The importance of learning English for biology study

英英字典

www.dictionary.com

gene - 7 dictionary results

每得科技-DNA extracti
Multi,Micro,Midi, Macro 您所需要的DNA
www.medclub.com.tw
Sponsored Results

真人發音

gene [jeen] ? Show IPA

–*noun*
the basic physical unit of heredity; a linear sequence of nucleotides along a segment of DNA that provides the coded instructions for synthesis of RNA, which, when translated into protein, leads to the expression of hereditary character.

Origin:
1911; < G *Gen* (1909), appar. abstracted from *-gen* -GEN; introduced by Danish geneticist Wilhelm L. Johannsen (1857-1927)

每得科技-DNA extraction
Multi,Micro,Midi, Macro Abgarose 有效而清楚地分離出您所需要的DNA
www.medclub.com.tw
Sponsored Results

Gene [jeen] ? Show IPA

–*noun*
a male given name, form of EUGENE.

Dictionary.com Unabridged
Based on the Random House Dictionary, © Random House, Inc. 2009.
Cite This Source | Link To gene

專有名詞

www.onelook.com

General (27 matching dictionaries)

1. gene: Compact Oxford English Dictionary [home, info]
2. gene: American Heritage Dictionary of the English Language [home, info]
3. -gene, gene: Encarta® World English Dictionary, North American Edition [home, info]
4. gene: Merriam-Webster's Online Dictionary, 11th Edition [home, info]
5. gene: Cambridge International Dictionary of English [home, info]
6. Gene: Wiktionary [home, info]
7. gene: Webster's New World College Dictionary, 4th Ed. [home, info]
8. gene: The Wordsmyth English Dictionary-Thesaurus [home, info]
9. gene: Infoplease Dictionary [home, info]
10. -gene, gene: Dictionary.com [home, info]
11. gene: Online Etymology Dictionary [home, info]
12. Gene, gene: UltraLingua English Dictionary [home, info]

英文的發音

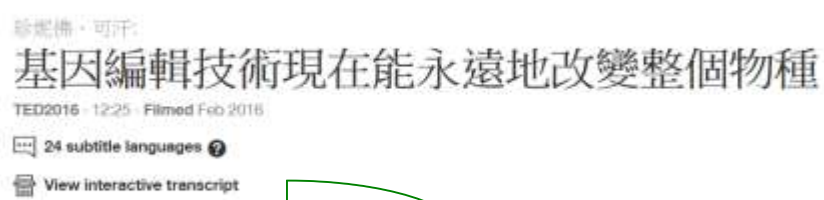
www.howjsay.com

TEDxTalk

CRISPR 基因編輯技術可以讓科學家改變 DNA 序列，並保證所得到的編輯遺傳特徵可以被未來的後代所遺傳，這項技術開闢了永遠改變整個物種可能性的大門。

更重要的是，該技術引發了新的問題：這項新技術將如何影響人類？我們將用它來改變甚麼呢？我們現在是上帝了嗎？

讓我們一起與記者珍妮佛．可汗（Jennifer Kahn）思考這些問題，並聽她分享基因編輯技術的潛在應用方式：根除瘧疾和Zika病毒的抗病基改蚊的發展。



[Link](#)

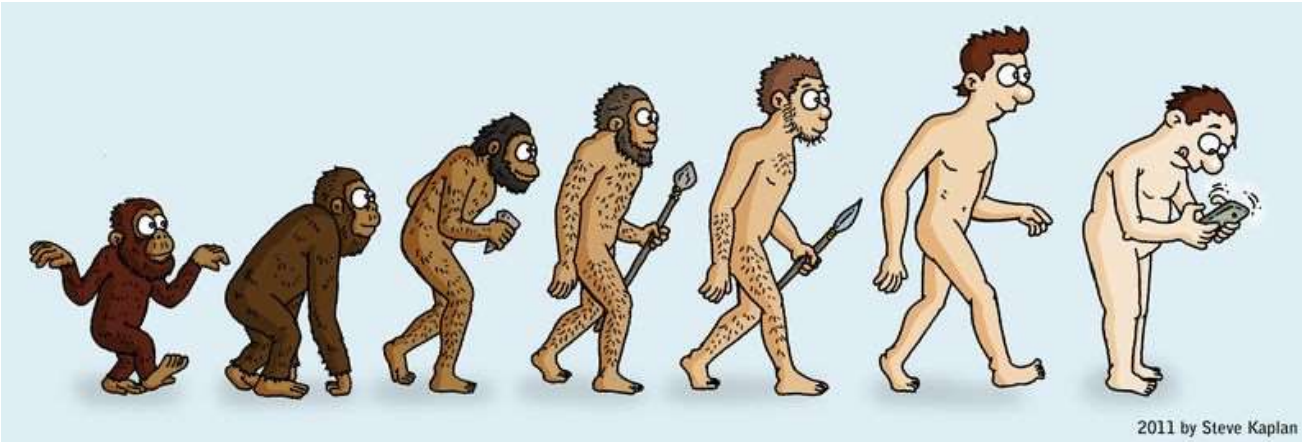
Learning Science and English by TED

- **Subtitles (字幕)**
- **Interactive transcript (互動文稿)**

Chapter 14
Mendel and the Gene Idea

Pea plants

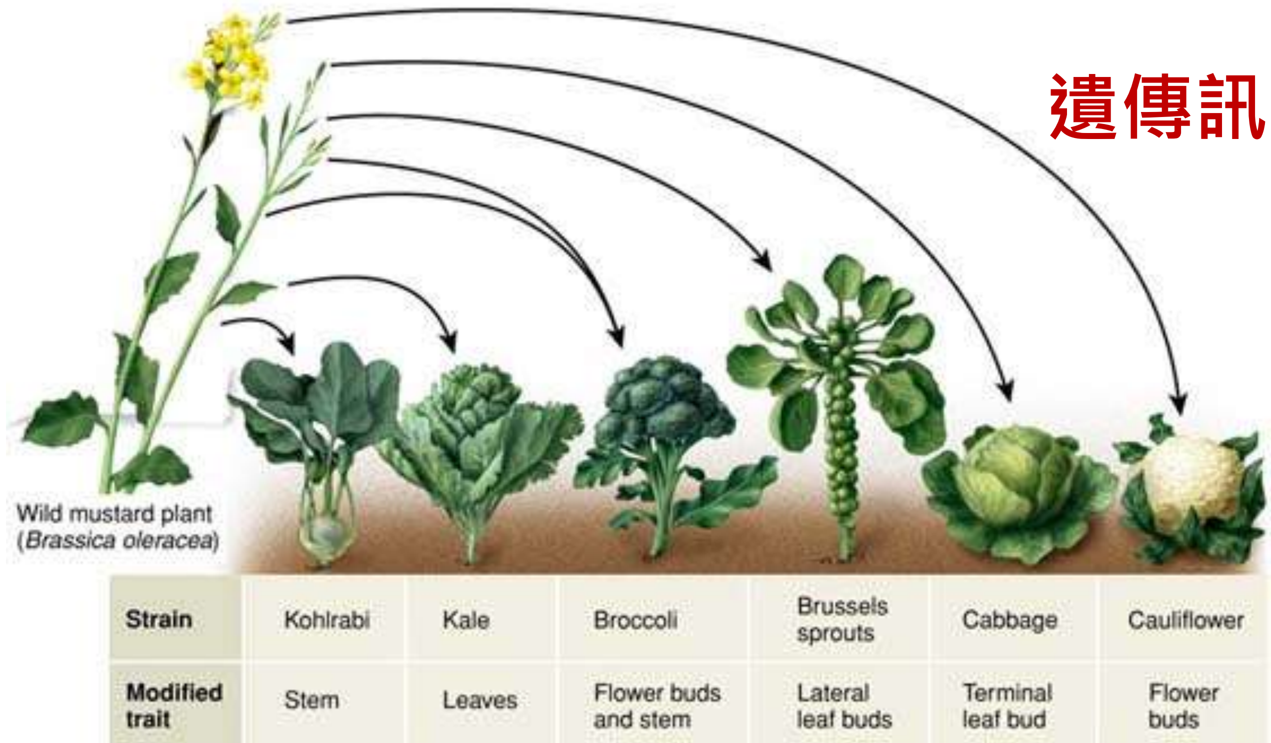
Modified by YJ Chuang at NTHU-DMS



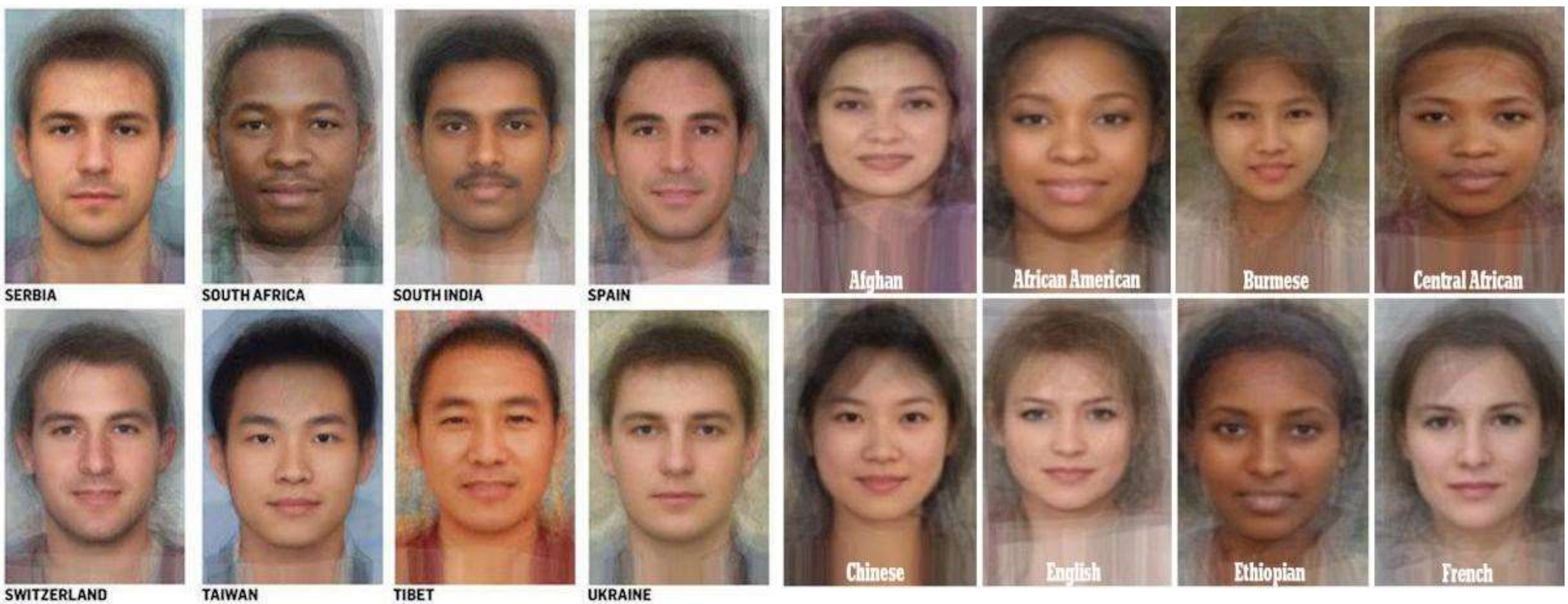
Evolution

遺傳訊息的漸變與重組

Breeding



“問題”源自於”觀察”

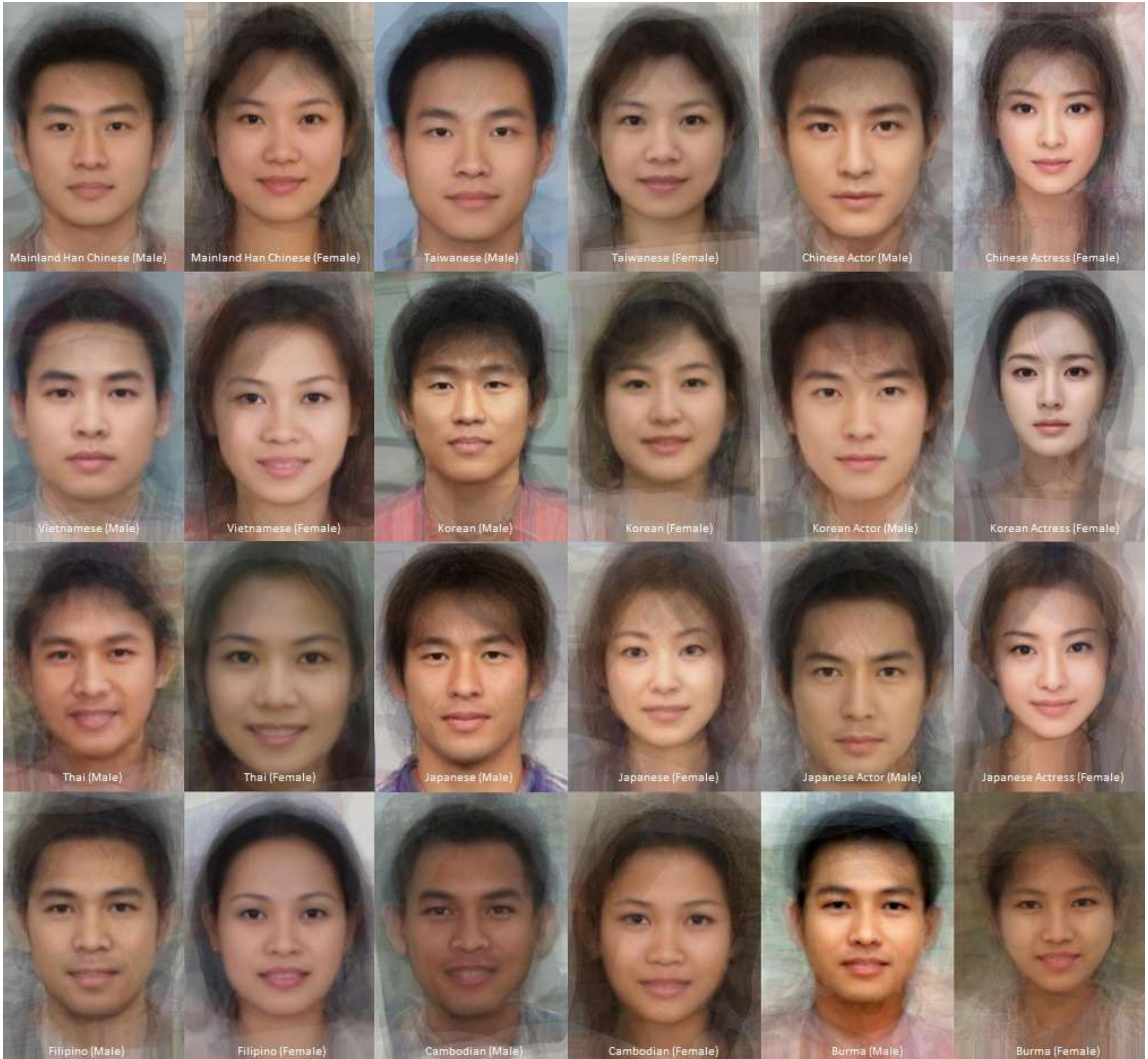


Similarity vs. Diversity

Method: Composite portraiture; Experimental psychologists at University of Glasgow

1 2 3 4 5 6

A B C D

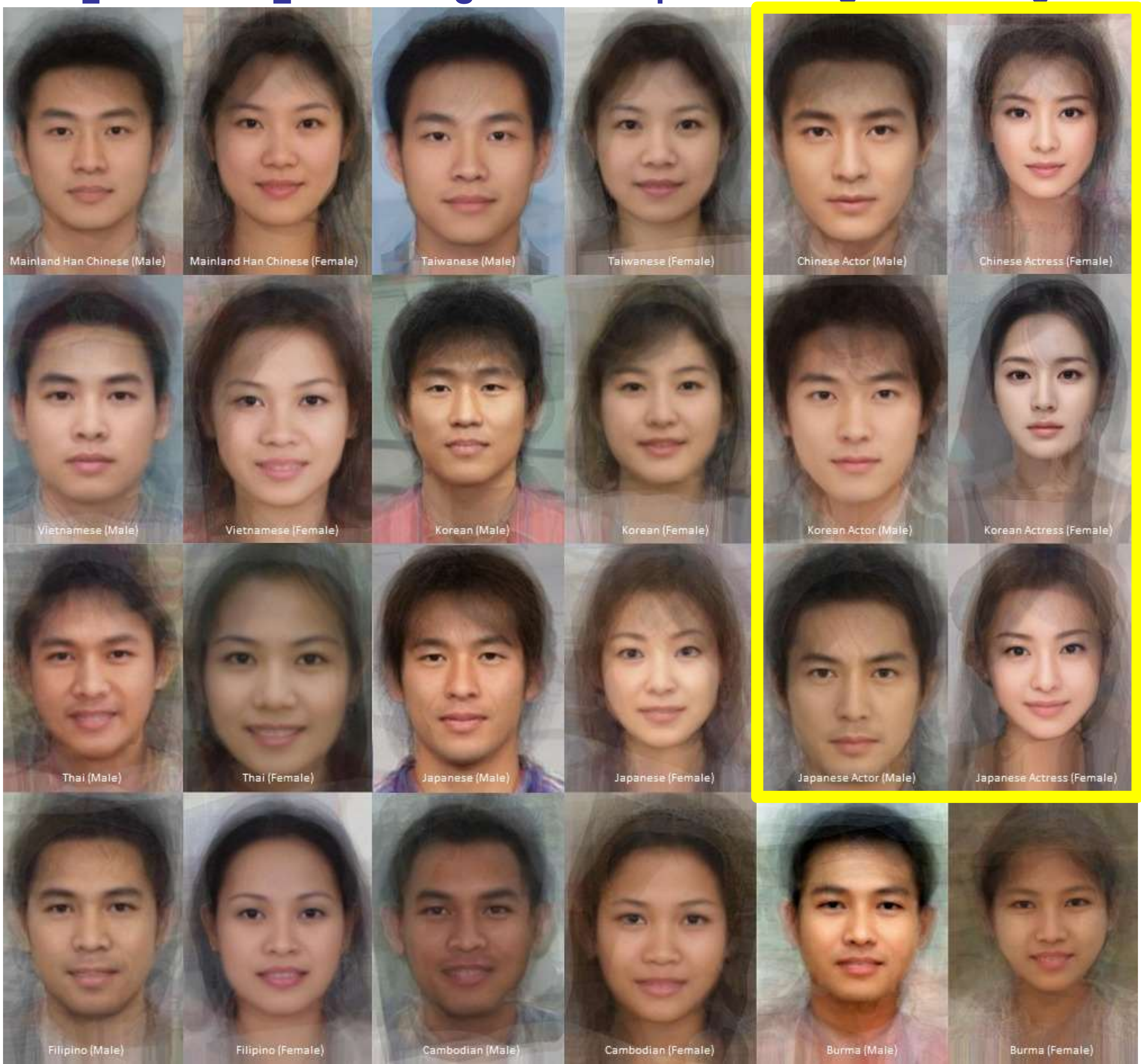


限縮至亞洲人

問題:有魅力的臉部特徵為何?

1 2 3 4 5 6

A B C D



再限縮至 東亞裔 演員

Key Question in Genetics 選定關鍵領域課題提問

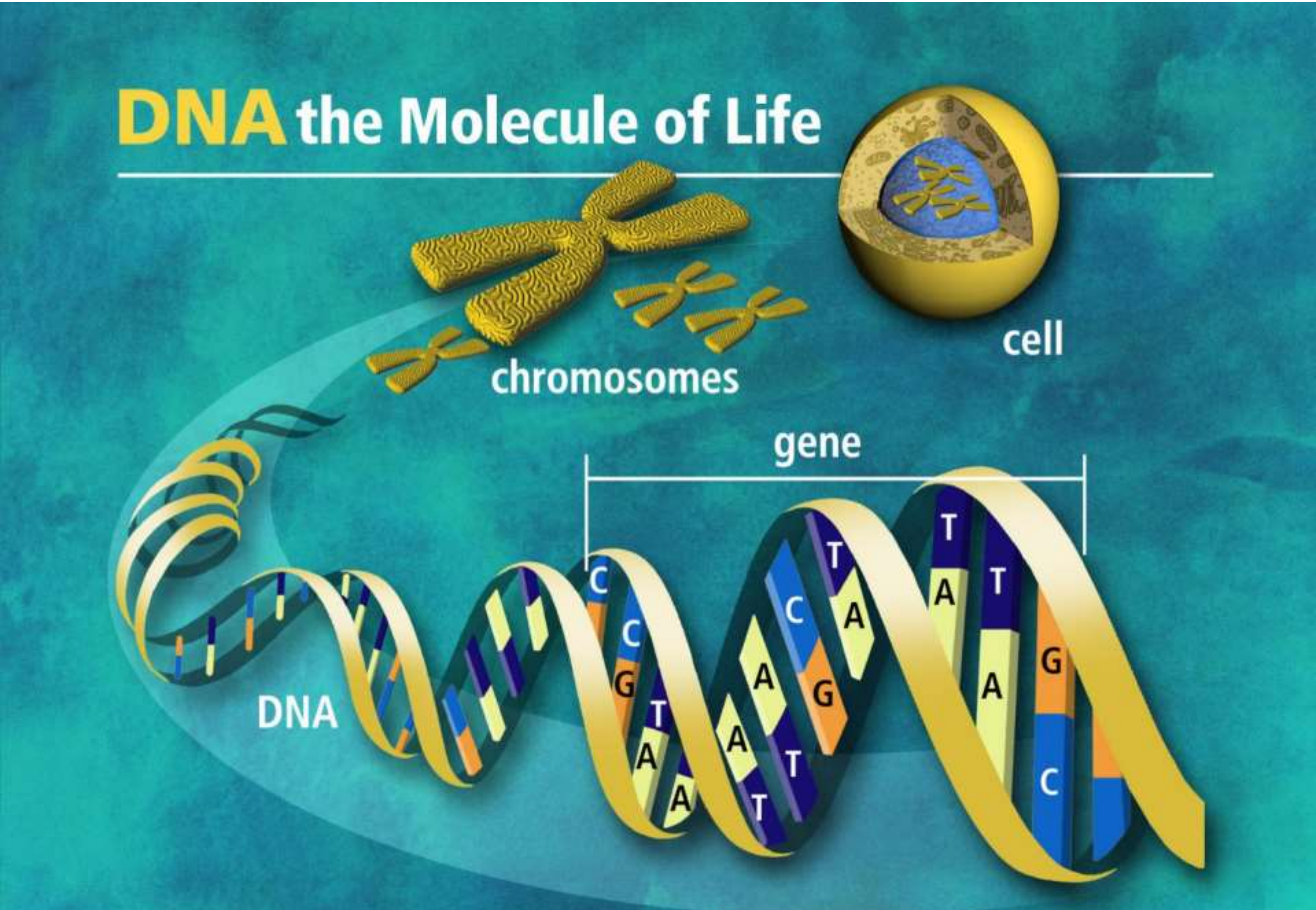
Inheritance - How to pass the genes to next generation?

Long history of live stock and agriculture breeding to select for more food and textile production

http://www.caas.cn/en/newsroom/research_update/251062.shtml

https://pixabay.com/p-57705/?no_redirect

200年來，研究者如何探索、理解基因? (知識的形成與累積)



Inheritance (繼承;有其父必有其子?)

