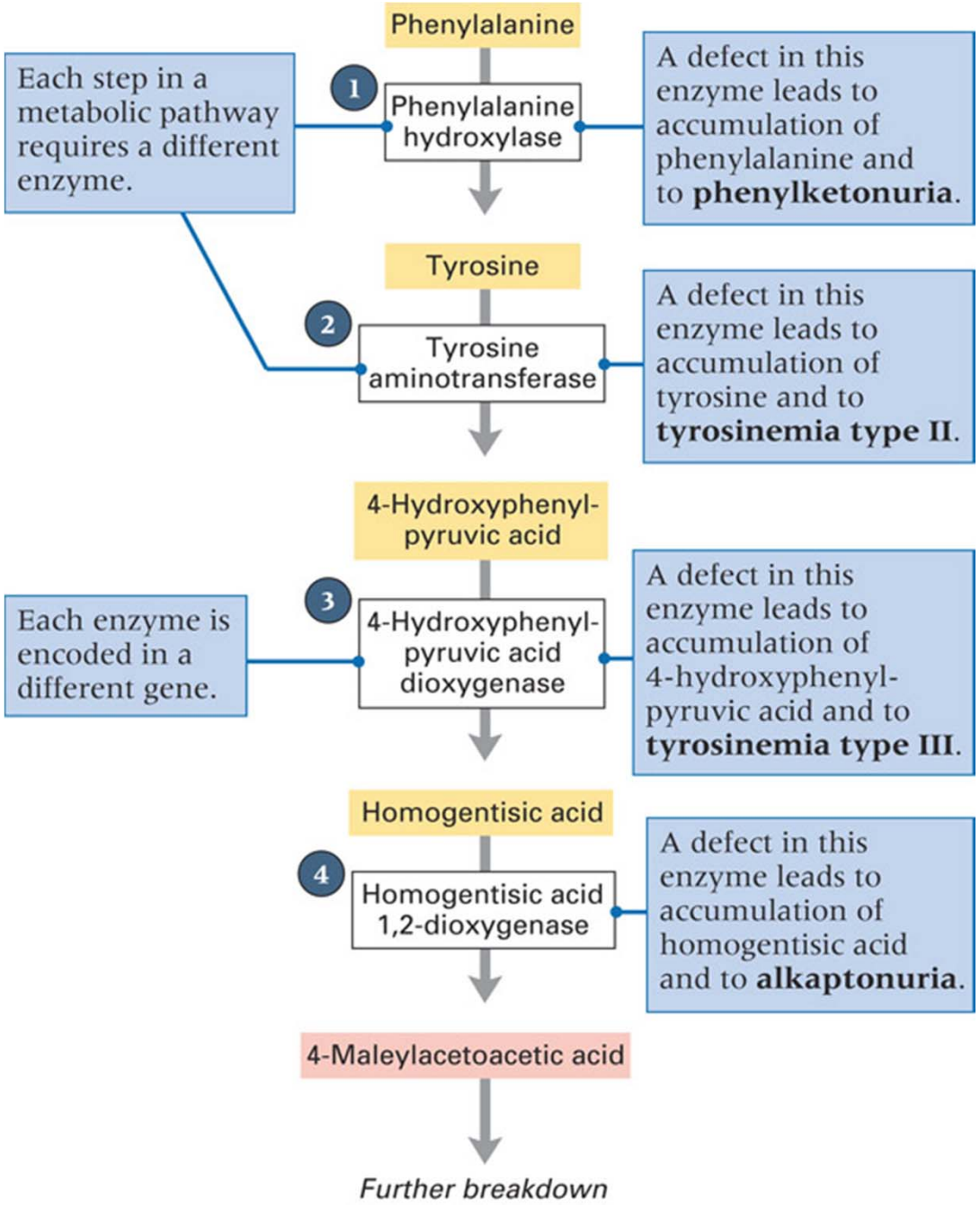


A classical example of one gene-one enzyme in human disease

Inborn errors of metabolism: Alkaptonuria (A black urine disease)