- Plant and animal breeders question about the inheritance of **flower colors, fur length, and other characters** of organisms.

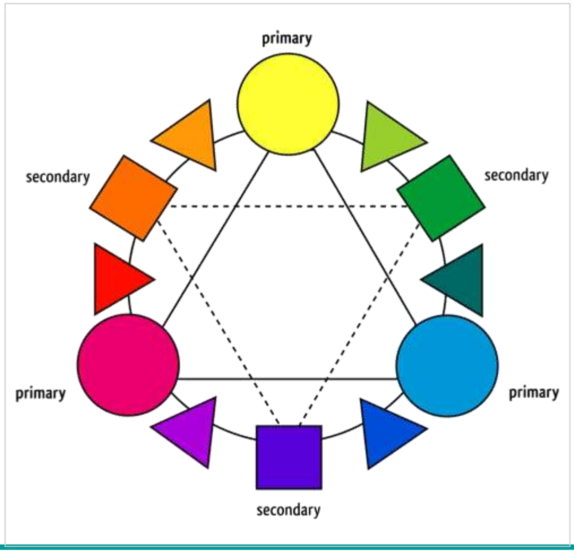
- How do these traits pass from generation to generation? Observations alone could not answer all questions.

+

Overview: Drawing from the Deck of Genes (抽牌?)

Question: What genetic principles account for **the passing of traits** from parents to offspring?

- During the 1800s, the "blending" hypothesis is the idea that genetic material from the two parents blends together (like blue and yellow paint blend to make green)



The Blending Hypothesis *(is not supported by the evidences)*

But people observed many exceptions to blending hypothesis. For example, red-flowered parents sometimes produced yellow-flowered offspring. The blending hypothesis could not explain how traits that disappear in one generation can reappear in later ones.

實際驗證…

YIELDS

+

The Gene idea!

- The "particulate" hypothesis (散粒; 不是 particular !) is the idea that parents pass on discrete (離散的;不連接的) heritable units (genes)
- Mendel documented a particulate mechanism through his experiments with garden peas.

Fig. 14-1

Fuchsia 倒掛金鐘

Gregor Johann Mendel

[**greg**-er **yoh**-hann **men**-dl]

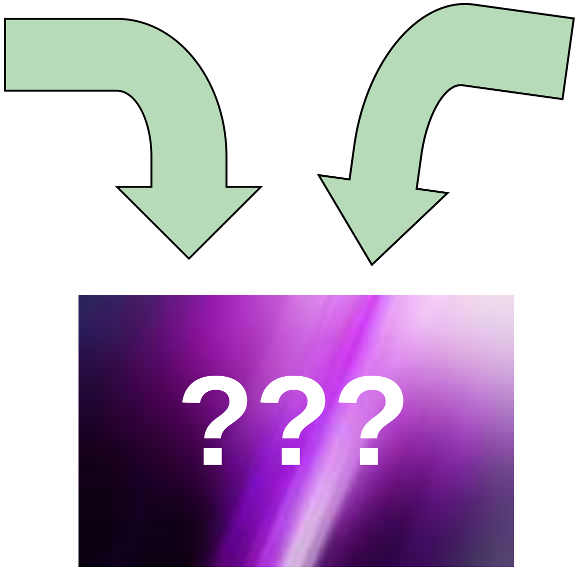
Mendelian [men-**dee**-lee-*uh*n]

Mendel was a curious monk. (好奇心是學習的起點)

He wondered, "if a **purple-flowered** pea plant were fertilized with pollen from a **white-flowered** plant, what color flowers would the offspring have?"

觀察後,

發想可驗證問題



Concept 14.1: Mendel used the scientific approach to identify two laws of inheritance

- Mendel discovered the basic principles of heredity by breeding garden peas in carefully planned experiments

假設答案，並設計實驗查證

If you were Mendel,

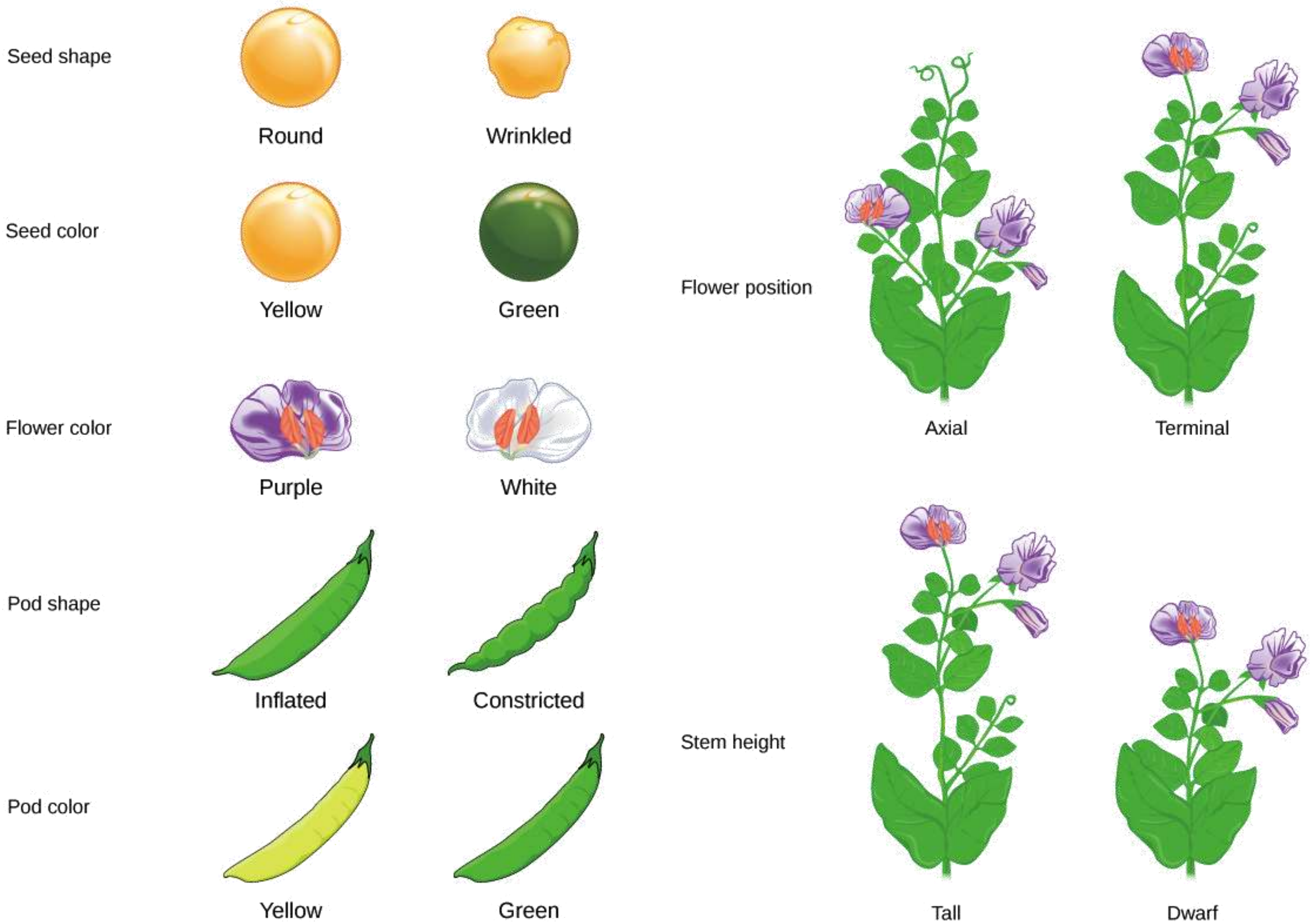
How will you design the experiment?

(你會如何設計實驗，藉以解釋遺傳的控制?)

Mendel's Experimental Design → Quantitative Approach
實驗設計 定量

- Advantages of pea plants for genetic study:
  - Pea plant has many varieties with distinct heritable features, or **characters** (such as flower color); character variants (such as purple or white flowers) are called **traits (表徵/特徵)**see next page
  - Mating of pea plants can be controlled (實驗條件)
    - Each pea plant has sperm-producing organs (stamens) and egg-producing organs (carpels)
    - Cross-pollination (fertilization between different plants) can be achieved by dusting one plant with pollen from another

Traits 表徵/特徵


Seed shape
Round
Wrinkled
Seed color
Yellow
Green
Flower color
Purple
White
Pod shape
Inflated
Constricted
Pod color
Yellow
Green
Flower position
Axial
Terminal
Stem height
Tall
Dwarf

Mendel's Experimental Design → Quantitative Approach
實驗設計 定量

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    - Cross-pollination (fertilization between different plants) can be achieved by dusting one plant with pollen from another

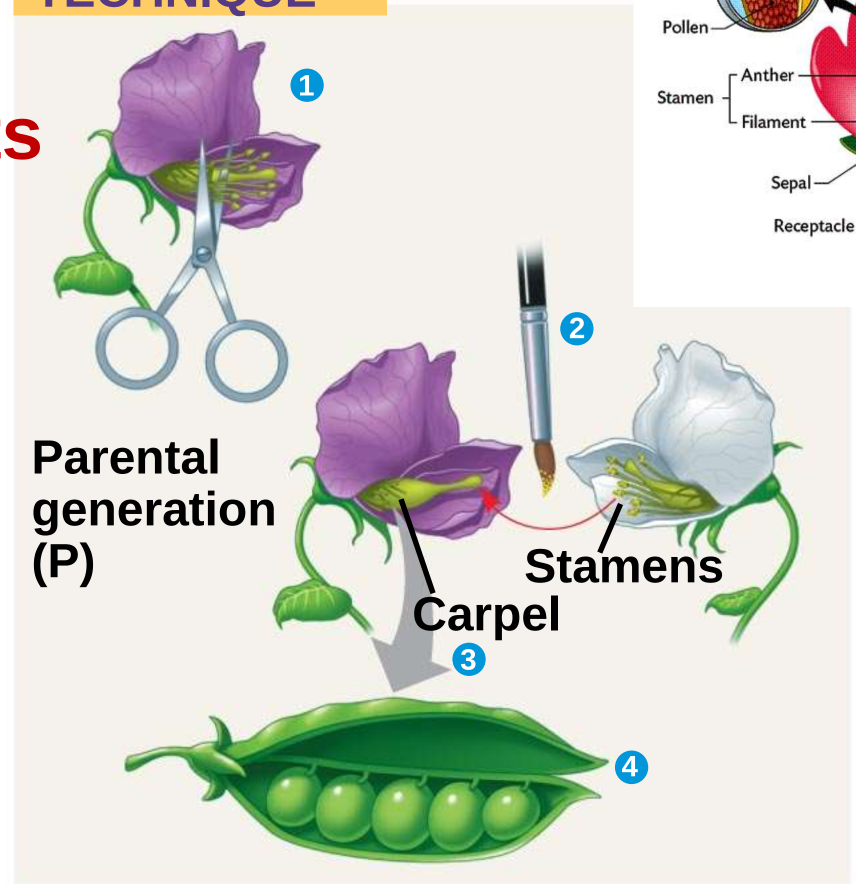
Fig. 14-2

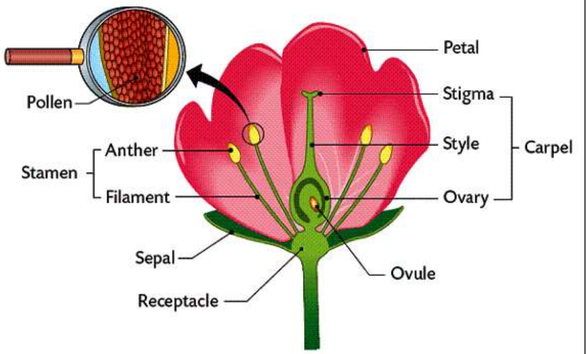
Research Method: Crossing Pea Plants

P: Parental (親代)

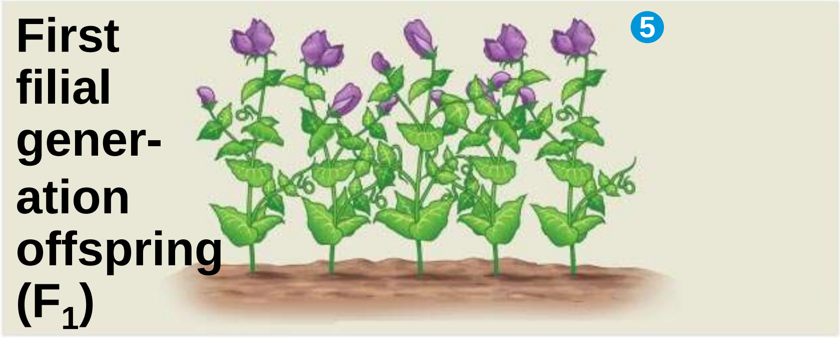
F: Filial (子代)

TECHNIQUE





RESULTS



Selecting the experimental variable/parameter

(可定性、定量分析的 實驗變數 與 遺傳因子)

- Mendel chose to track only those characters that varied in an **either-or** manner (either-or; in this case - 非白即紫)
- He also used varieties that were **true-breeding** (純種品系: plants that produce offspring of the same variety when they self-pollinate)

Hybridization (雜交) < experimental approach

- In a typical experiment, Mendel mated two contrasting, true-breeding varieties, a process called **hybridization**

- In Mendel's experiment:
  - The true-breeding parents are the **P generation** (parental generation)
  - The hybrid offspring of the P generation are called the **F_1 generation** (1^{st} filial generation)
  - When F_1 individuals self-pollinate, the **F_2 generation** is produced

See illustration next page

Fig. 14-3-1

EXPERIMENT

P Generation (true-breeding parents)

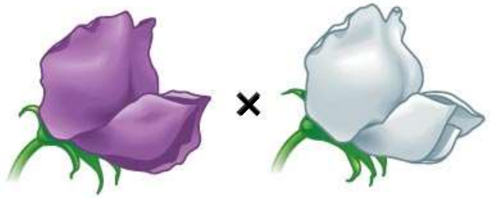
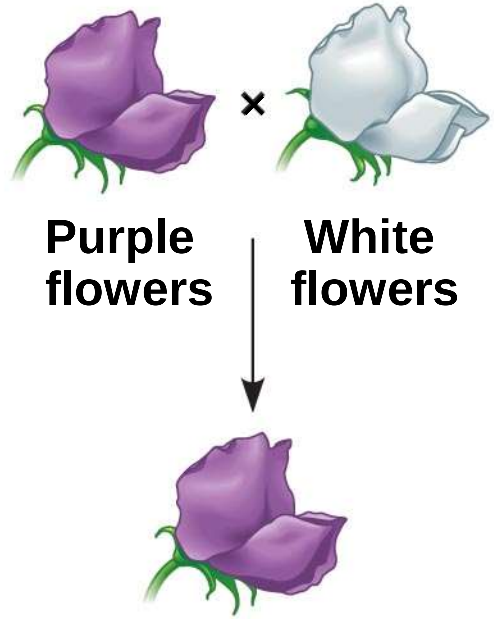


Fig. 14-3-2

EXPERIMENT

P Generation (true-breeding parents)



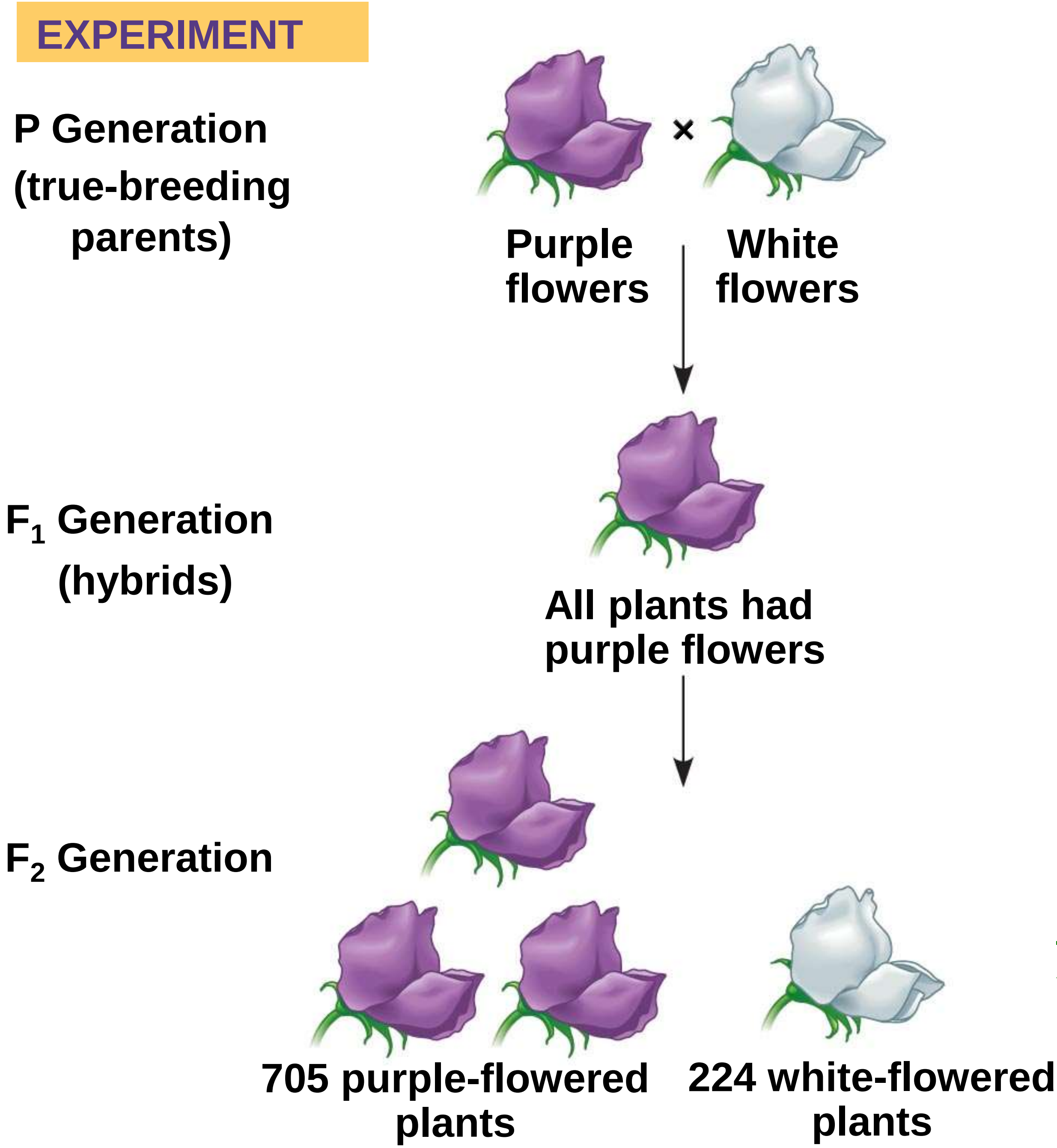
Purple flowers

White flowers

F_1 Generation (hybrids)

All plants had purple flowers

Fig. 14-3-3



實驗結果

Data Analysis and Interpretation 數據分析與解讀

- Mendel reasoned that *only the purple flower factor* was affecting flower color in the F_1 hybrids
- Mendel called the purple flower color a dominant trait and the white flower color a recessive trait
- Mendel observed the same pattern of inheritance in six other pea plant characters, each represented by two traits (擴大驗證)
- What Mendel called a "heritable factor" is what we now call a gene

The Law of Segregation 導出規則、定律

- When Mendel crossed contrasting, true-breeding white and purple flowered pea plants, he found all of the F_1 hybrids were purple
- When Mendel crossed the F_1 hybrids, many of the F_2 plants had purple flowers, but some had white
- Mendel discovered a ratio of about 3 to 1, purple to white flowers, in the F_2 generation

Table 14-1

Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	x	Recessive Trait	F_2 Generation Dominant:Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Seed color	Yellow 	×	Green 	6,022:2,001	3.01:1
Seed shape	Round 	×	Wrinkled 	5,474:1,850	2.96:1
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1
Pod color	Green 	×	Yellow 	428:152	2.82:1
Stem length	Tall 	×	Dwarf 	787:277	2.84:1

3:1 ratio

定性、定量分析

擴大驗證 (Repeat)

導出規則

Why 3 to 1? (實驗發現引出新問題)

Mendel's Model – hypothesis to explain the data

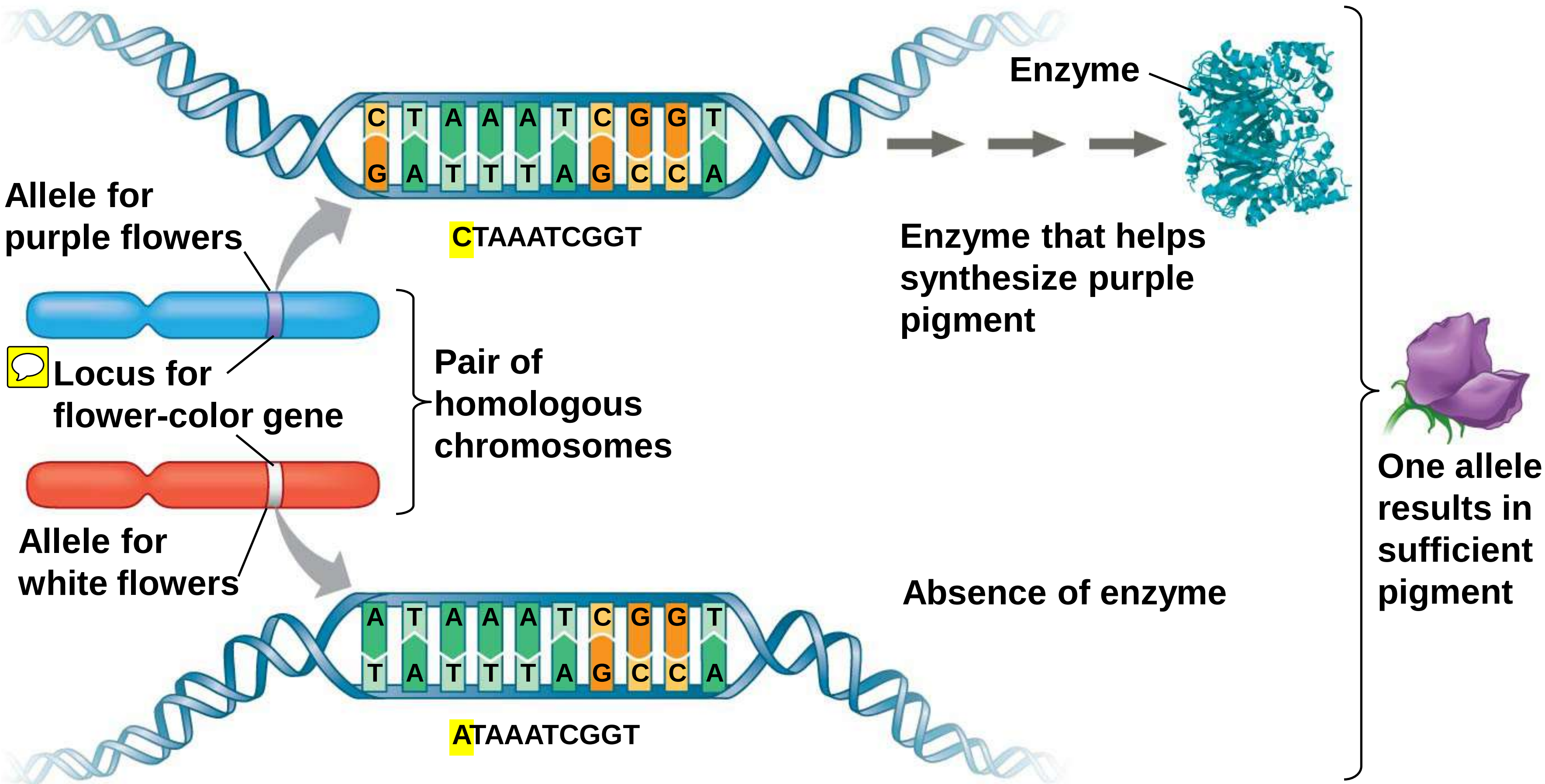
- Mendel developed a hypothesis to explain the 3:1 inheritance pattern he observed in F_2 offspring
- **Four related concepts** make up this model to support the hypothesis
- These concepts can be related to what we now know about **genes** and **chromosomes**

建立新的、更詳盡的 [假設與理論模型] 去解釋實驗發現

(Concept 1/4) Alleles: alternative versions of genes

- The first concept is that alternative versions of genes account for variations in inherited characters
  - For example, the gene for flower color in pea plants exists in two versions, one for purple flowers and the other for white flowers
- These alternative versions of a gene are now called **alleles**
- Each gene resides at a specific locus on a specific chromosome

Figure 14.4



Gene → Protein → Phenotype

(Concept 2/4) Each parent offers one allele

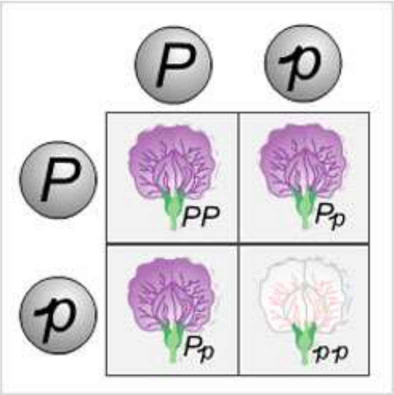
- The second concept is that for each character an organism inherits two alleles, one from each parent
- Mendel made this deduction *without knowing about the role of chromosomes*
- The two alleles at a locus on a chromosome may be identical, as in the true-breeding plants of Mendel's P generation
- Alternatively, the two alleles at a locus may differ, as in the F_1 hybrids

Useful Genetic Vocabulary

- An organism with two identical alleles for a character is said to be **homozygous**同型合子 (PP or pp) for the gene controlling that character
- An organism that has two different alleles for a gene is said to be **heterozygous** (Pp) for the gene controlling that character
- Unlike homozygotes, heterozygotes are not true-breeding

(Concept 3/4) Dominant 顯性 vs. Recessive 隱性 allele

- The third concept is that if the two alleles at a locus differ, then one (the **dominant allele**) determines the organism's appearance, and the other (the **recessive allele**) has no noticeable effect on appearance
- In the flower-color example, the F_1 plants had purple flowers because the allele for that trait is dominant



(Concept 4/4) One heritable character in one gamete

- The fourth concept, now identified as the **law of segregation**, states that the two alleles for a heritable character separate (segregate) during gamete formation and end up in different gametes (配子=精子或卵子)
- Thus, an egg or a sperm gets only one of the two alleles that are present in the somatic cells (of the body) of an organism
- This segregation of alleles corresponds to the distribution of homologous chromosomes to different gametes (sperm or egg) in meiosis

Figure 14.3-1

Experiment

P Generation (true-breeding parents)

Purple flowers

White flowers

Figure 14.3-2

Experiment

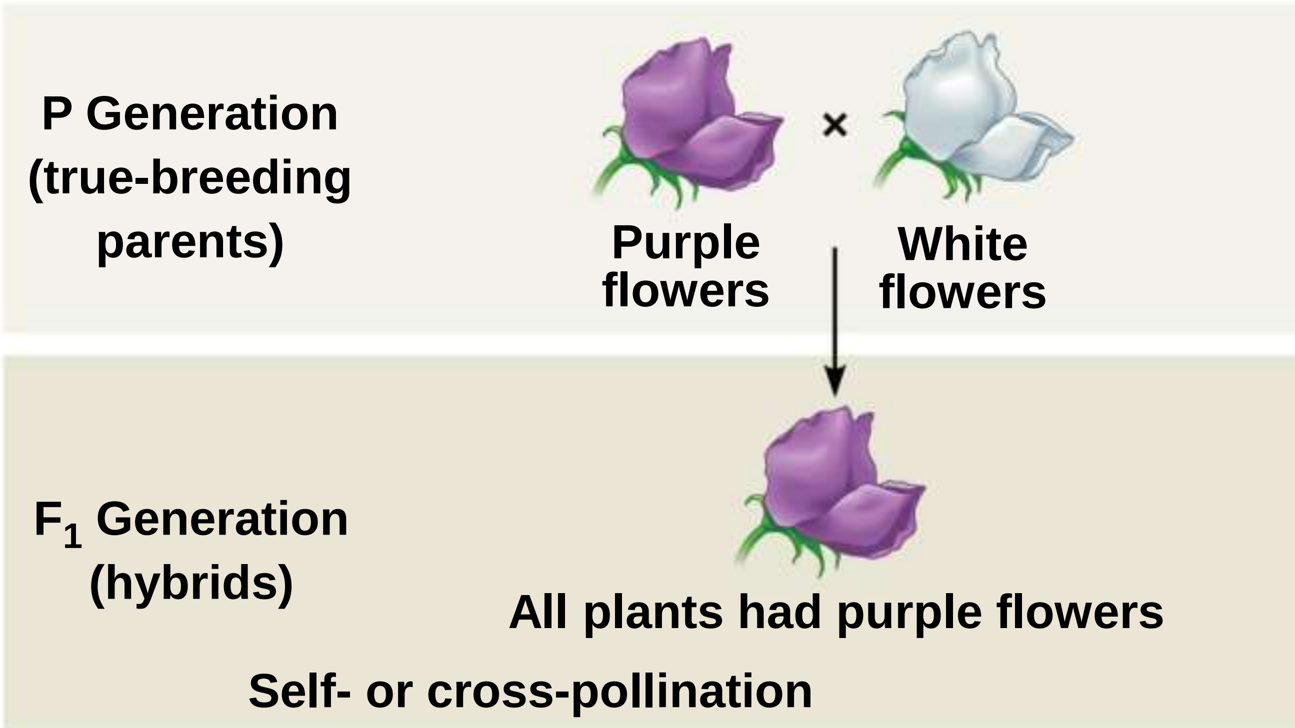
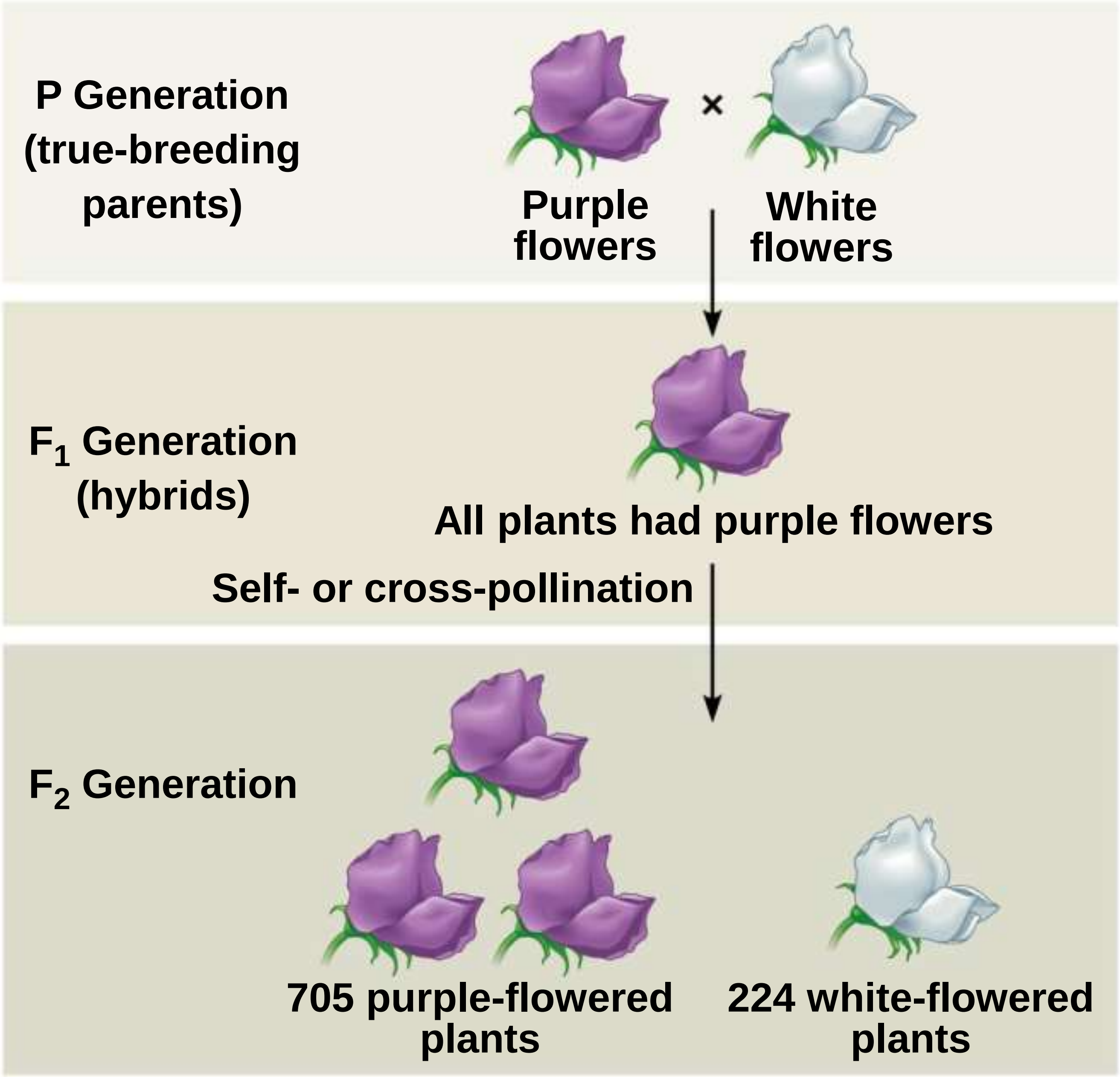


Figure 14.3-3

Experiment



- Mendel observed the same pattern of inheritance in six other pea plant characters, each represented by two traits
- What Mendel called a “heritable factor” is what we now call a gene

實驗結論必需重複驗證 - Reproducibility

Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Flower color	Purple 	×	White 	705:224	3.15:1
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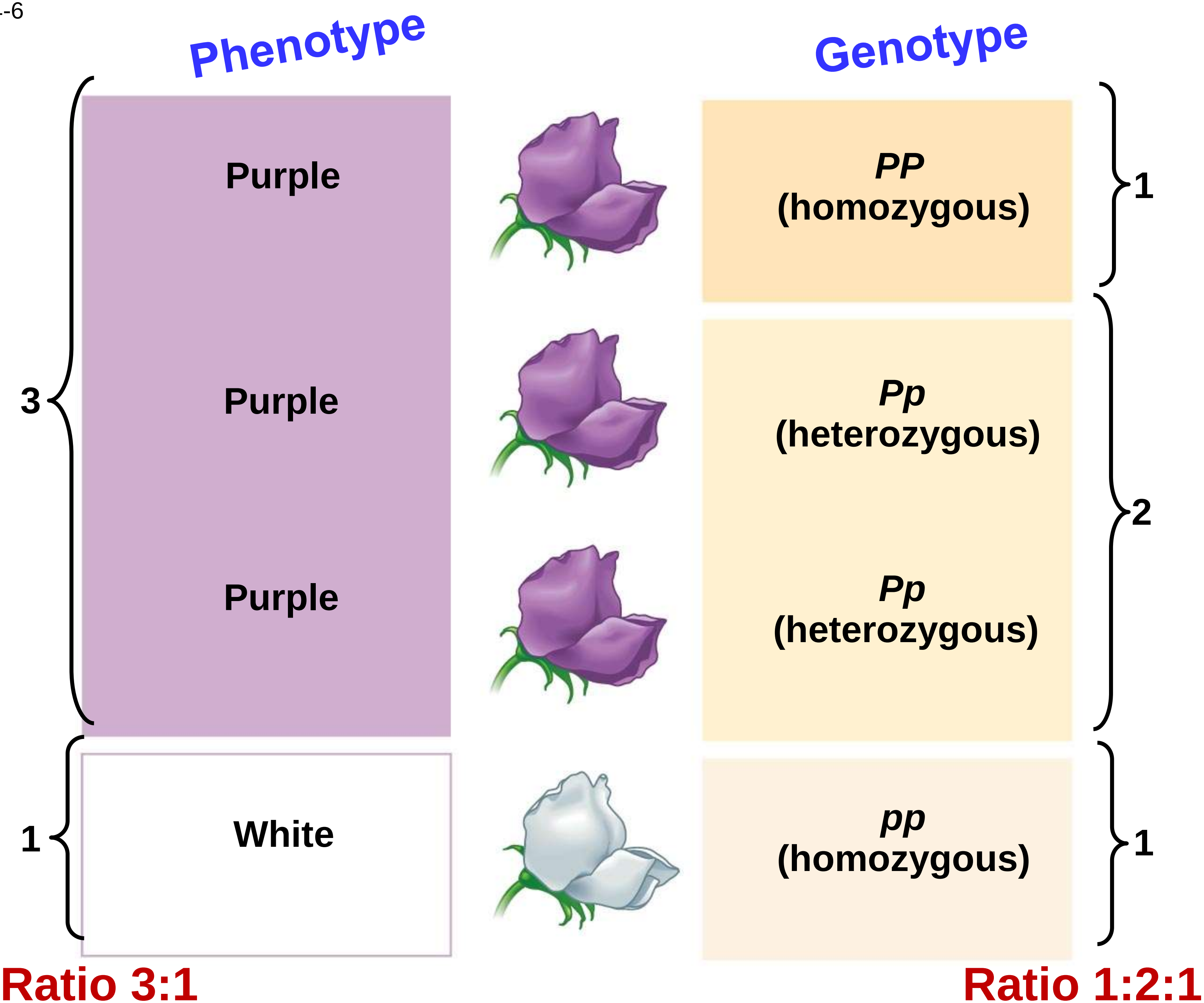
Table 14.1 The Results of Mendel's F_1 Crosses for Seven Characters in Pea Plants

Character	Dominant Trait	×	Recessive Trait	F_2 Generation Dominant: Recessive	Ratio
Pod shape	Inflated 	×	Constricted 	882:299	2.95:1
Pod color	Green 	×	Yellow 	428:152	2.82:1
Flower position	Axial 	×	Terminal 	651:207	3.14:1
Stem length	Tall 	×	Dwarf 	787:277	2.84:1

Phenotype vs. Genotype

- Because of the different effects of dominant and recessive alleles, an organism’s traits do not always reveal its genetic composition
- Therefore, we distinguish between an organism’s **phenotype** 顯型;表現型, or physical appearance, and its **genotype** 基因型, or genetic makeup
- In the example of flower color in pea plants, *PP* and *Pp* plants have the same phenotype (purple) but different genotypes

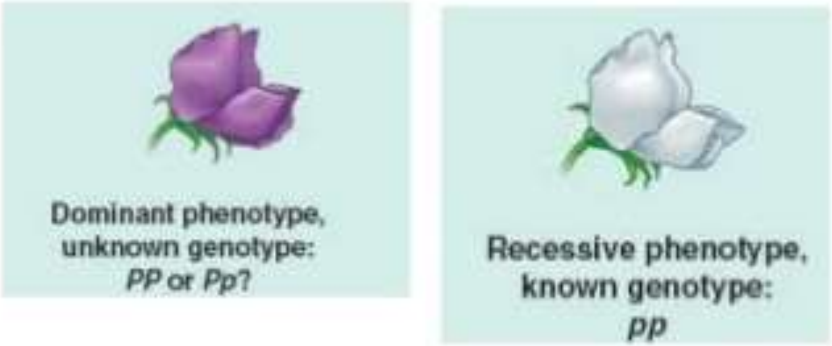
Fig. 14-6


Phenotype
Genotype
Purple
PP
(homozygous)
1
3
Purple
Pp
(heterozygous)
2
Purple
Pp
(heterozygous)
1
White
pp
(homozygous)
1
Ratio 3:1
Ratio 1:2:1

The Testcross (試交)

- **Why is it done?**

  To find if an organism is homozygous dominant or heterozygous.



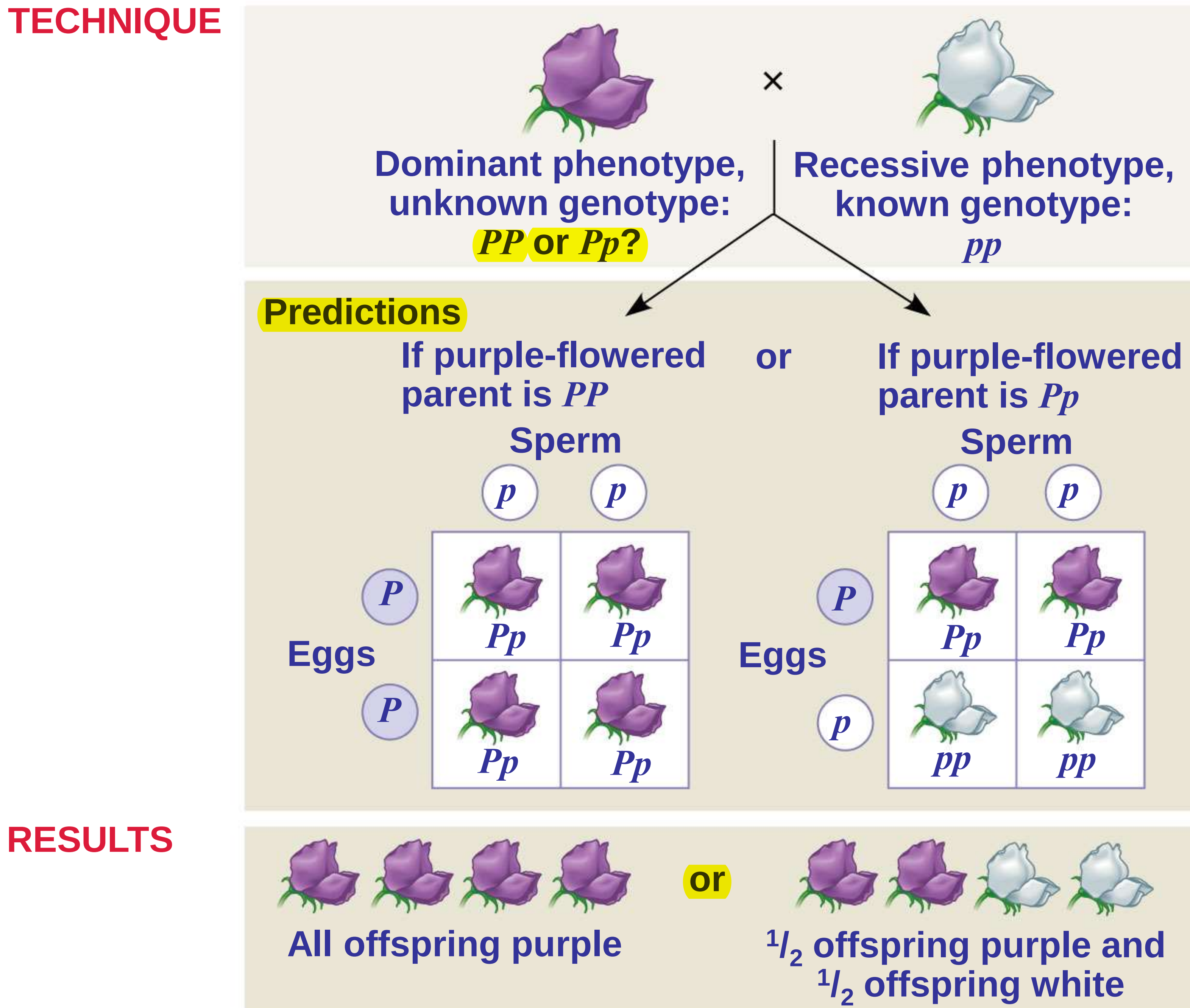
- **How is it done?**

  Organism of unknown genotype is mated with a homozygous recessive one.