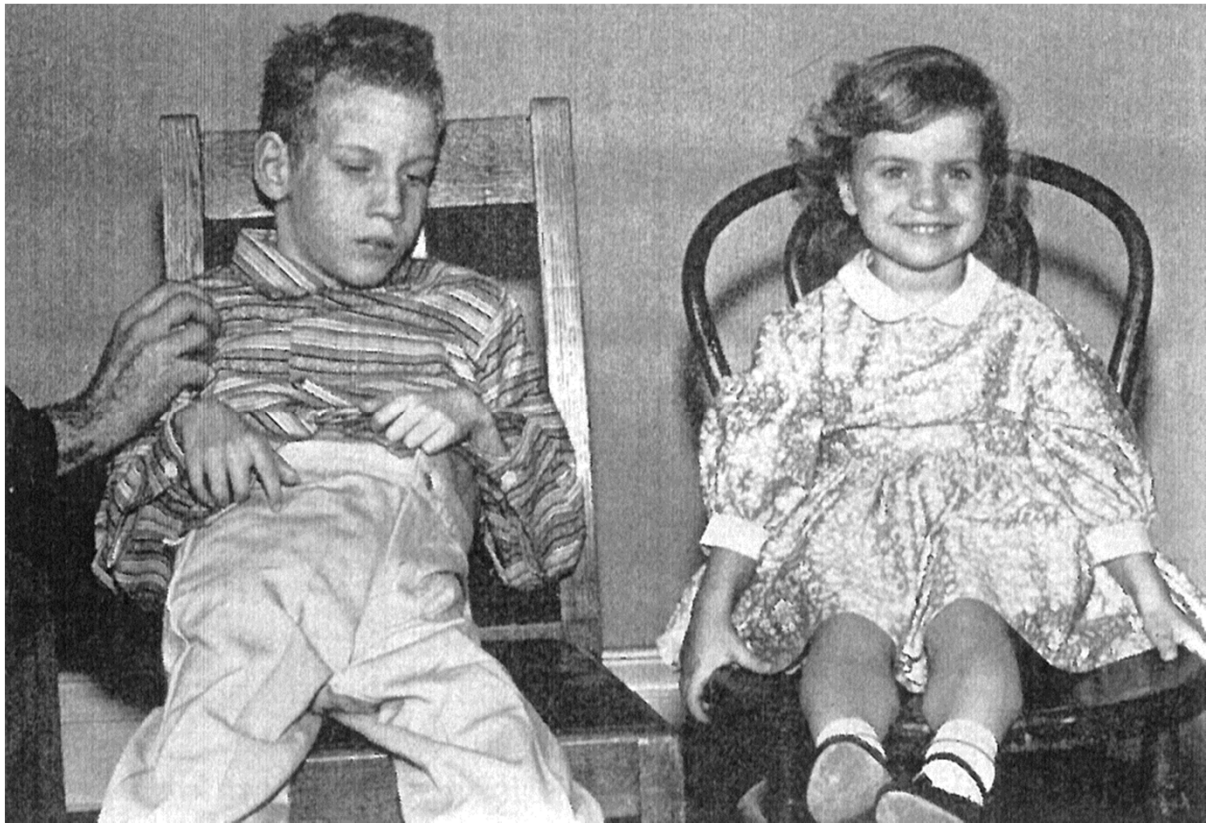


Metabolic pathway for the breakdown of phenylalanine and tyrosine



One gene-one enzyme model in a biochemical pathway

Phenylketonuria (PKU)



Phenotype & Genotype

- Most biochemical pathways have several steps, not just one.
- The outward characteristics at the final step of the a biochemical pathway is referred to as the phenotype.
- The genetic make-up of the phenotype is referred to as the genotype, and the phenotype is a visible effect of the genotype.
- A defective gene (mutant allele) near the beginning of a pathway will make later reaction irrelevant by making the effect of alterations in another gene (epistasis)