- **How is a conclusion drawn?**

Look at offspring phenotype ratios

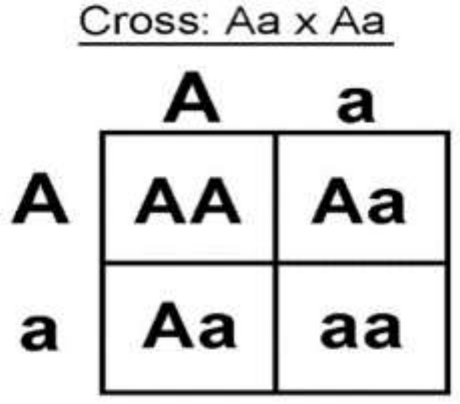
Figure 14.7 Research Methods: the Testcross



Punnett square

圖式表達 Phenotype-Genotype 關聯性

- Mendel's segregation model accounts for the 3:1 ratio he observed in the F_2 generation of his numerous crosses
- The possible combinations of sperm and egg can be shown using a **Punnett square**, a diagram for predicting the results of a genetic cross between individuals of known genetic makeup



A: dominant

a: recessive

Deriving the "Laws" of inheritance

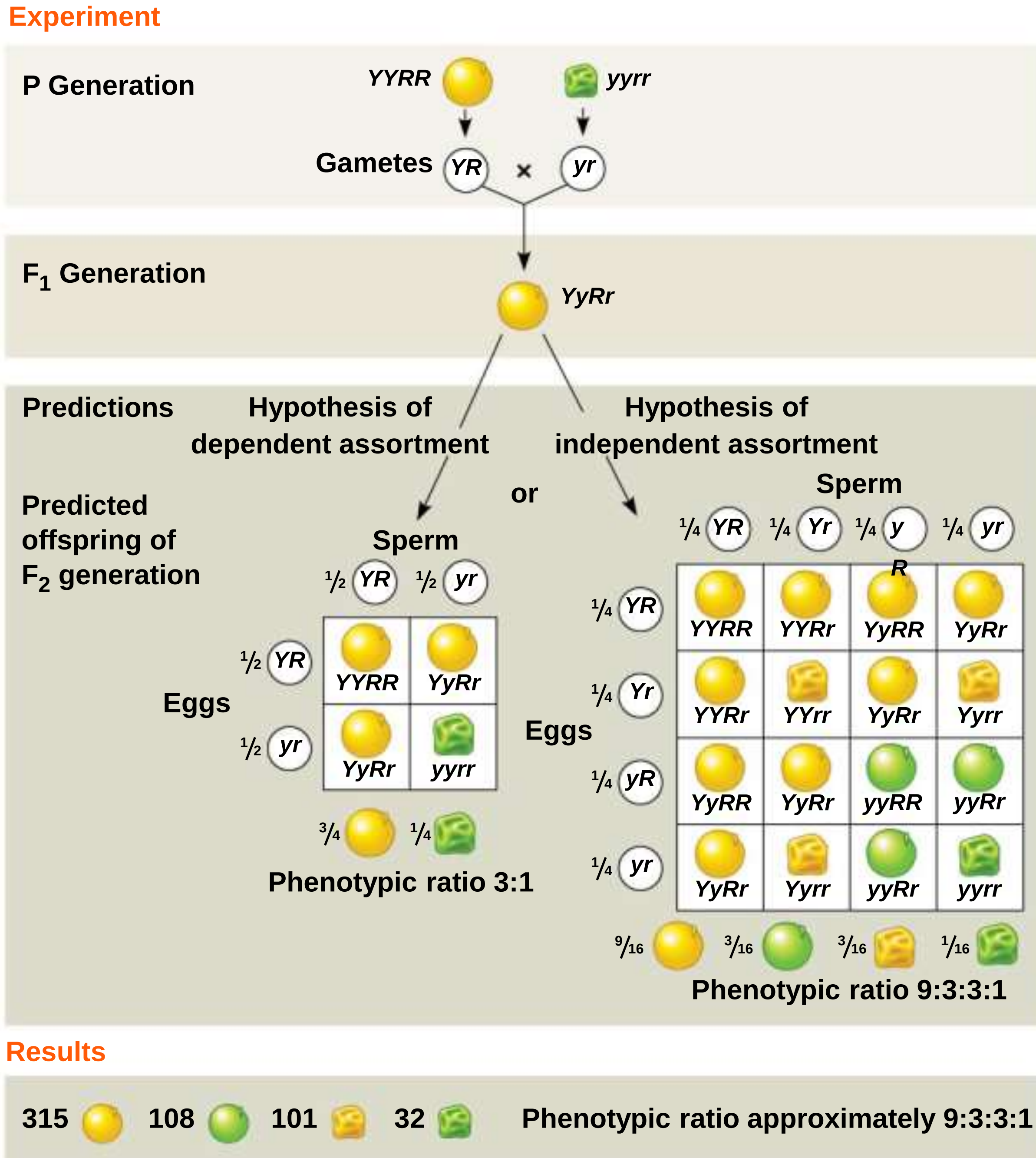
- Mendel derived the **law of segregation** by following a single character
- The F_1 offspring produced in this cross were **monohybrids (單性雜種),** individuals that are heterozygous for one character
- A cross between such heterozygotes is called a *monohybrid cross*

mono-hybrid		
Aa x Aa		
	A	a
A	AA	Aa
a	aA	aa

Changing experimental conditions and finds second law

- Mendel identified his second **law of inheritance** by following two characters at the same time
- Crossing two true-breeding parents differing in two characters produces **dihybrids** in the F_1 generation, heterozygous for both characters
- A dihybrid cross, a cross between F_1 dihybrids, can determine whether two characters are transmitted to offspring as a package or independently (see next silde)

Figure 14.8


Experiment
P Generation
YYRR
yyrr
Gametes
YR
×
yr
F1 Generation
YyRr
Predictions
Hypothesis of dependent assortment
or
Hypothesis of independent assortment
Predicted offspring of F2 generation
Sperm
½ YR
½ yr
Eggs
½ YR
½ yr
YYRR
YyRr
YyRr
yyrr
¾
¼
Phenotypic ratio 3:1
Sperm
¼ YR
¼ Yr
¼ yR
¼ yr
Eggs
¼ YR
¼ Yr
¼ yR
¼ yr
YYRR
YYRr
YyRR
YyRr
YYRr
YYrr
YyRr
Yyrr
YyRR
YyRr
yyRR
yyRr
YyRr
Yyrr
yyRr
yyrr
9/16
3/16
3/16
1/16
Phenotypic ratio 9:3:3:1
Results
315
108
101
32
Phenotypic ratio approximately 9:3:3:1

Law of independent assortment

- Using a dihybrid cross, Mendel developed the **law of independent assortment**:

  Each pair of alleles segregates independently of each other pair of alleles during gamete 配子 formation

- Strictly speaking, this law applies only to genes on different, nonhomologous chromosomes
  - Genes located near each other on the same chromosome tend to be inherited together

孟德爾 的遺傳研究，讓人類超越想像，把知識的極限推前了一大步

1. 選定關鍵領域
2. 觀察現象
3. 發想可驗證問題
4. 提出假設(可能答案)，並設計實驗查證
5. 實驗設計 - 可[定性、定量]分析的變數
6. 數據分析與解讀
7. 導出規則、定律
8. 實驗結論可被重複驗證
9. 清楚而正確的闡述

孟德尔(Johann Gregor Mendel, 1822-1884)

Concept 14.2: Probability laws govern Mendelian inheritance (衍申的相關學理)

- Mendel's laws of segregation and independent assortment reflect the rules of probability
  - When tossing a coin, the outcome of one toss has no impact on the outcome of the next toss
  - In the same way, the alleles of one gene segregate into gametes independently of another gene's alleles

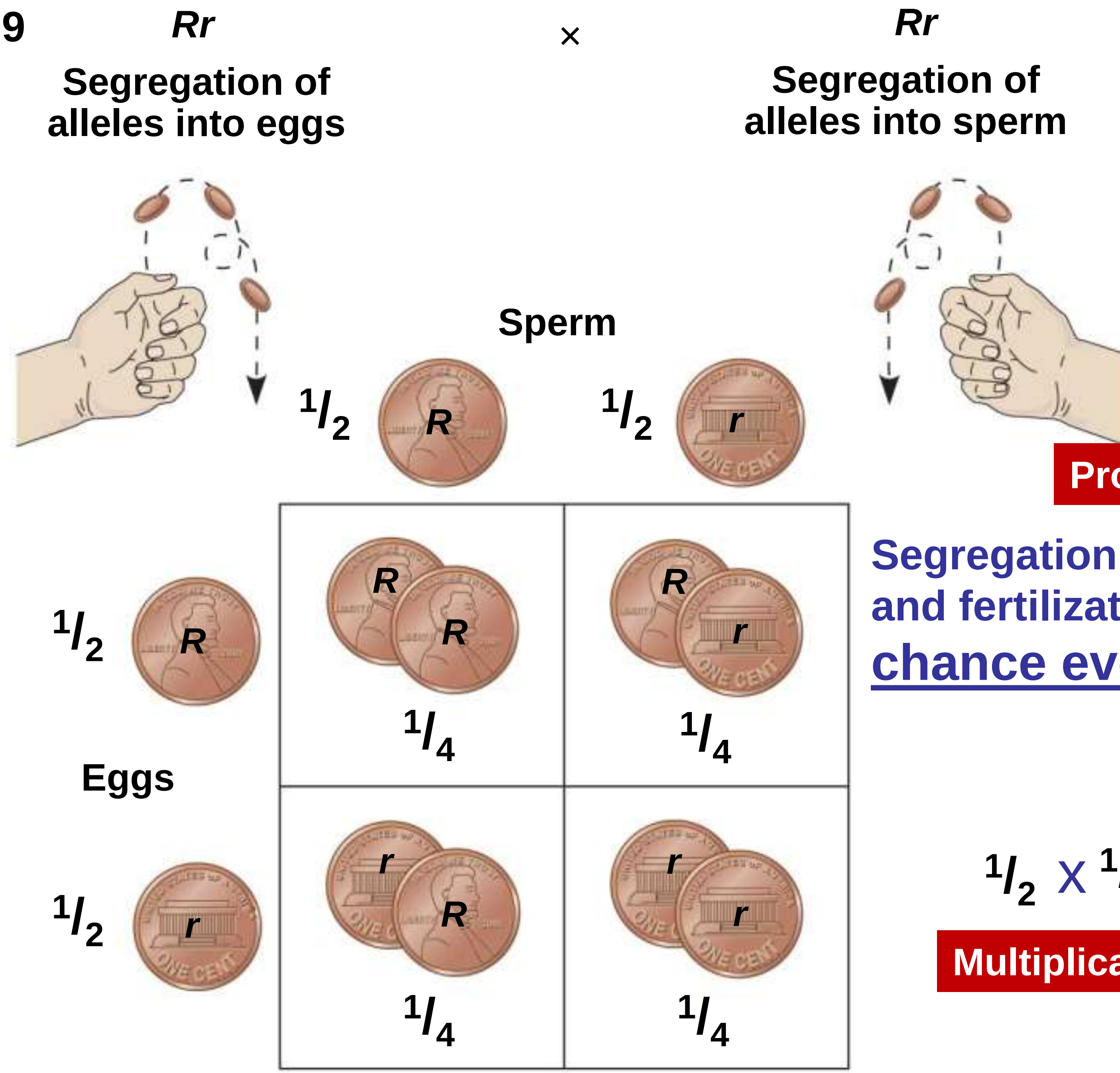
See Fig.14.9

The Multiplication and Addition Rules Applied to Monohybrid Crosses

- The multiplication rule states that the probability that two or more independent events will occur together is the product of their individual probabilities
- Probability in an F_1 monohybrid cross can be determined using the multiplication rule
- Segregation in a heterozygous plant is like flipping a coin: Each gamete has a ½ chance of carrying the dominant allele and a ½ chance of carrying the recessive allele

See Fig.14.9

Fig. 14-9



Probability

Segregation of alleles and fertilization as **chance events**

$^{1}/_{2} \times {}^{1}/_{2} = {}^{1}/_{4}$

Multiplication

Rule of addition

- The rule of addition states that the probability that any one of two or more exclusive events will occur is calculated by adding together their individual probabilities
- The rule of addition can be used to figure out the probability that an F_2 plant from a monohybrid cross will be heterozygous rather than homozygous

Solving Complex Genetics Problems with the Rules of Probability

- We can apply the multiplication and addition rules to predict the outcome of crosses involving multiple characters
- A dihybrid or other multicharacter cross is equivalent to two or more independent monohybrid crosses occurring simultaneously
- In calculating the chances for various genotypes, each character is considered separately, and then the individual probabilities are multiplied together

See Calculation next page

Calculating the chances for various genotypes

Question:

What fraction of offspring from PpYyRr x Ppyyrr are predicted to exhibit the recessive phenotypes for <u>at least two of the three</u> characters?

List the possible combinations

independent

ppyyRr	$\frac{1}{4}$ (probability of *pp*) $\times$ $\frac{1}{2}$ (*yy*) $\times$ $\frac{1}{2}$ (*Rr*)	$= \frac{1}{16}$
ppYyrr	$\frac{1}{4} \times \frac{1}{2} \times \frac{1}{2}$	$= \frac{1}{16}$
Ppyyrr	$\frac{1}{2} \times \frac{1}{2} \times \frac{1}{2}$	$= \frac{2}{16}$
PPyyrr	$\frac{1}{4} \times \frac{1}{2} \times \frac{1}{2}$	$= \frac{1}{16}$
ppyyrr	$\frac{1}{4} \times \frac{1}{2} \times \frac{1}{2}$	$= \frac{1}{16}$
Chance of *at least two* recessive traits		$= \frac{6}{16}$ **or** $\frac{3}{8}$

Addition

Concept 14.3: Inheritance patterns are often more complex than predicted by simple Mendelian genetics

- The relationship between genotype and phenotype is *rarely as simple* as in the pea plant characters Mendel studied
- Many heritable characters are not determined by only one gene with two alleles (真實世界的複雜~)
- However, the basic principles of segregation and independent assortment apply even to more complex patterns of inheritance

Extending Mendelian Genetics for a Single Gene

- Inheritance of characters by a single gene may deviate 偏離 from simple Mendelian patterns in the following situations:
  - When alleles are not completely dominant or recessive (example: some flower color)
  - When a gene has more than two alleles (example: ABO blood type)
  - When a gene produces multiple phenotypes (example: Phenylketonuria - mental retardation, eczema, pigment defects)

Degrees of Dominance

- **Complete dominance** occurs when phenotypes of the heterozygote and dominant homozygote are identical
- In **incomplete dominance**, the phenotype of F_1 hybrids is somewhere between the phenotypes of the two parental varieties
- In **co-dominance**, two dominant alleles affect the phenotype in separate, distinguishable ways

Fig. 14-10

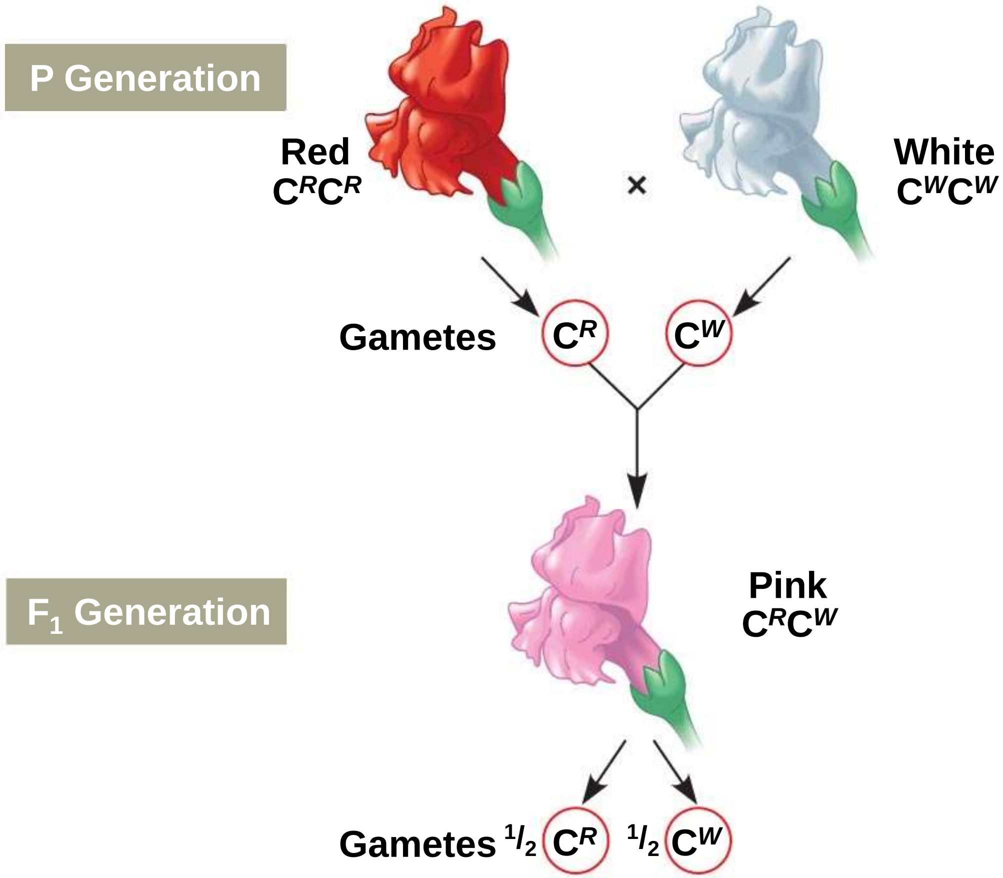
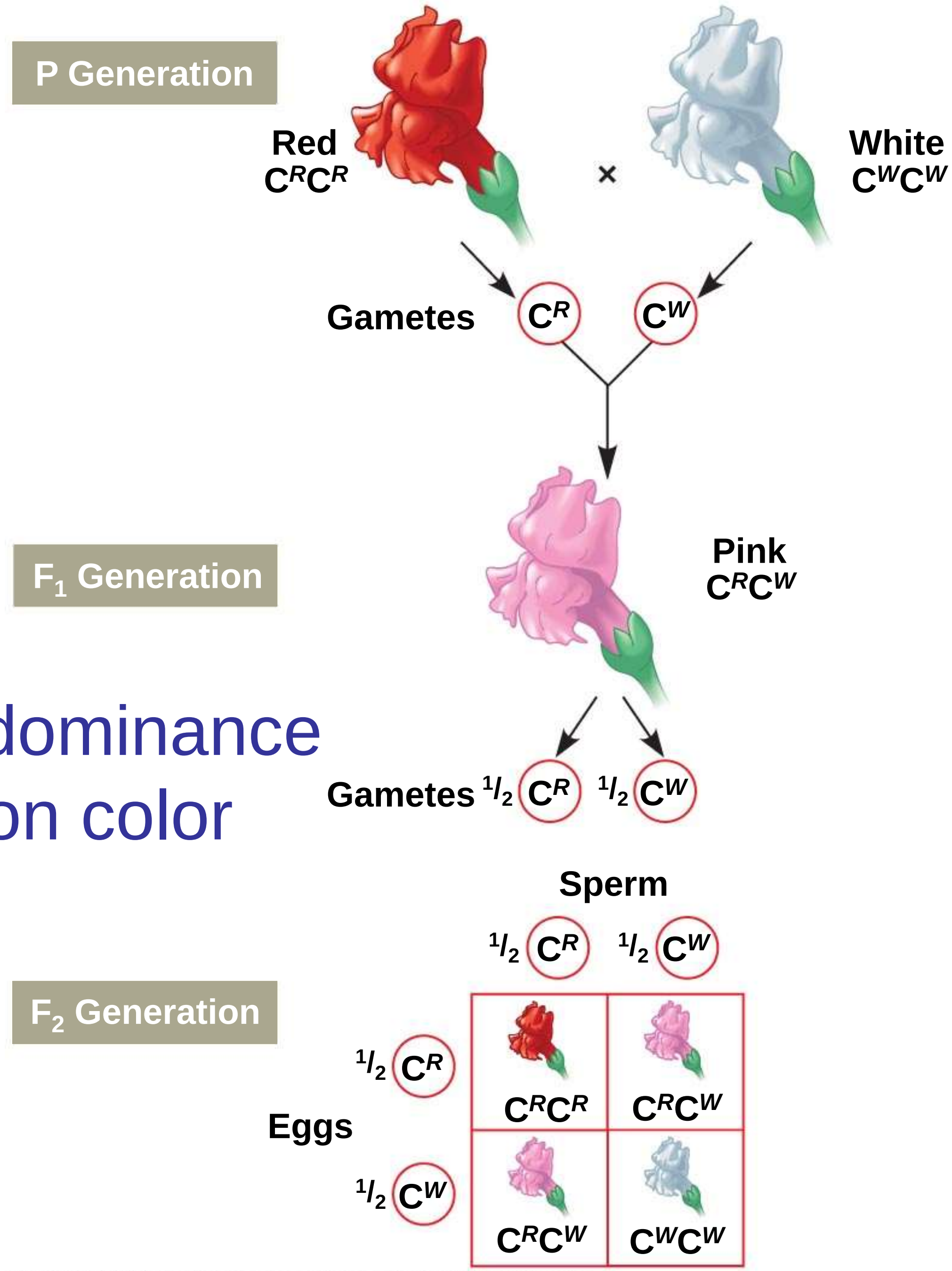

P Generation
Red
C^RC^R
×
White
C^WC^W
Gametes
C^R
C^W
F_1 Generation
Pink
C^RC^W
Gametes
$^1/_2$ C^R
$^1/_2$ C^W

Fig. 14-10



Incomplete dominance in snapdragon color

Incomplete dominance and Codominance

Incomplete

1. the offspring is showing a **3rd phenotype**. The parents each have one, and the offspring are different from the parents.
2. the trait in the offspring is a blend (mixing) of the parental traits.

RED x WHITE
---> PINK

Codominance

1. "CO-" is "together".
2. In CO-dominance, the "recessive" & "dominant" traits appear together in the phenotype of hybrid organisms.

RED x WHITE
---> Red - White Spotted

Key Question in Genetics (遺傳)

When thing goes wrong (or condition changes) during gene passage…

What may happen?

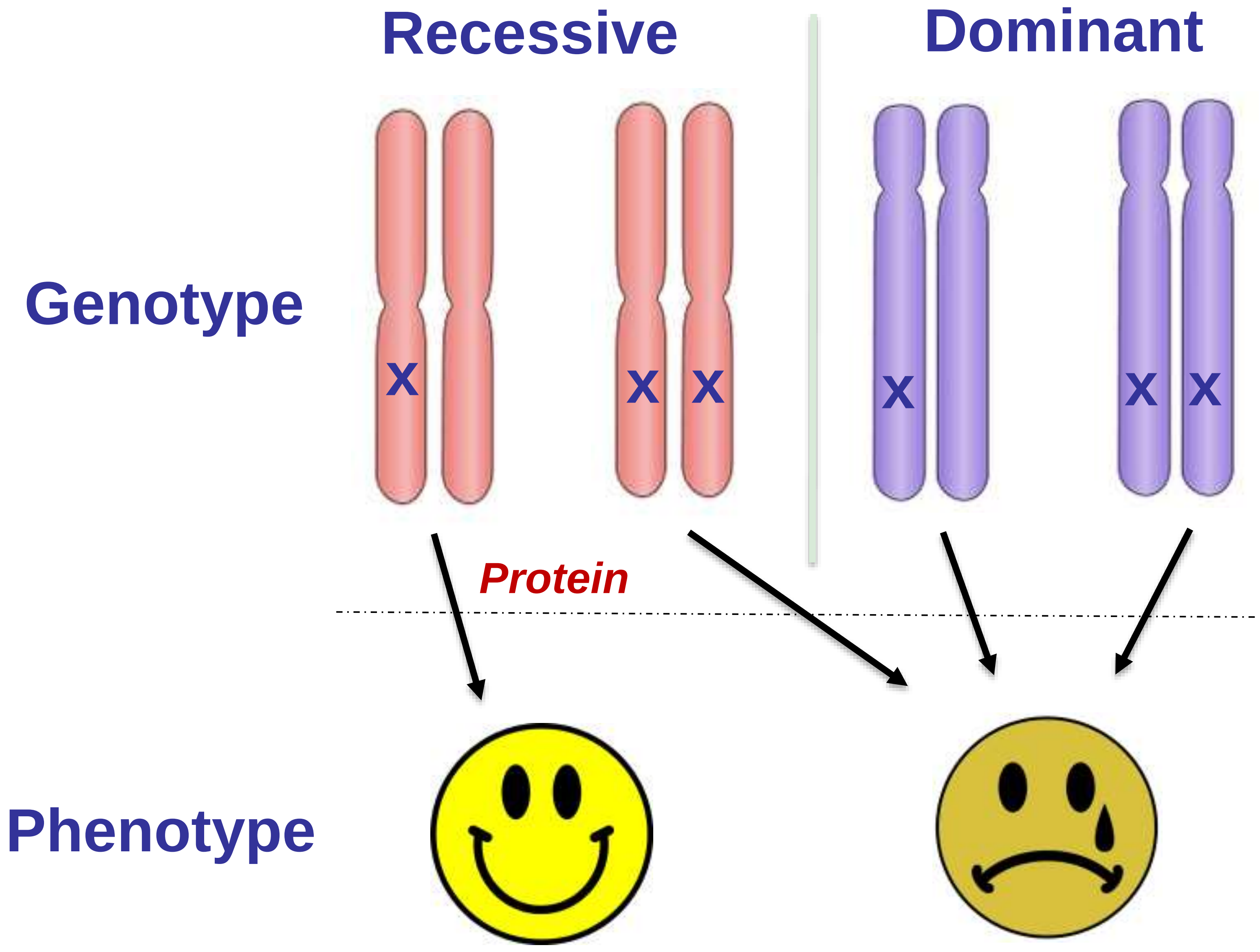
?

Alleles are simply variations in a gene's sequence

The Relation Between Dominance and Phenotype

- A dominant allele does not subdue (壓制) a recessive allele; alleles don't interact
  - Alleles are simply variations in a gene's nucleotide sequence
- For any character, dominance/recessiveness relationships of alleles depend on the level at which we examine the phenotype

Dominant and Recessive mutations



Tay-Sachs Disease – varied degree of dominance at different levels

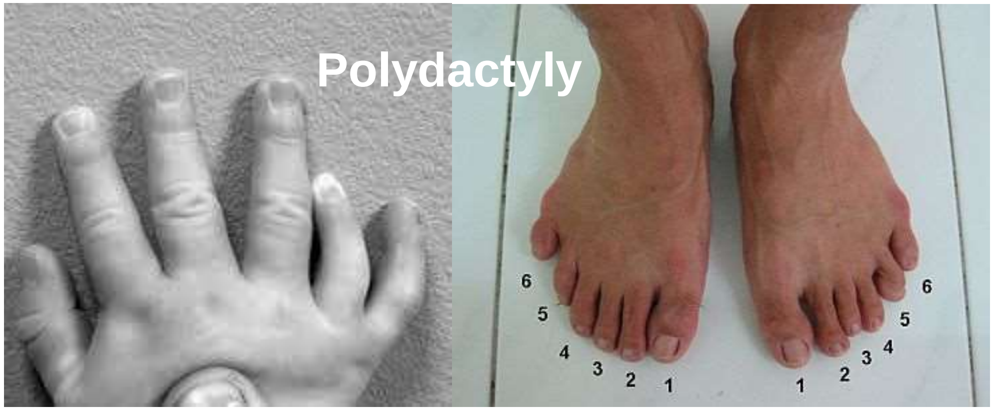
- **Tay-Sachs disease** is fatal; a dysfunctional enzyme causes an accumulation of lipids in the brain – Child with 2 copies of Tay-Sachs allele (homozygotes) has the disease
  - At the *organismal* level, the allele is recessive
  - At the *biochemical* level, the phenotype (i.e., partial loss of the enzyme activity) is incompletely dominant

  - At the *molecular* level, the alleles are codominant (both normal & dysfunctional enzymes are found)

Do you have a extra finger or toe?

Frequency of Dominant Alleles

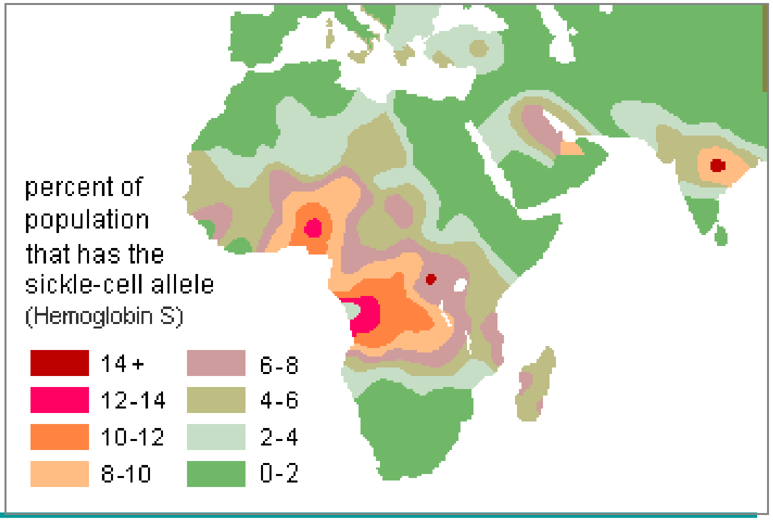
- Dominant alleles are not necessarily more common in populations than recessive alleles
- For example, one baby out of 400 in the United States is born with extra fingers or toes



Prevalence (盛行率；流行度) of a dominant allele

- The allele for this unusual trait is dominant to the allele for the more common trait of five digits per appendage
- In some examples, the recessive allele may be far more prevalent than the population's dominant allele

Sickle cell anemia is a recessive trait. People with one sickle cell gene are said to be carriers.



Multiple Alleles

- Most genes exist in populations in more than two allelic forms

- For example, the four phenotypes of the ABO blood group in humans are determined by three alleles for the enzyme (I) that attaches **A or B carbohydrates** to red blood cells: I^A, I^B, and *i*.

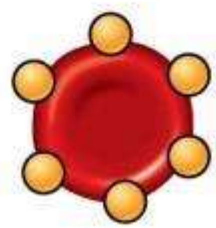
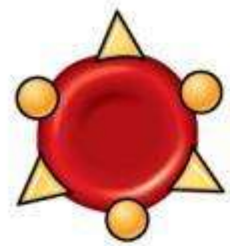
- The enzyme encoded by the $\boldsymbol{I^A}$ allele adds the A carbohydrate, whereas the enzyme encoded by the $\boldsymbol{I^B}$ allele adds the B carbohydrate; the enzyme encoded by the *i* allele adds neither

Figure 14.11

(a) The three alleles for the ABO blood groups and their carbohydrates

Allele	I^A	I^B	i
Carbohydrate	A △	B ○	none

(b) Blood group genotypes and phenotypes

Genotype	I^AI^A or I^Ai	I^BI^B or I^Bi	I^AI^B	ii
Red blood cell appearance				
Phenotype (blood group)	A	B	AB	O

Pleiotropy (多重作用性)

- Most genes have multiple phenotypic effects, a property called **pleiotropy**
- For example, pleiotropic alleles are responsible for the multiple symptoms of certain hereditary diseases, such as
  - cystic fibrosis（囊腫纖維症）缺陷的基因造成身體產生過多的黏液
  - sickle-cell disease（鐮刀型細胞貧血）

Extending Mendelian Genetics for Two or More Genes

- Some traits may be determined by two or more genes

Gene 1

Gene 2

⋮

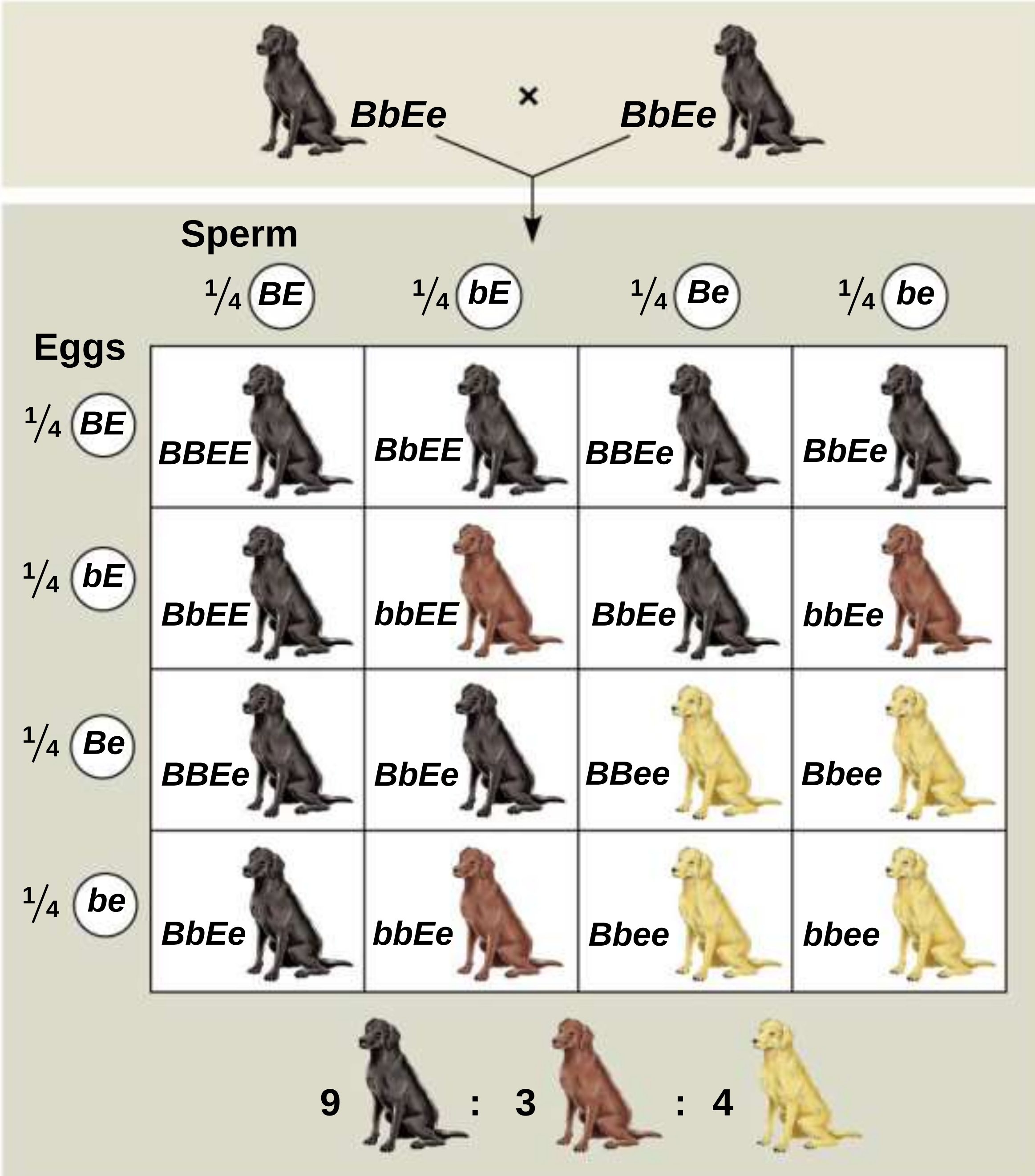
Gene N

} **Trait 1**

Epistasis (離位、越位調控; 基因的上位作用)

- In **epistasis**, a gene at one locus alters the phenotypic expression of a gene at a second locus
- For example, in mice and many other mammals, coat color depends on two genes
  - One gene determines the pigment color (with alleles ***B*** for black and ***b*** for brown)
  - The other gene (with alleles ***C*** for color and ***c*** for no color) determines whether the pigment will be deposited in the hair

Figure 14.12

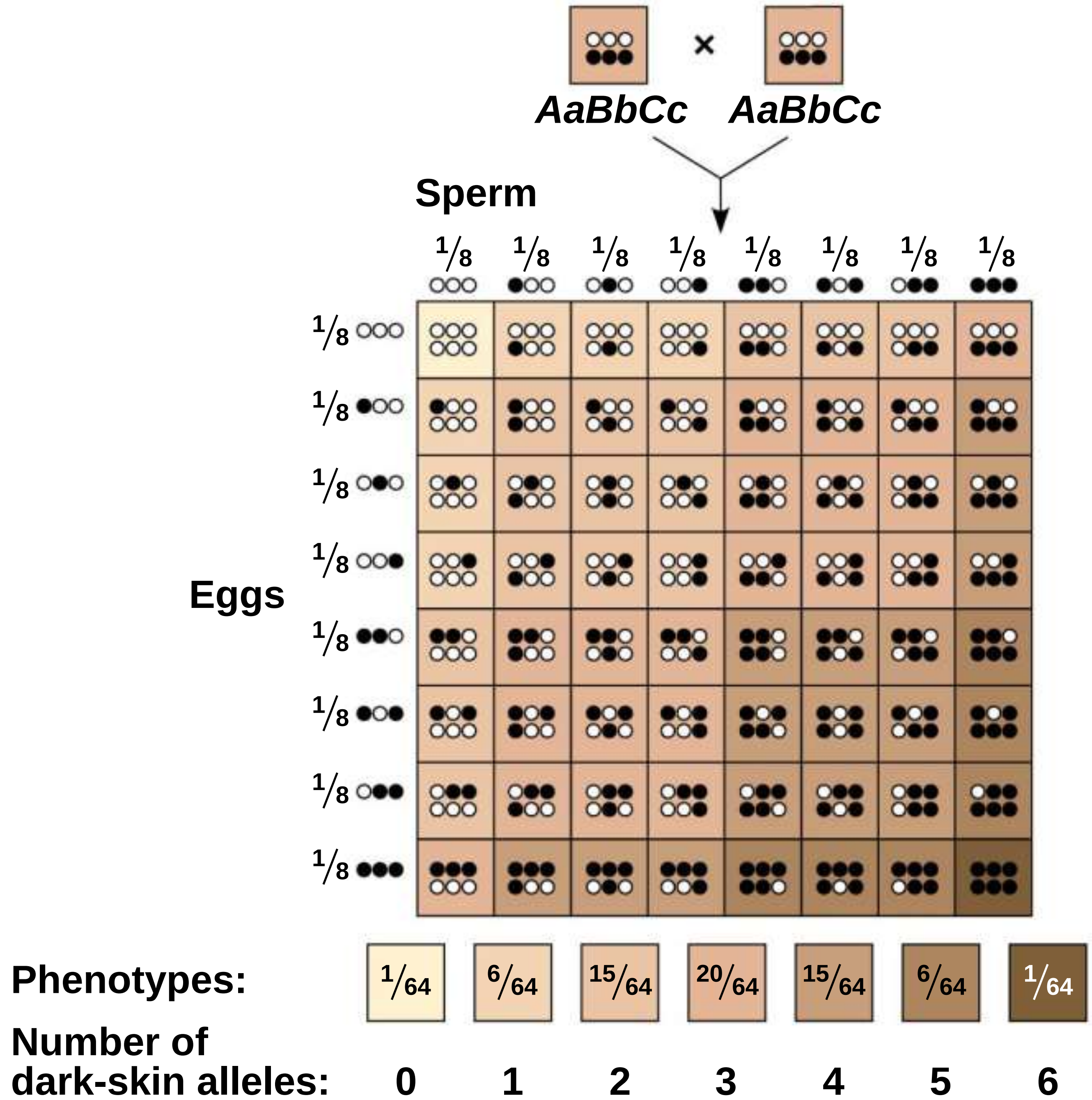


表現黃色毛色的基因對表現黑色或巧克力毛色的基因具有Epistasis /上位作用

Polygenic Inheritance

- **Quantitative characters** are those that vary in the population along a continuum
- Quantitative variation usually indicates **polygenic inheritance**, an additive effect of two or more genes on a single phenotype
- Skin color in humans is an example of polygenic inheritance

Figure 14.13



AaBbCc
×
AaBbCc
Sperm
Eggs
1/8
Phenotypes:
1/64
6/64
15/64
20/64
15/64
6/64
1/64
Number of dark-skin alleles:
0
1
2
3
4
5
6

Nature and Nurture: The Environmental Impact on Phenotype

- Another departure from Mendelian genetics arises when the phenotype for a character depends on **environment** as well as genotype
- The **norm of reaction** is the phenotypic range of a genotype influenced by the environment
- For example, hydrangea flowers (繡球花) of the same genotype range from blue-violet to pink, depending on soil acidity…

Fig. 14-14

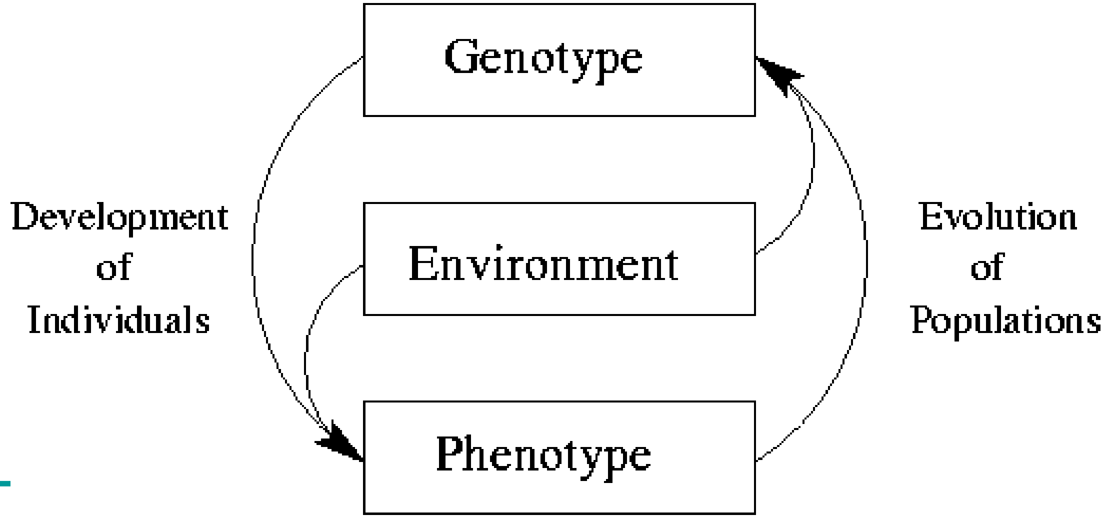
繡球花 (Hydrangea) -- 花色可隨土壤的pH值而改變

若在鹼性土壤 **(basic soil)** 種植**(pH**值比**7**大**)**，花色是粉紅； 若在酸性土壤 **(acidic soil)** 種植**(pH**值比**7**小**)**，花色是藍紫色。

- Norms of reaction are generally broadest for polygenic characters
- Such characters are called **multifactorial**多因素 because genetic and environmental factors collectively influence phenotype.

Integrating a Mendelian View of Heredity and Variation

- An organism's phenotype includes:
  - physical appearance, internal anatomy, physiology, and behavior
- An organism's phenotype reflects its overall genotype and unique environmental history



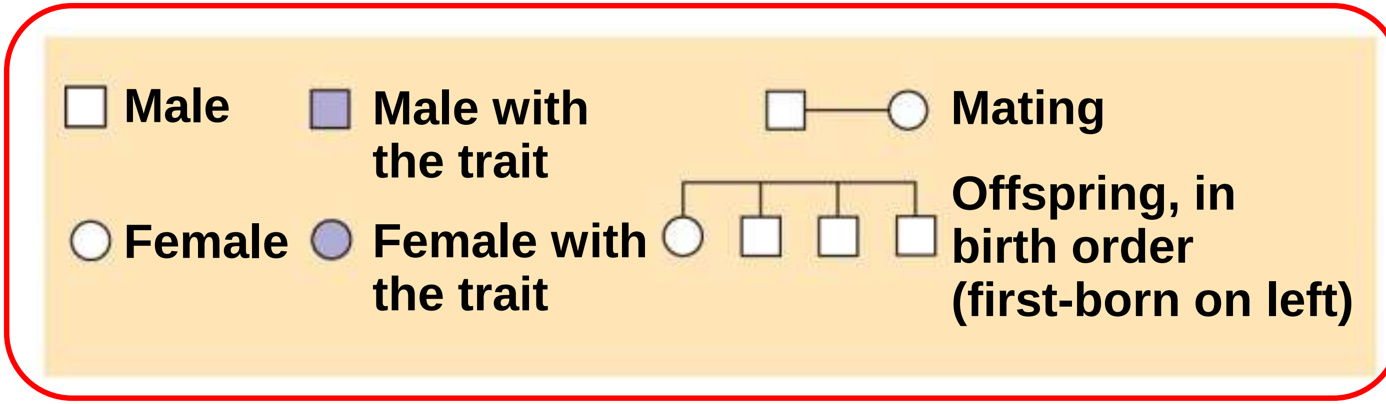
Concept 14.4: Many human traits follow Mendelian patterns of inheritance

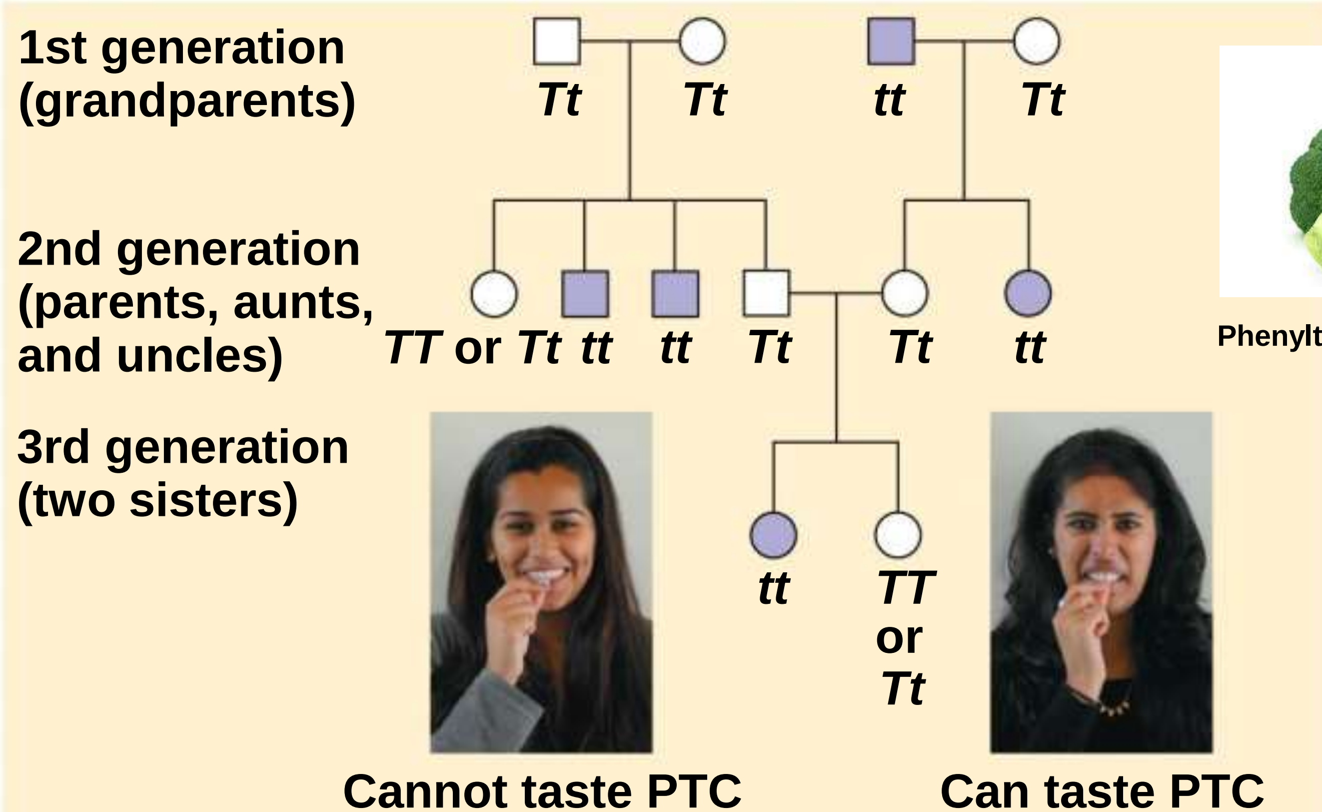
- Humans are not good subjects for genetic research
  - Generation time is too long
  - Parents produce relatively few offspring
  - Breeding experiments are unacceptable
- However, basic Mendelian genetics endures as the foundation of human genetics

Pedigree Analysis (族譜)

- A **pedigree** is a family tree that describes the inter-relationships of parents and children across generations
- Inheritance patterns of particular traits can be traced and described using pedigrees