Ploidy and Allelism

- Genes consist of DNA and the genes belonging to each cell are arranged on chromosomes, a giant molecules of DNA

- Genes are a portion of a chromosome

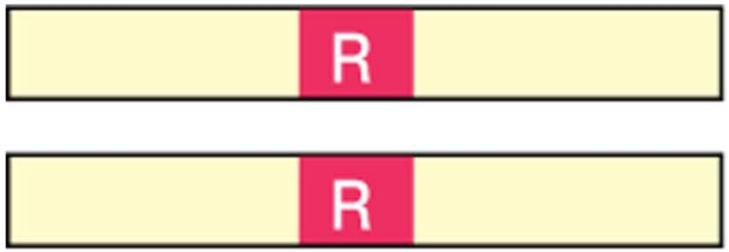
- Lower organisms have only a single copy (one allele) of each gene (haploid), but higher organisms have duplicate copies (two alleles) of each gene (diploid)

Ploidy and Allelism

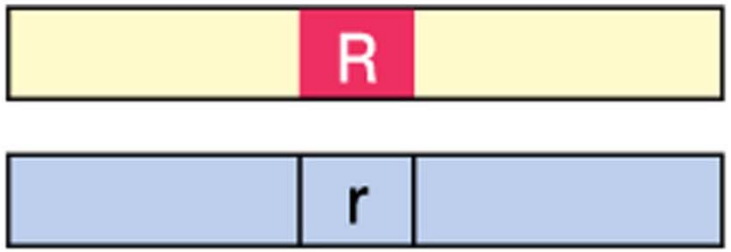
- The allele whose properties are expressed as the phenotype is called dominant allele, and the allele masked its expression by dominant allele is called recessive allele

- If you have two identical alleles of the same gene, you are homozygous for that gene, if you have to different alleles, you are heterozygous for that gene

Allelism and dominance effect on the flower color phenotype

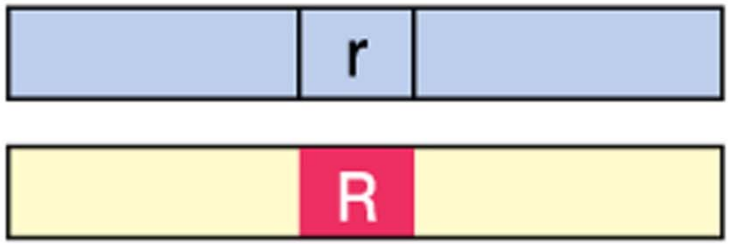


우성의 동형접합
(RR = 빨강 꽃)



이형접합
(Rr= 빨강 꽃)

열성의 동형접합
(rr = 하양 꽃)



이형접합
(rR = 빨강 꽃)

Homologous chromosomes in human: each chromosome contains an allele for a gene(locus)

Figure 6-4c Principles of Genetics, 4/e

TABLE 3.3

Inherited Conditions in Human Beings
Dominant Traits
Achondroplasia (dwarfism)
Brachydactyly (short fingers)
Congenital night blindness
Ehler-Danlos syndrome (a connective tissue disorder)
Huntington's disease (a neurological disorder)
Marfan syndrome (tall, gangly stature)
Neurofibromatosis (tumorlike growths on the body)
Phenylthiocarbamide (PTC) tasting
Widow's peak
Woolly hair
Recessive Traits
Albinism (lack of pigment)
Alkaptonuria (a disorder of amino acid metabolism)
Ataxia telangiectasia (a neurological disorder)
Cystic fibrosis (a respiratory disorder)
Duchenne muscular dystrophy
Galactosemia (a disorder of carbohydrate metabolism)
Glycogen storage disease
Phenylketonuria (a disorder of amino acid metabolism)
Sickle-cell anemia (a hemoglobin disorder)
Tay-Sachs disease (a lipid storage disorder)