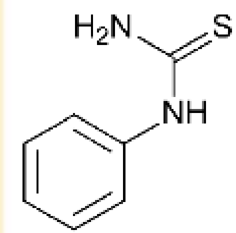
Figure 14.15b
Ed11





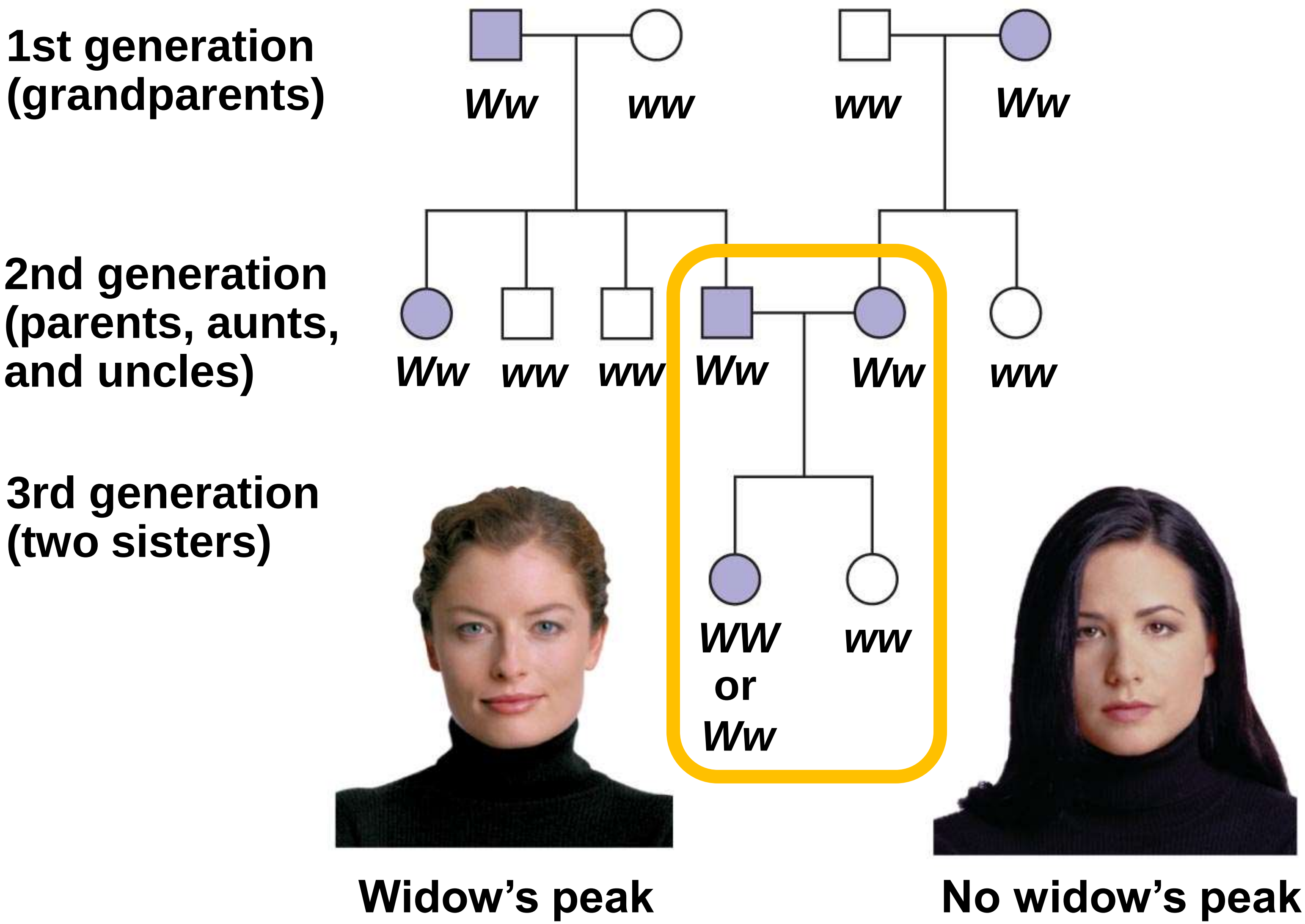
(b) Is the inability to taste a chemical called PTC a dominant or recessive trait? → **Recessive**

Phenylthiocarbamide (PTC)



一個人能否嘗出PTC苦澀味是由其基因決定的

Fig. 14-15 2

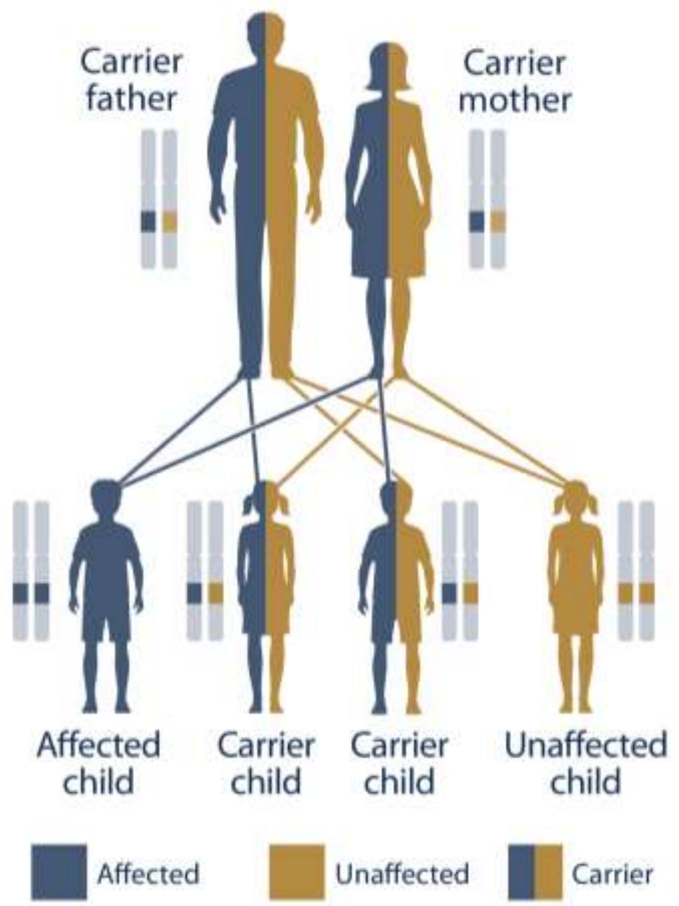


(a) Is a widow's peak a dominant or recessive trait? → **dominant**

- Pedigrees can also be used to make **predictions** about future offspring
- We can use the multiplication and addition rules to predict the probability of specific phenotypes

Recessively Inherited Disorders

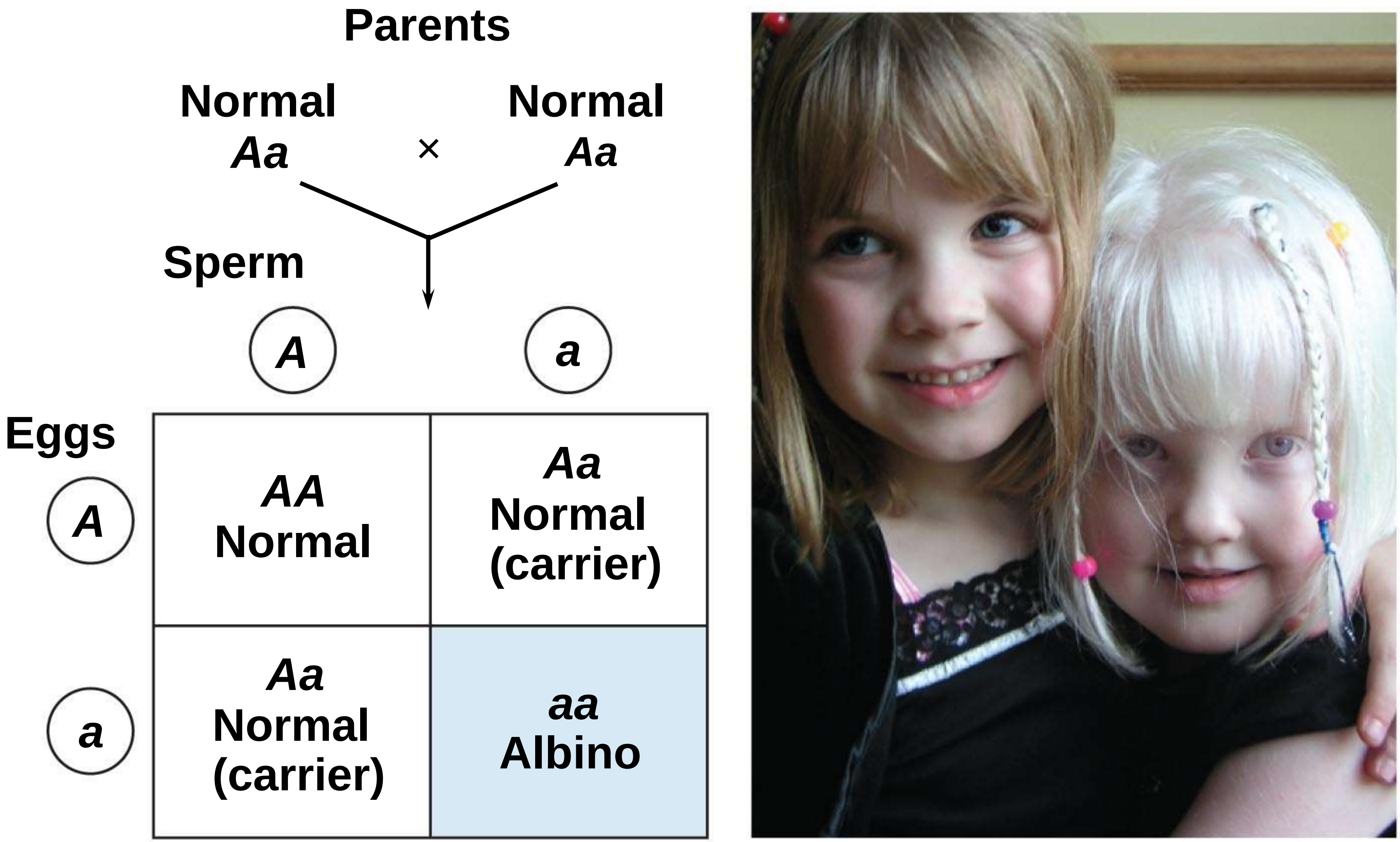
- Many genetic disorders are inherited in a recessive manner



The Behavior of Recessive Alleles

- Recessively inherited disorders show up only in individuals homozygous for the allele
- **Carriers** are heterozygous individuals who carry the recessive allele but are phenotypically normal (i.e., pigmented)
- Albinism is a recessive condition characterized by a lack of pigmentation in skin and hair

Fig. 14-16


Parents
Normal
Aa
×
Normal
Aa
Sperm
A
a
Eggs
A
a
AA
Normal
Aa
Normal
(carrier)
Aa
Normal
(carrier)
aa
Albino

Why no consanguineous mating? [kon-**sang**-gwin-ee-*uh* s]

- If a recessive allele that causes a disease is rare, then the chance of two carriers meeting and mating is low

近親

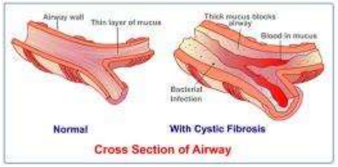
- Consanguineous matings (i.e., matings between close relatives) increase the chance of mating between two carriers of the same rare allele

- Most societies and cultures have laws or taboos against marriages between close relatives

Lethal genetic disease - *Cystic Fibrosis* 囊狀纖維化

- **Cystic fibrosis** is the most common lethal genetic disease in the United States,striking one out of every 2,500 people of European descent

- The cystic fibrosis allele results in defective or absent chloride transport channels in plasma membranes



- Symptoms include **mucus buildup** in some internal organs and abnormal absorption of nutrients in the small intestine

Evolutionary advantage - *Sickle-Cell Disease* 鎌刀型貧血症

- **Sickle-cell disease** affects one out of 400 African-Americans
- The disease is caused by the substitution of a single amino acid in the hemoglobin protein in red blood cells
- Symptoms include physical weakness, pain, organ damage, and even paralysis

Reduction of malaria symptoms in heterozygote individual with sickle-cell trait!

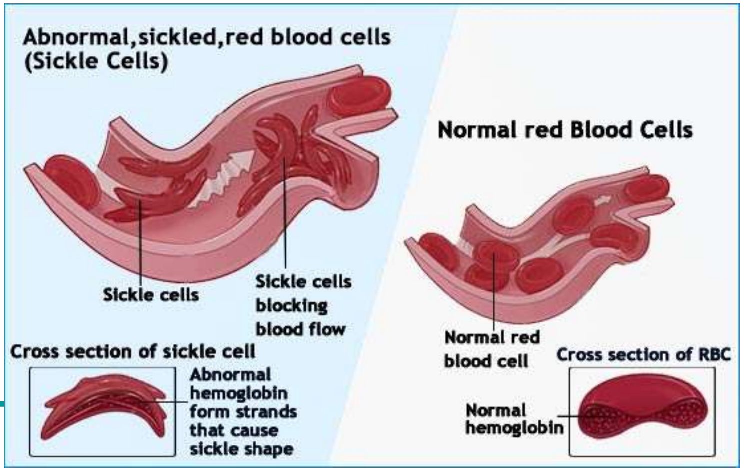
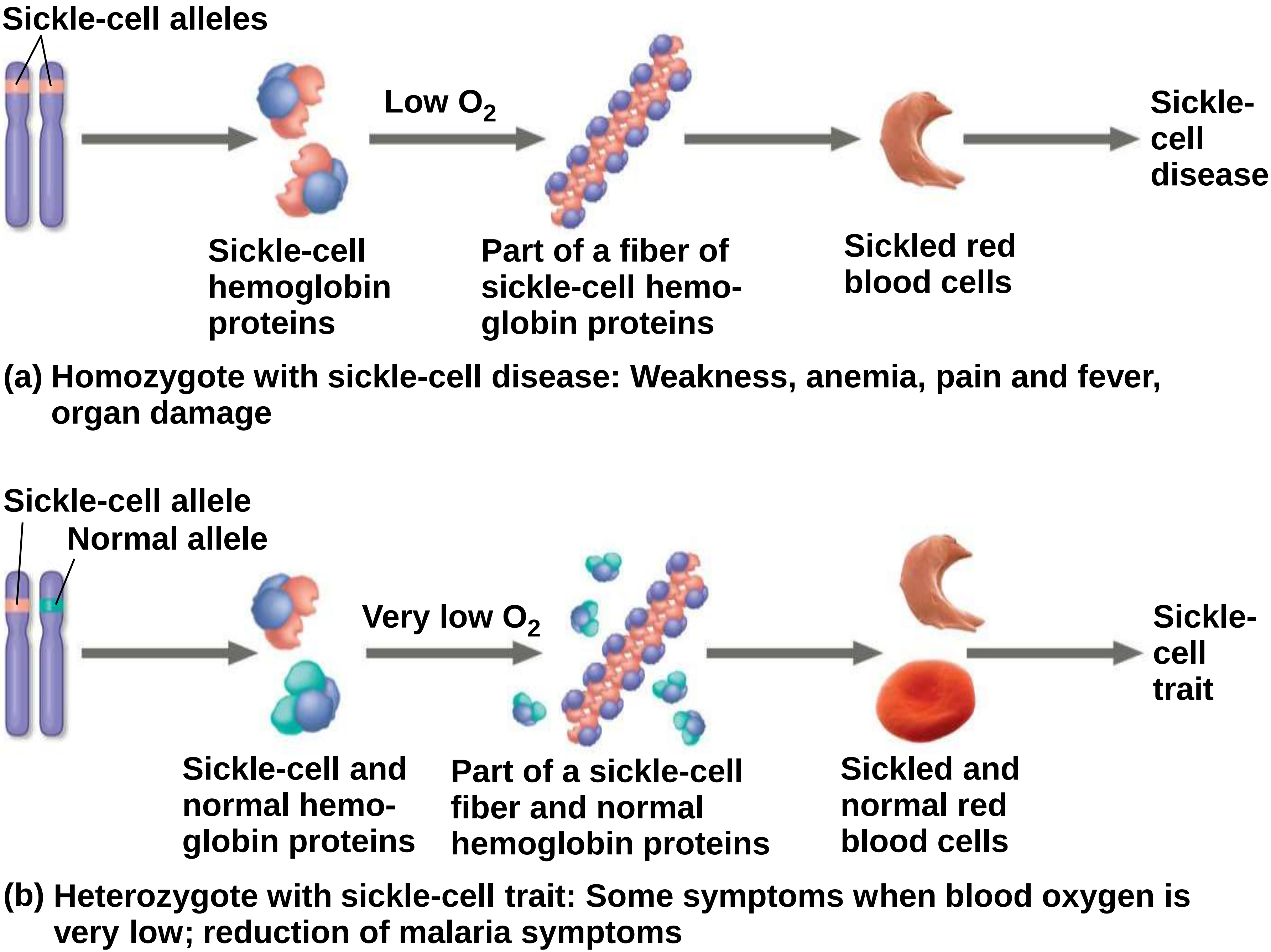


Figure 14.17



(a) Homozygote with sickle-cell disease: Weakness, anemia, pain and fever, organ damage

(b) Heterozygote with sickle-cell trait: Some symptoms when blood oxygen is very low; reduction of malaria symptoms

Dominantly Inherited Disorders

- Some human disorders are caused by dominant alleles
- Dominant alleles that cause a lethal disease are rare and arise by mutation
- *Achondroplasia* [ey-kon-dr*uh*-**pley**-zh*uh*] 軟骨發育不全 is a form of dwarfism caused by a rare dominant allele

Achondroplasia: absence of cartilage formation results in bone growth disorder

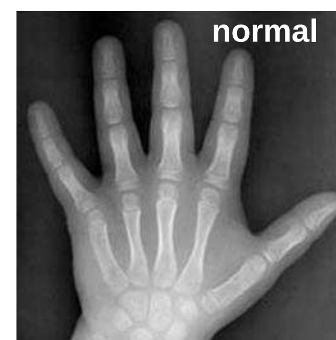


Figure 14.17

Achondroplasia: a dominant trait

Parents

Dwarf *Dd* × Normal *dd*

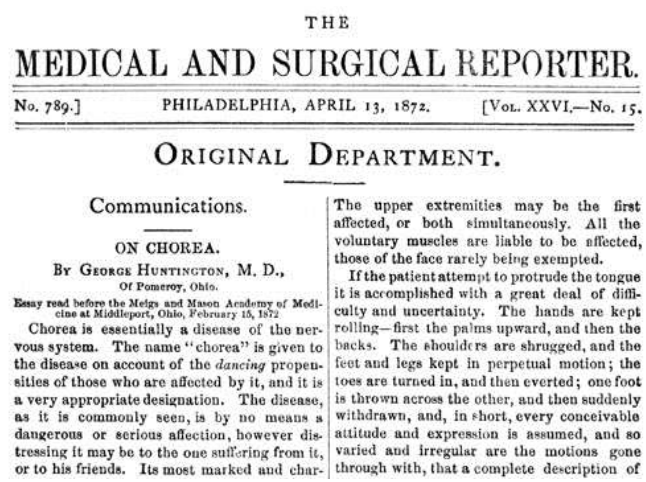
Sperm / Eggs

Eggs \ Sperm	*D*	*d*
d	*Dd* Dwarf	*dd* Normal
d	*Dd* Dwarf	*dd* Normal

Huntington's Disease 亨丁頓舞蹈症

Late-onset disease with lethal dominant allele

- **Huntington's disease** is a degenerative disease of the nervous system
- The disease has no obvious phenotypic effects until the individual is about 35 to 40 years of age



THE

MEDICAL AND SURGICAL REPORTER.

No. 789.] PHILADELPHIA, APRIL 13, 1872. [Vol. XXVI.—No. 15.

ORIGINAL DEPARTMENT.

Communications.

ON CHOREA.

By George Huntington, M. D.,

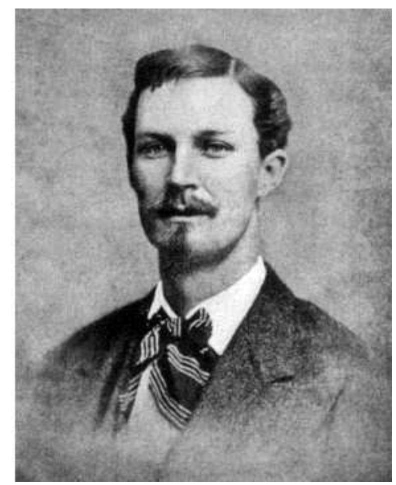
Of Pomeroy, Ohio.

Essay read before the Meigs and Mason Academy of Medicine at Middleport, Ohio, February 15, 1872

Chorea is essentially a disease of the nervous system. The name "chorea" is given to the disease on account of the *dancing* propensities of those who are affected by it, and it is a very appropriate designation. The disease, as it is commonly seen, is by no means a dangerous or serious affection, however distressing it may be to the one suffering from it, or to his friends. Its most marked and char-

The upper extremities may be the first affected, or both simultaneously. All the voluntary muscles are liable to be affected, those of the face rarely being exempted.

If the patient attempt to protrude the tongue it is accomplished with a great deal of difficulty and uncertainty. The hands are kept rolling—first the palms upward, and then the backs. The shoulders are shrugged, and the feet and legs kept in perpetual motion; the toes are turned in, and then everted; one foot is thrown across the other, and then suddenly withdrawn, and, in short, every conceivable attitude and expression is assumed, and so varied and irregular are the motions gone through with, that a complete description of



In 1872 George Huntington described the disorder in his first paper "On Chorea" at the age of 22

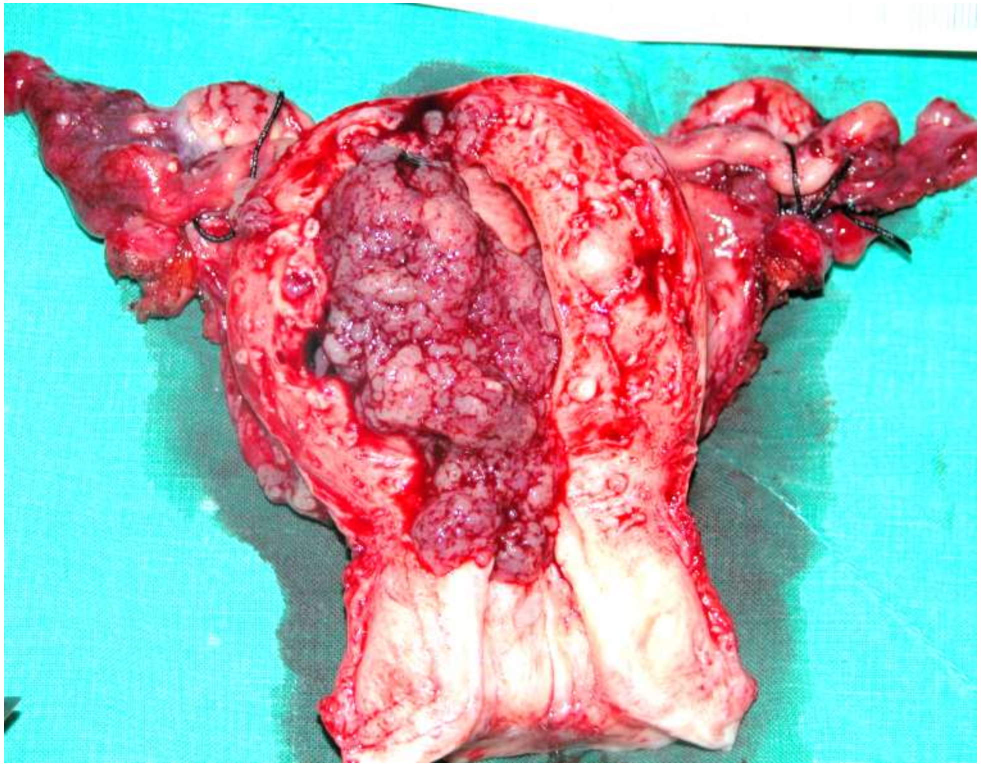
Multifactorial Disorders

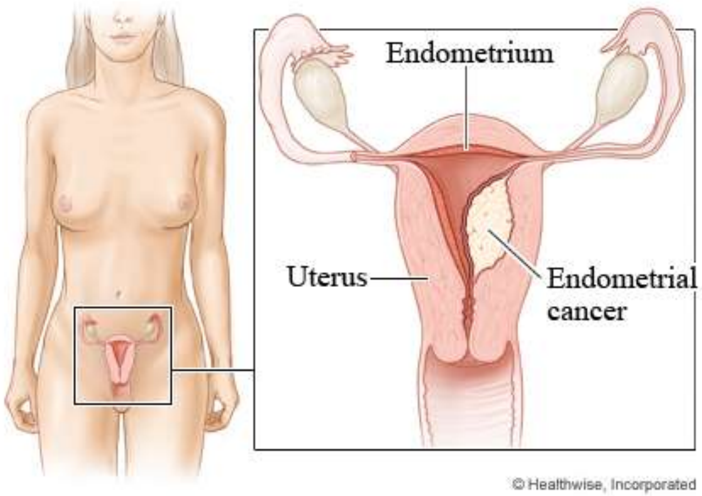
- Many diseases, such as heart disease and cancer, have both genetic and environmental components
- Little is understood about the genetic contribution to most multifactorial diseases

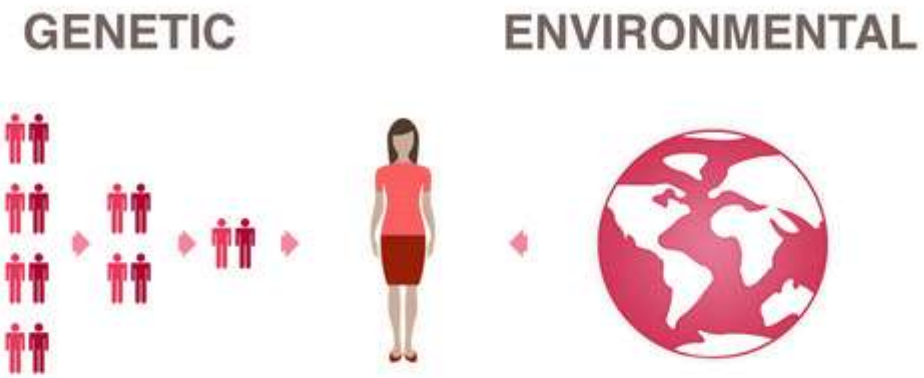
(bloody picture next)

An example of Multifactorial disorder - Endometrial Ovarian Cancer

MMH Hsinchu, October 2004



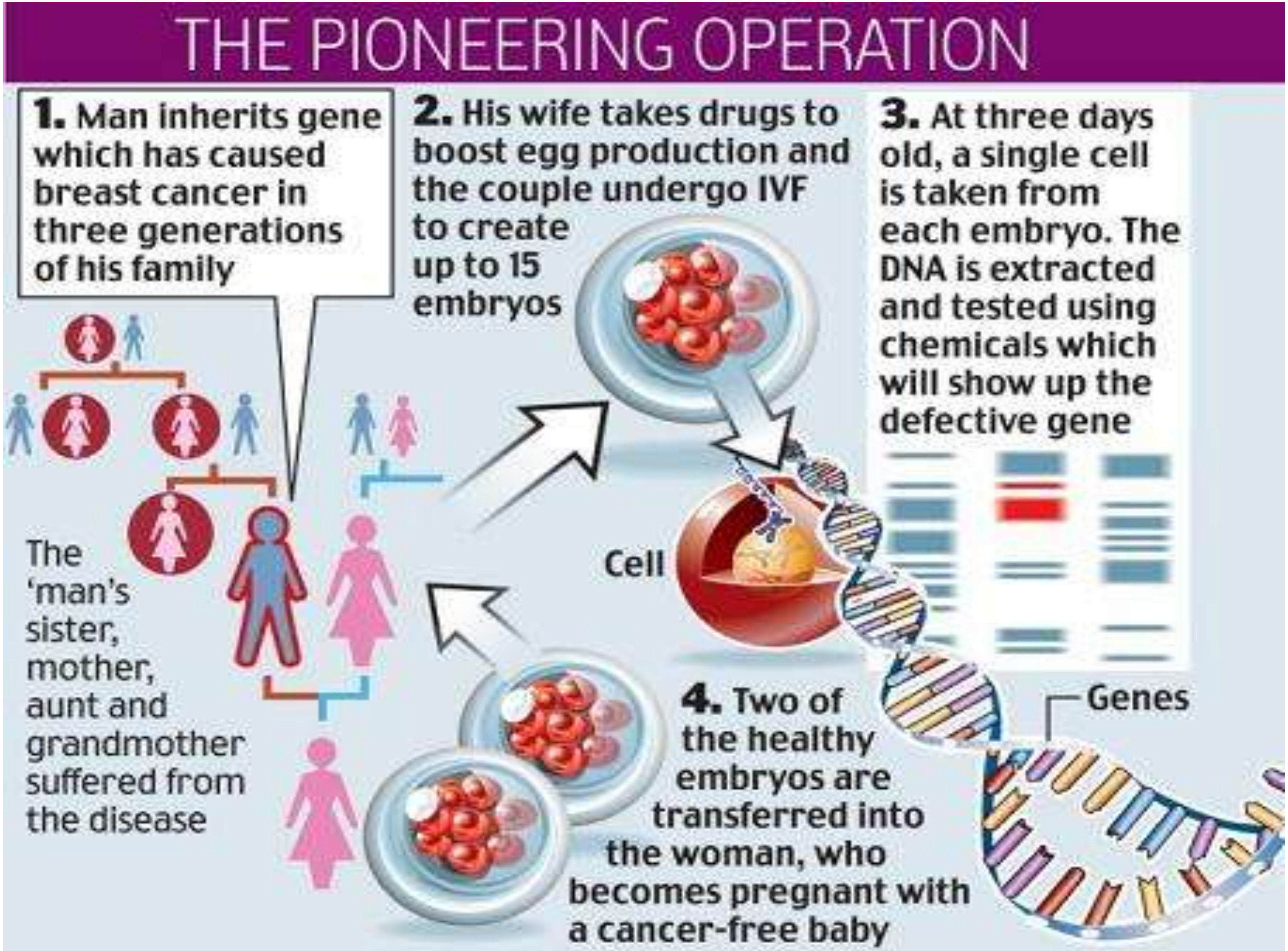




Genetic Testing and Counseling

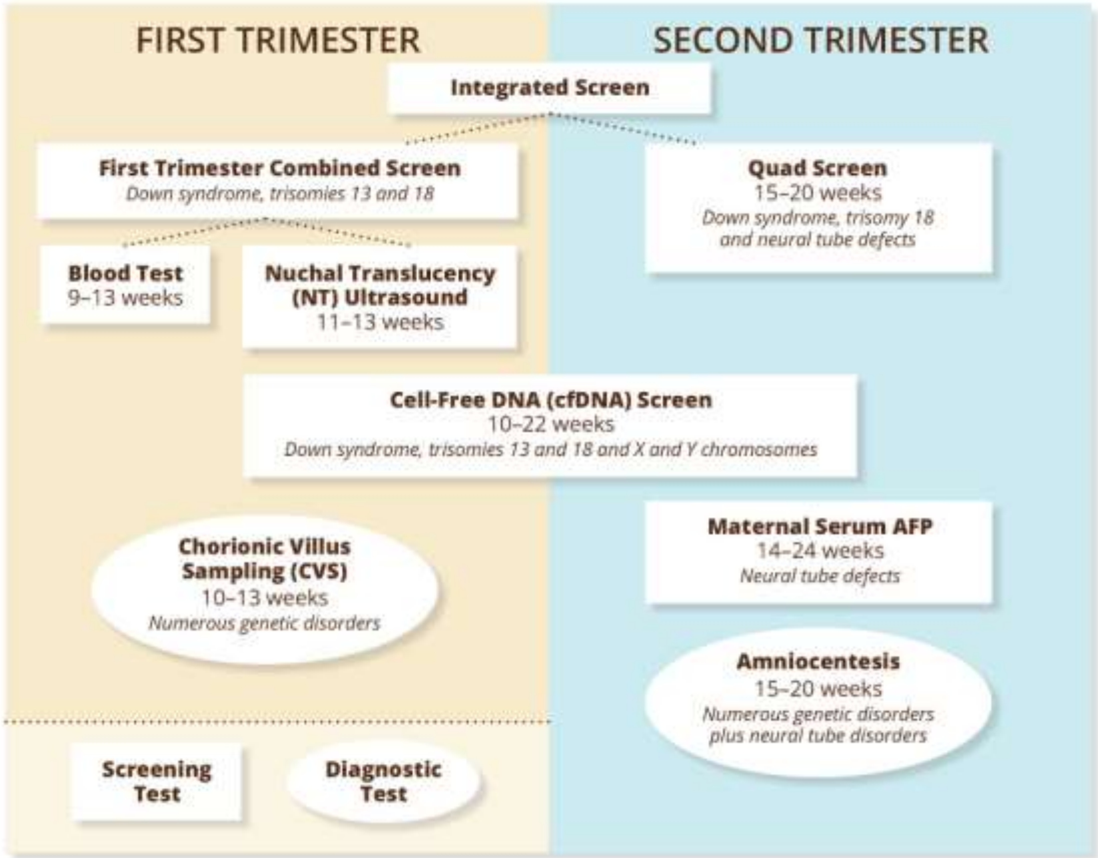
- Genetic counselors can provide information to prospective parents concerned about a family history for a specific disease

Designer Babies: The Fertility Institutes



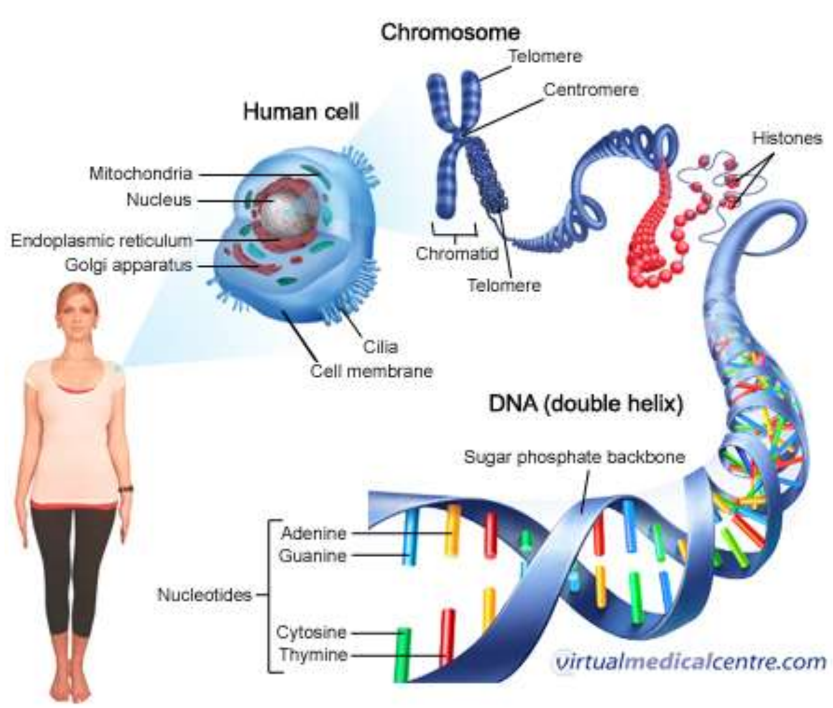
Counseling Based on Mendelian Genetics and Probability Rules

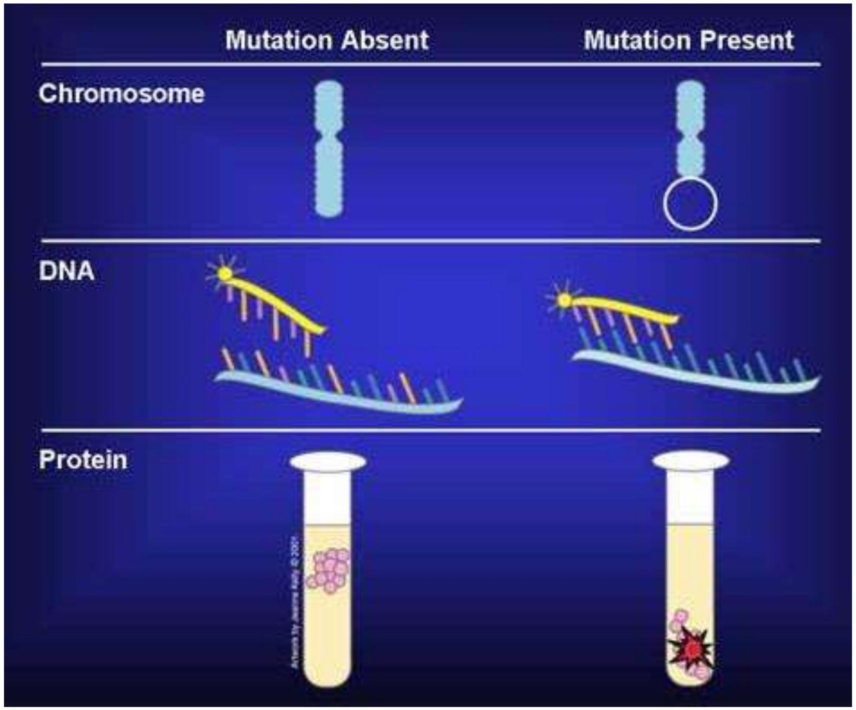
- Using family histories, genetic counselors help couples determine the odds that their children will have genetic disorders



Tests for Identifying Carriers

- For a growing number of diseases, tests are available that identify carriers and help define the odds more accurately





Fetal Testing

- In **amniocentesis** 羊膜穿刺術, the liquid that bathes the fetus is removed and tested
- In **chorionic villus sampling (CVS**) 絨膜絨毛取樣, a sample of the placenta is removed and tested

See illustration on next page

- Other techniques, such as ***ultrasound*** and ***fetoscopy*** 胎兒鏡, allow fetal health to be assessed visually in utero

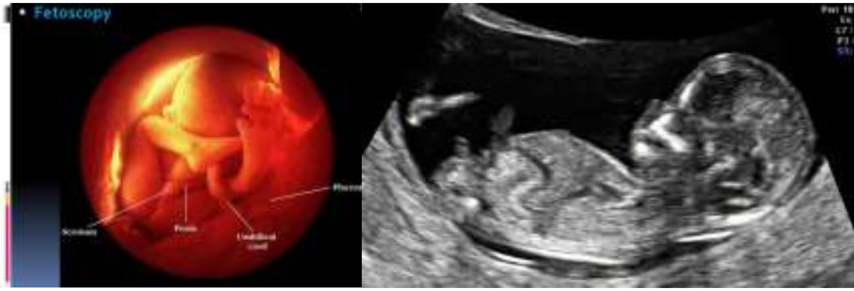
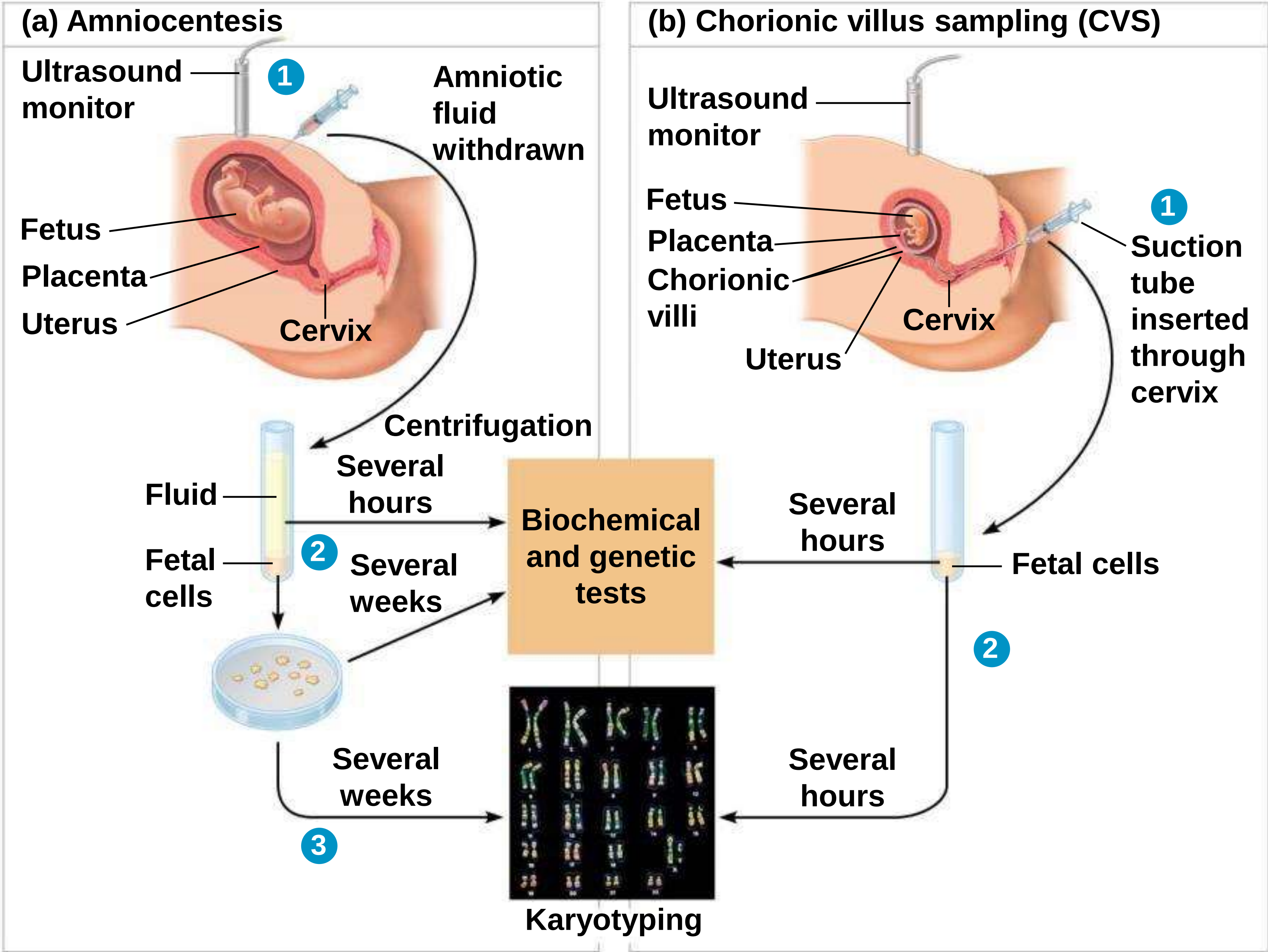


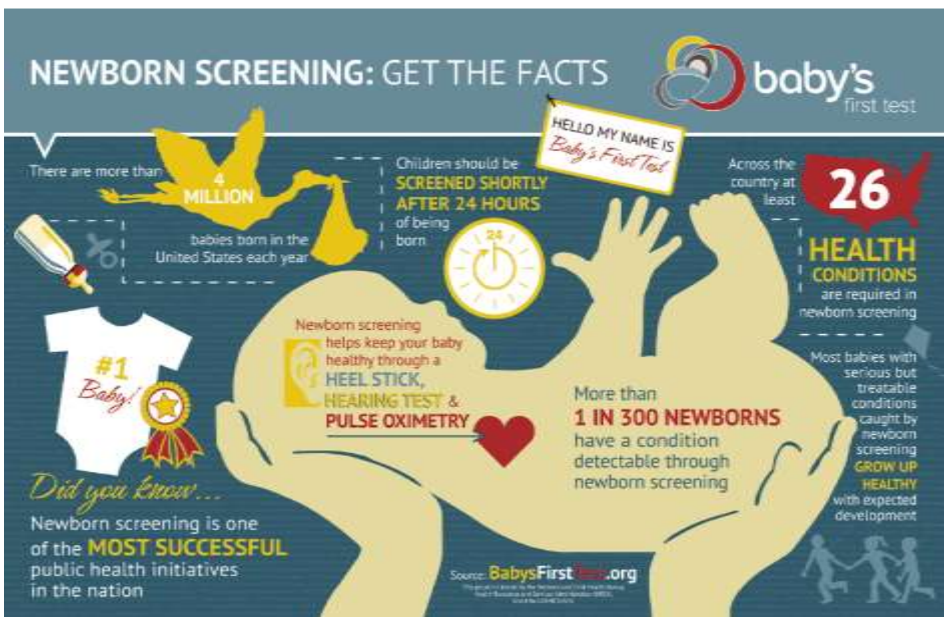
Figure 14.19



(a) Amniocentesis
Ultrasound monitor
1
Amniotic fluid withdrawn
Fetus
Placenta
Uterus
Cervix
Centrifugation
Fluid
Several hours
Fetal cells
2
Several weeks
Biochemical and genetic tests
Several weeks
3
Karyotyping
(b) Chorionic villus sampling (CVS)
Ultrasound monitor
Fetus
Placenta
Chorionic villi
Uterus
Cervix
1
Suction tube inserted through cervix
Several hours
Fetal cells
2
Several hours

Newborn Screening

- Some genetic disorders can be detected at birth by simple tests that are now routinely performed in most hospitals