Table 3-3 Principles of Genetics, 4/e

Phenotypes controlled by single gene in human

Achondroplasia (dwarfism)

Dominant allele

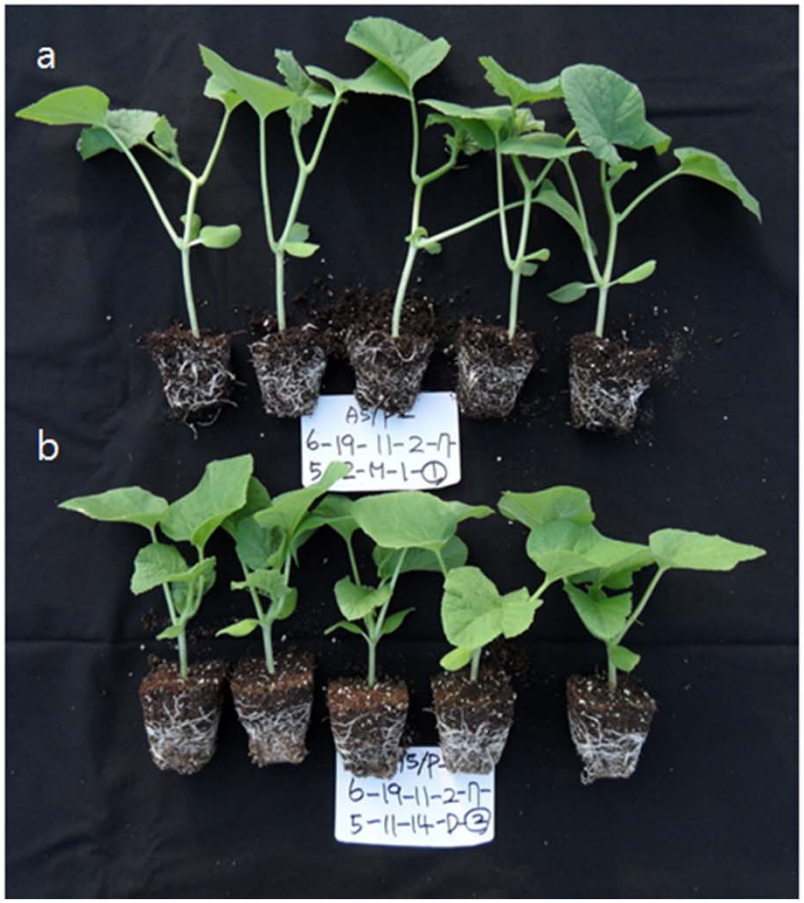
Albinism (lack of pigment)

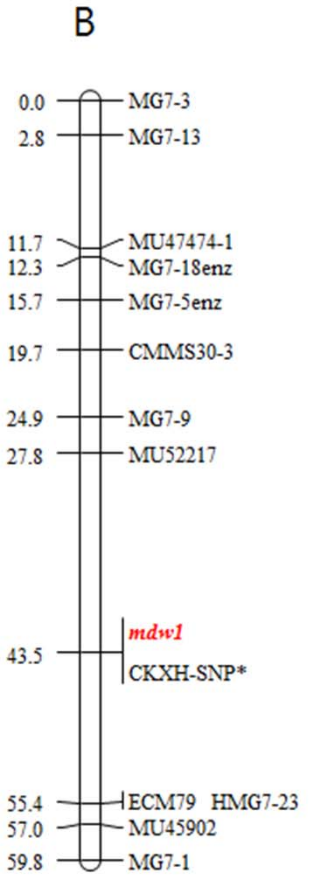
Recessive allele

Many important crops are polyploidy

Figure 6-6 Principles of Genetics, 4/e

CKX(cytokine oxidase) gene could controls dwarfism in melon





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