新生兒先天代謝異常疾病篩檢一覽表

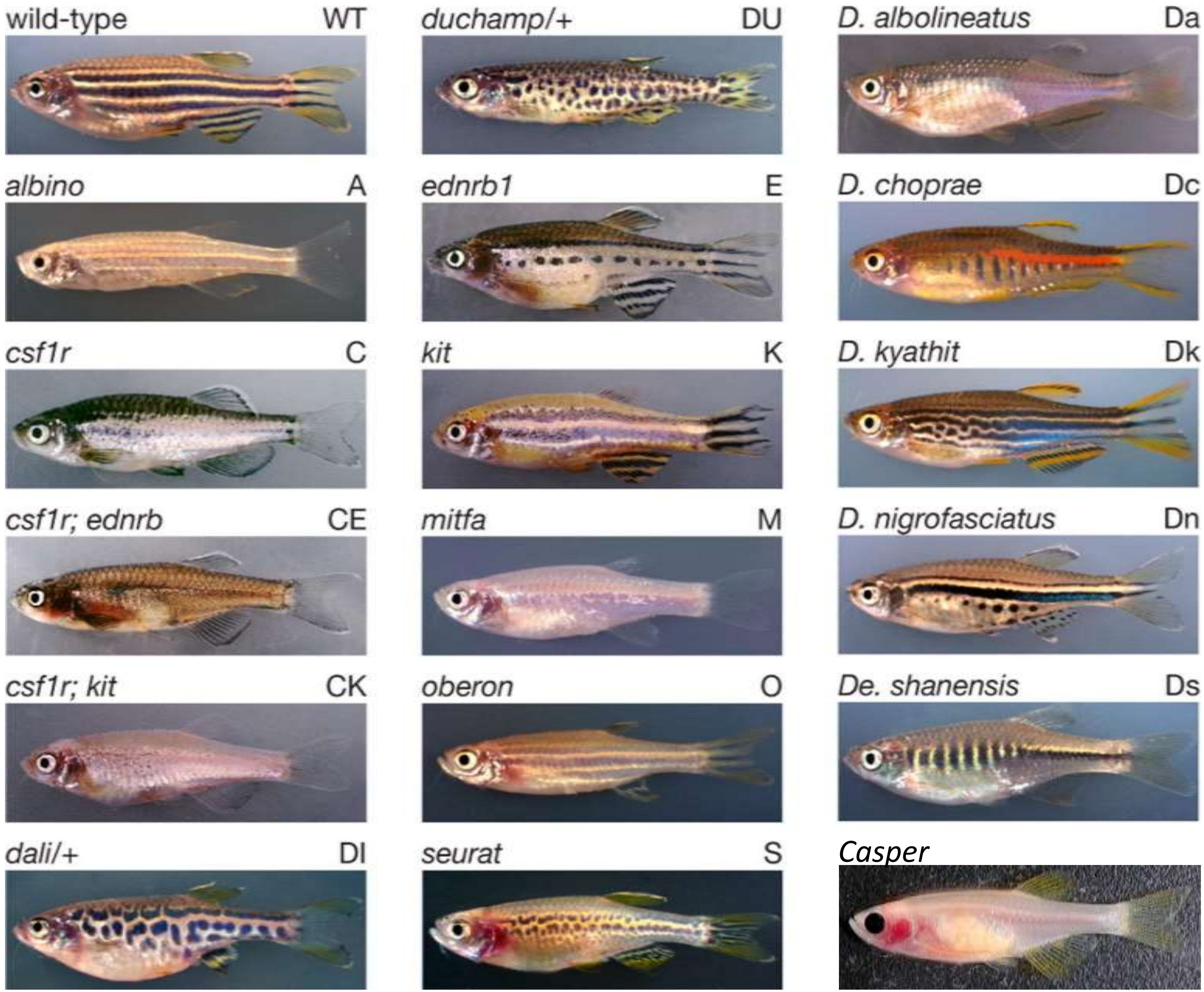
串聯質譜儀篩檢代謝疾病	非串聯質譜儀篩檢代謝疾病
MS01中鏈醯輔酶A去氫酶缺乏症	CHT先天性甲狀腺低能症
MS02短鏈脂肪酸代謝異常	GAL半乳糖血症
MS03長鏈脂肪酸代謝異常	G6PD葡萄糖-6-磷酸鹽去氫酵素缺乏症（蠶豆症）
MS04卡尼丁（肉鹼）吸收障礙	CAH先天性腎上腺增生症
MS05卡尼丁結合酵素缺乏	POMPE龐貝氏症
MS06卡尼丁穿透障礙	FABRY法布瑞氏症
MS07丙酸血症	SCID嚴重複合型免疫缺乏症
MS08甲基丙二酸血症	AADC芳香族 L-胺基酸類脫羧基缺乏症（尚未全面推廣）
MS09異戊酸血症	說明 1. 所有表列項目為目前國內可做的篩檢。 2. 有底色標示者為國健署補助的11項檢查。 3. 「串聯質譜儀」可一次篩檢出有機酸、胺基酸、脂肪酸等23項代謝疾病，但目前僅有7項在國民健康署補助篩檢項目內，因此，父母只會看到7項疾病的篩檢結果。如果父母希望同時看到其他項目結果，必須先填寫「新生兒篩檢先趨計畫同意書」，不需額外採血及增加費用，即可進行篩檢並獲得結果。 4. 在「非串聯質譜儀篩檢代謝疾病」中的「龐貝氏症」，目前為自費篩檢項目，選擇「龐貝氏症」可免費加驗先趨篩檢「法布瑞氏症」，家長可選擇放棄「法布瑞氏症」篩檢，但不能只檢驗「法布瑞氏症」。 5. 建議新生兒接種卡介苗前，考慮先進行「SCID嚴重複合型免疫缺乏症」篩檢，確認無病後，再施打包括卡介苗。
MS10戊二酸血症第一型	
MS11C5-OH上升相關疾病	
MS12楓糖尿症	
MS13瓜胺酸血症（第一、二型）	
MS14酪胺酸血症（第二、三型）	
MS15精胺丁二酸酶缺乏症	
MS17精胺酸血症	
MS18高鳥氨酸-高血氨-高瓜胺酸綜合症候群（HHH症候群）	
MS19其他疾病	
MS20苯酮尿症	
MS21高胱胺酸尿症及高甲硫胺酸血症	
MS22極長鏈醯輔酶A去氫酶缺乏症	
MS23戊二酸血症第二型	

You should now be able to:

1. Define the following terms: true breeding, hybridization, monohybrid cross, P generation, F_1 generation, F_2 generation
2. Distinguish between the following pairs of terms: dominant and recessive; heterozygous and homozygous; genotype and phenotype
3. Use a Punnett square to predict the results of a cross and to state the phenotypic and genotypic ratios of the F_2 generation

4. Explain how phenotypic expression in the heterozygote differs with complete dominance, incomplete dominance, and codominance
5. Define and give examples of pleiotropy and epistasis
6. Explain why lethal dominant genes are much rarer than lethal recessive genes
7. Explain how carrier recognition, fetal testing, and newborn screening can be used in genetic screening and counseling

Diverse mutant lines of zebrafish and closely related species



Engeszer R E et al. PNAS 2008;105:929-933

Transparent zebrafish- Casper

Developed in Leonard Zon's Lab

Children's Hospital Boston

Cell Stem Cells 2008

roy$^{-/-}$ x nacre$^{-/-}$

(no iridophore : no melanocyte)

VS.

AB wild type

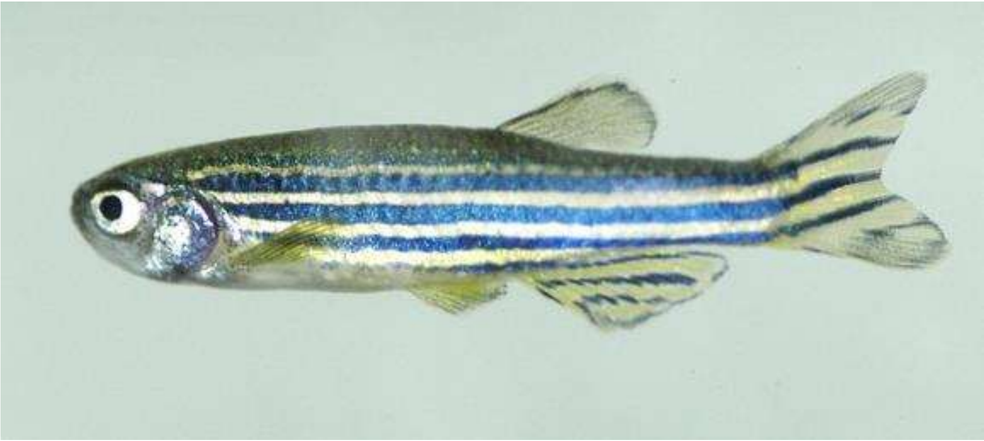
X

No melanocyte X No iridophore

Citrine

ZeTH Transparent zebafish – Citrine 琉璃斑馬魚品系

Citrine（黃水晶）：加強人們**理智體**，使人的行事作為更為邏輯、科學、條理，主偏財，屬**財富水晶**。



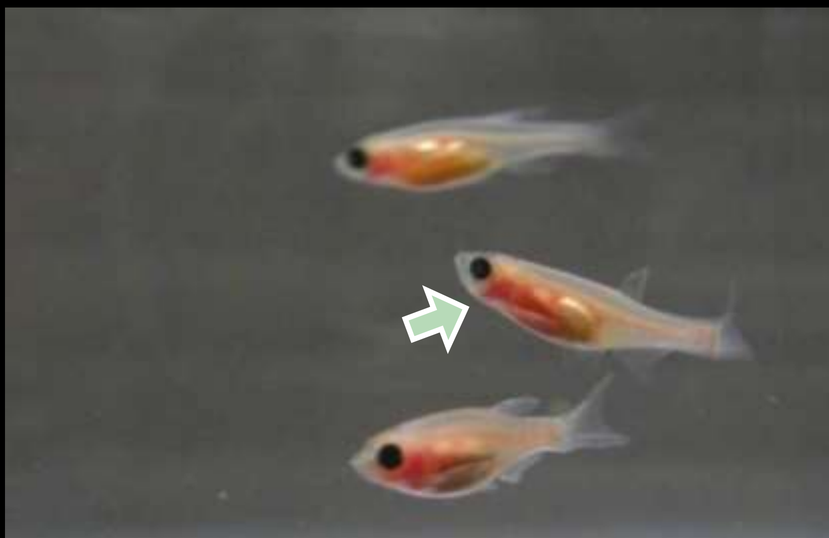
Children's Hospital Boston

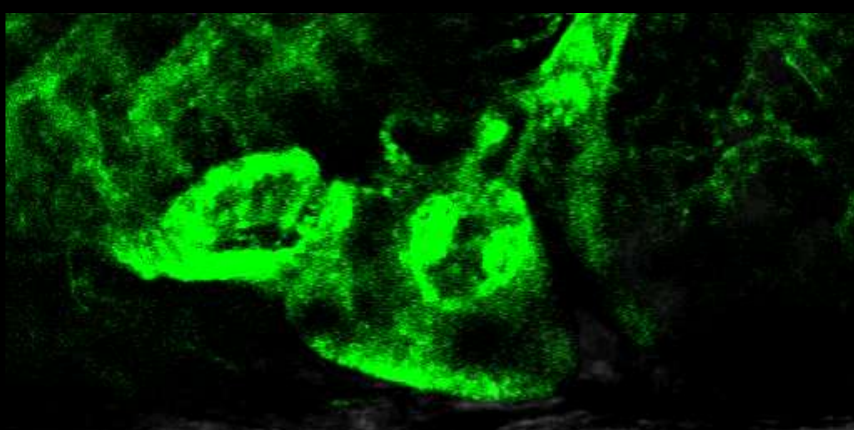
Transparent zebrafish - Casper

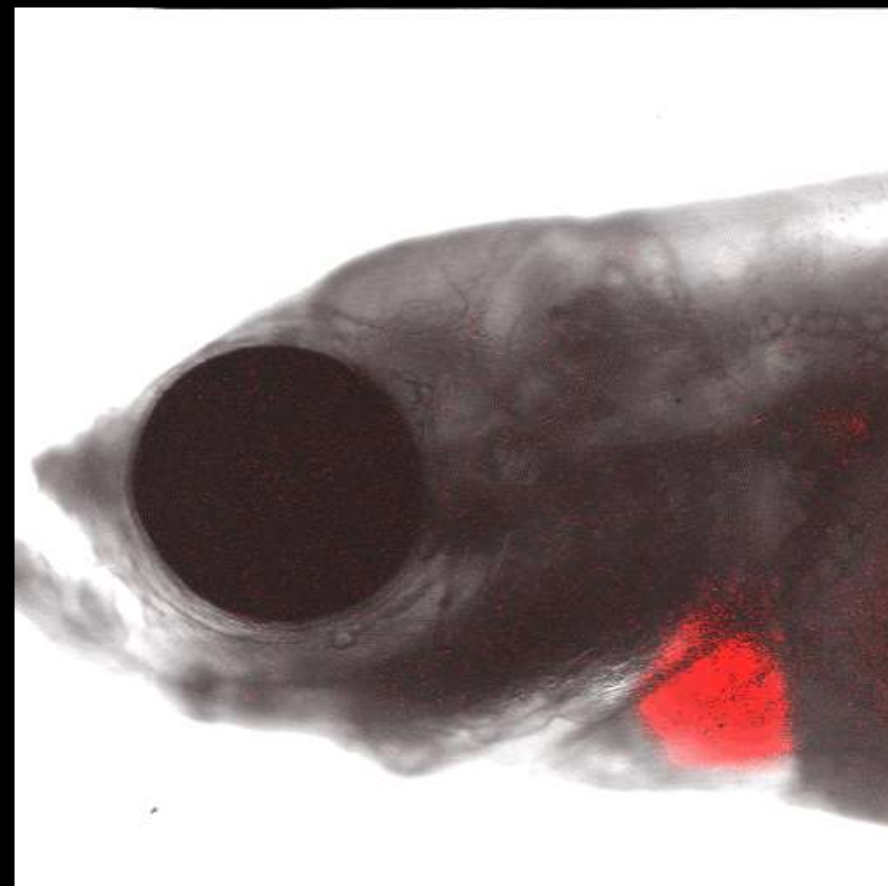
AB Strain Zebrafish


Transparent zebrafish line – Citrine
YJ Chuang Lab, Department of Medical Science, National Tsing Hua University
Photo credit: Jennifer Hsia

Real time observation of Internal Organ- Heart

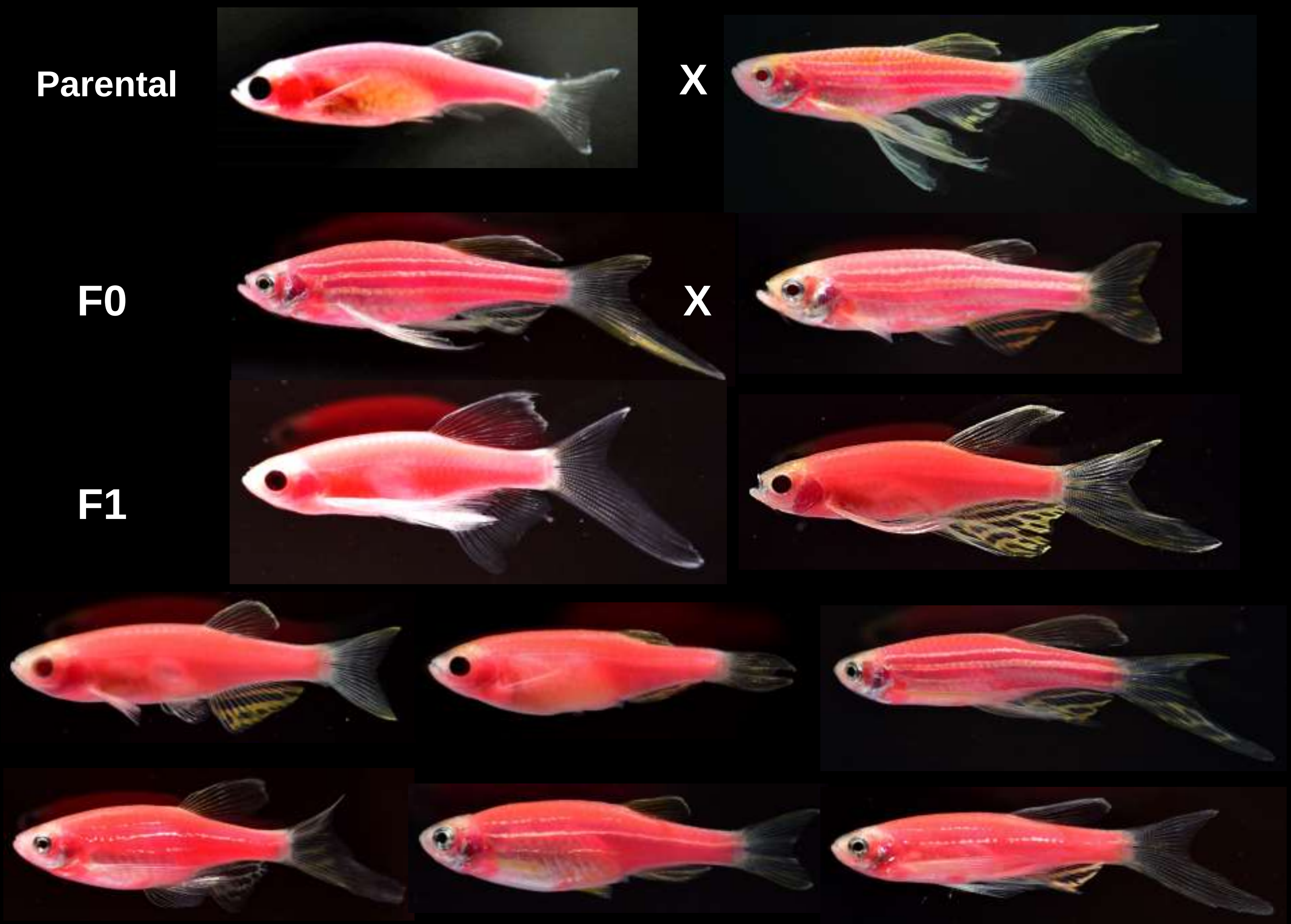


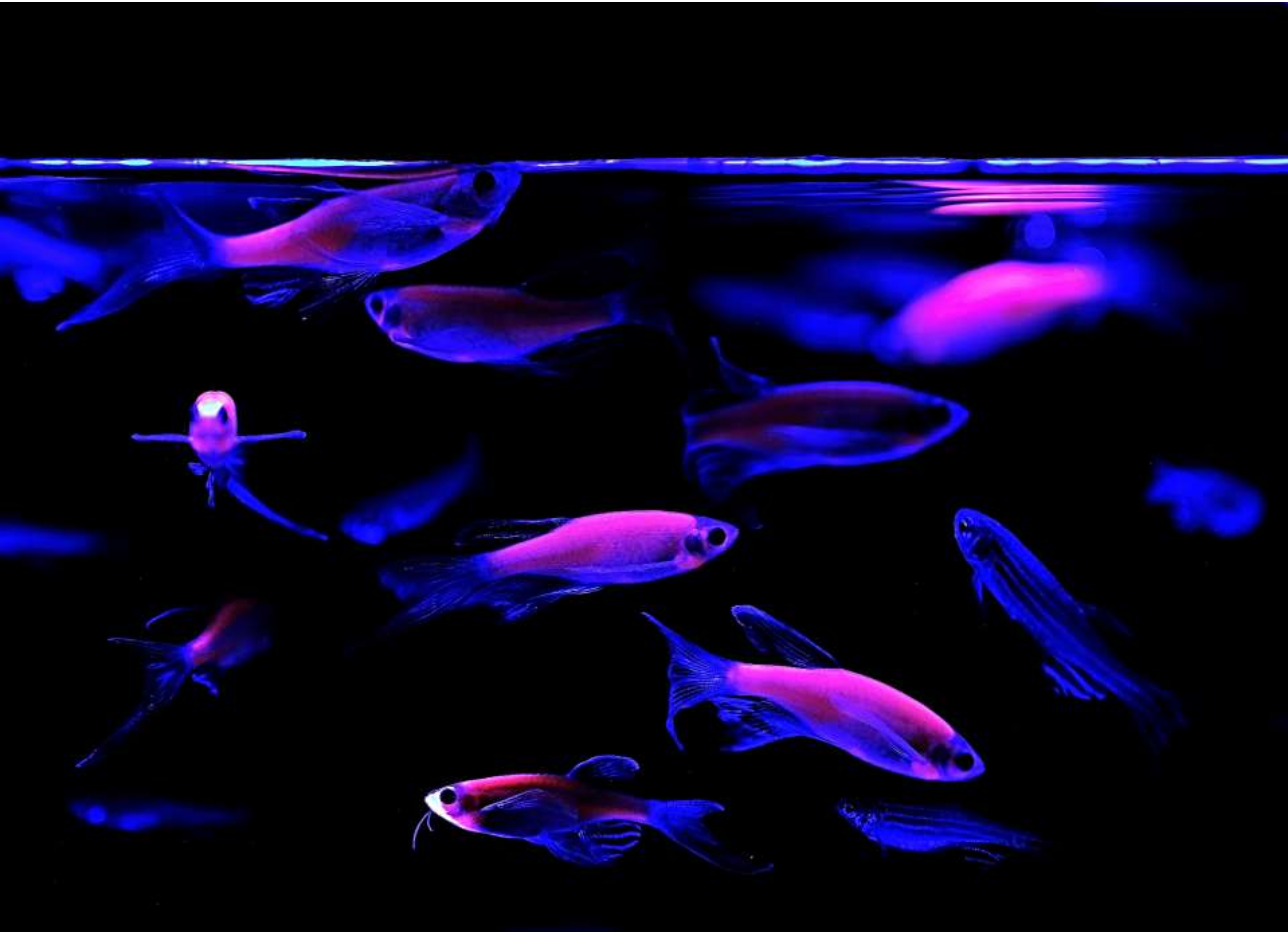




Transgenic Citrine with fluorescent heart

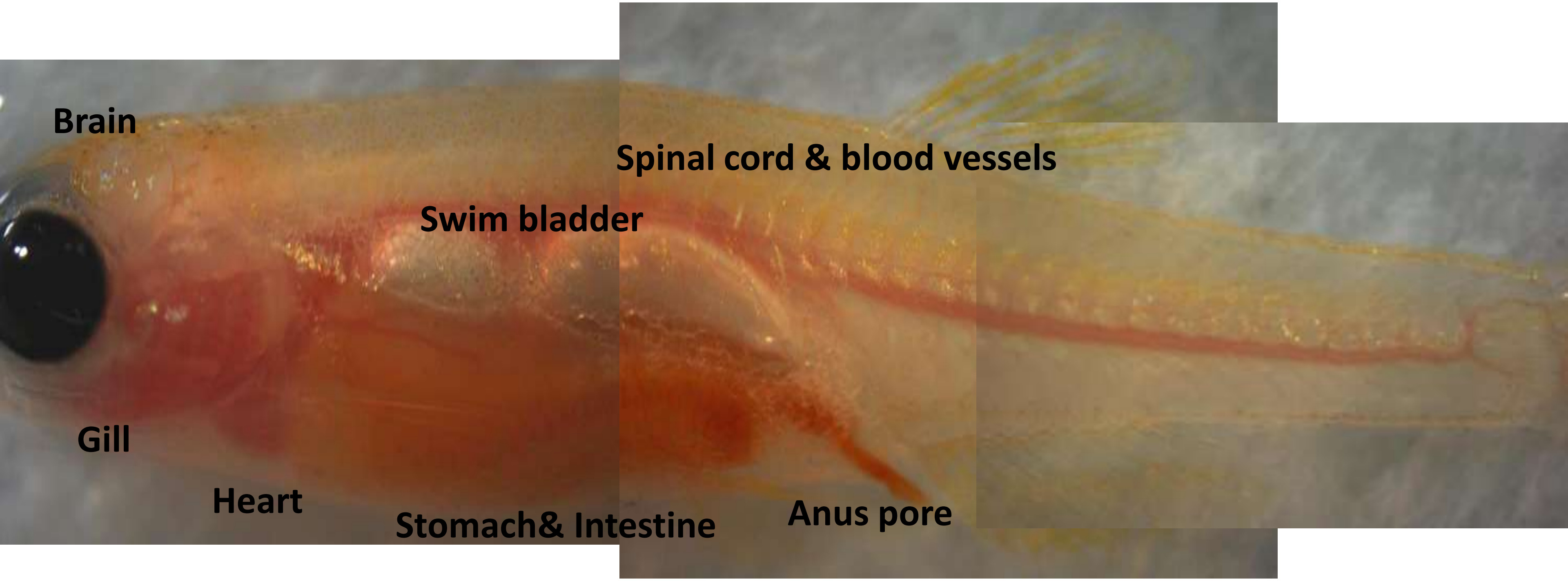

Transparent zebrafish line – Rose Quartz
YJ Chuang Lab, Department of Medical Science, National Tsing Hua University
Photo credit: Jennifer Hsia


Parental
X
F0
X
F1



Melanoma Zebrafish

Transparent zebrafish- Citrine



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