# Lecture 25:

## DNA Mutation

**Course 281**

# Lessons for life

**Jim Rohn Official**
@OfficialJimRohn

"If you don't like how things are, change it! You're not a tree." -- Jim Rohn

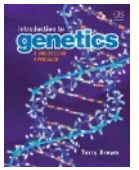
# AIMS

- Understand basic principle of genetics.
- Understand the progression in our understanding of the mutation concept.
- Understand the cell type and location where mutations have hereditary consequences.
- Understand the location of mutations within the genome.
- Understand the two major types of DNA mutation (general view)

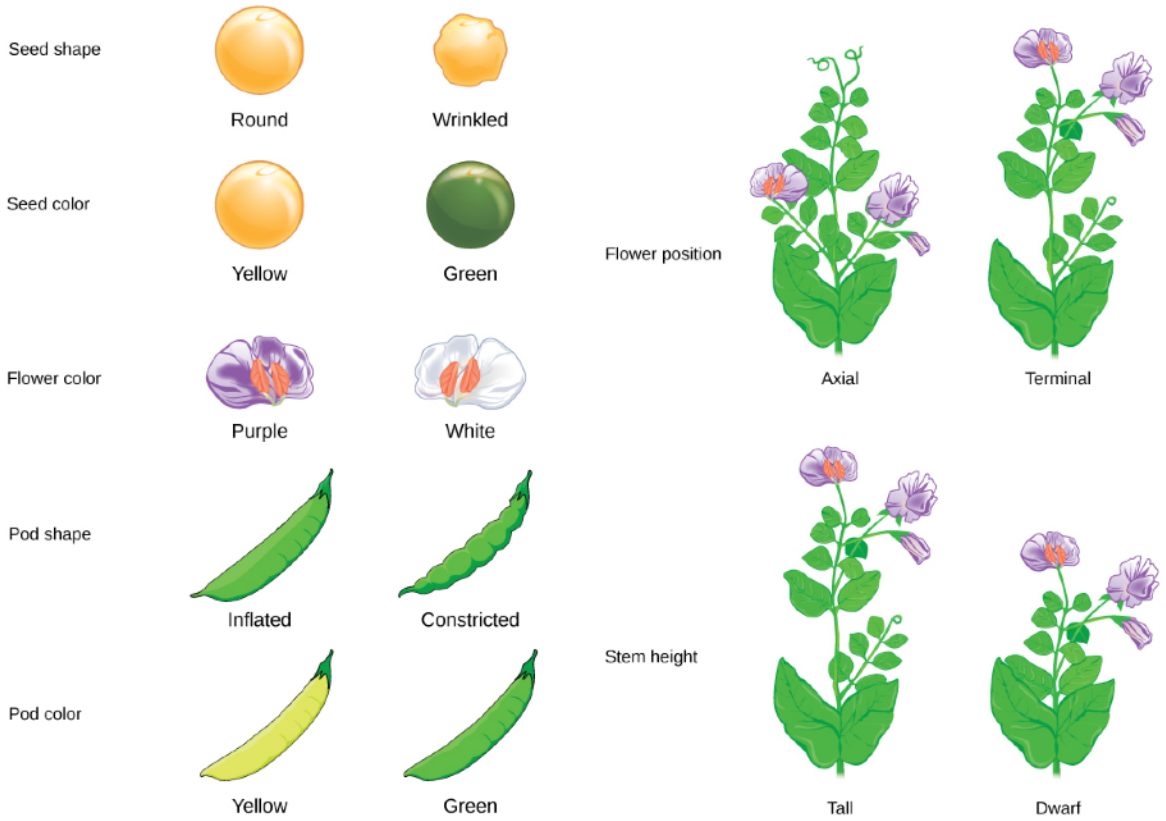
# Genetic Questions

- What is the link between inheritance and evolution?
- How the passage of genes from generation to generation result in genetic diversity and eventually evolution of new species?





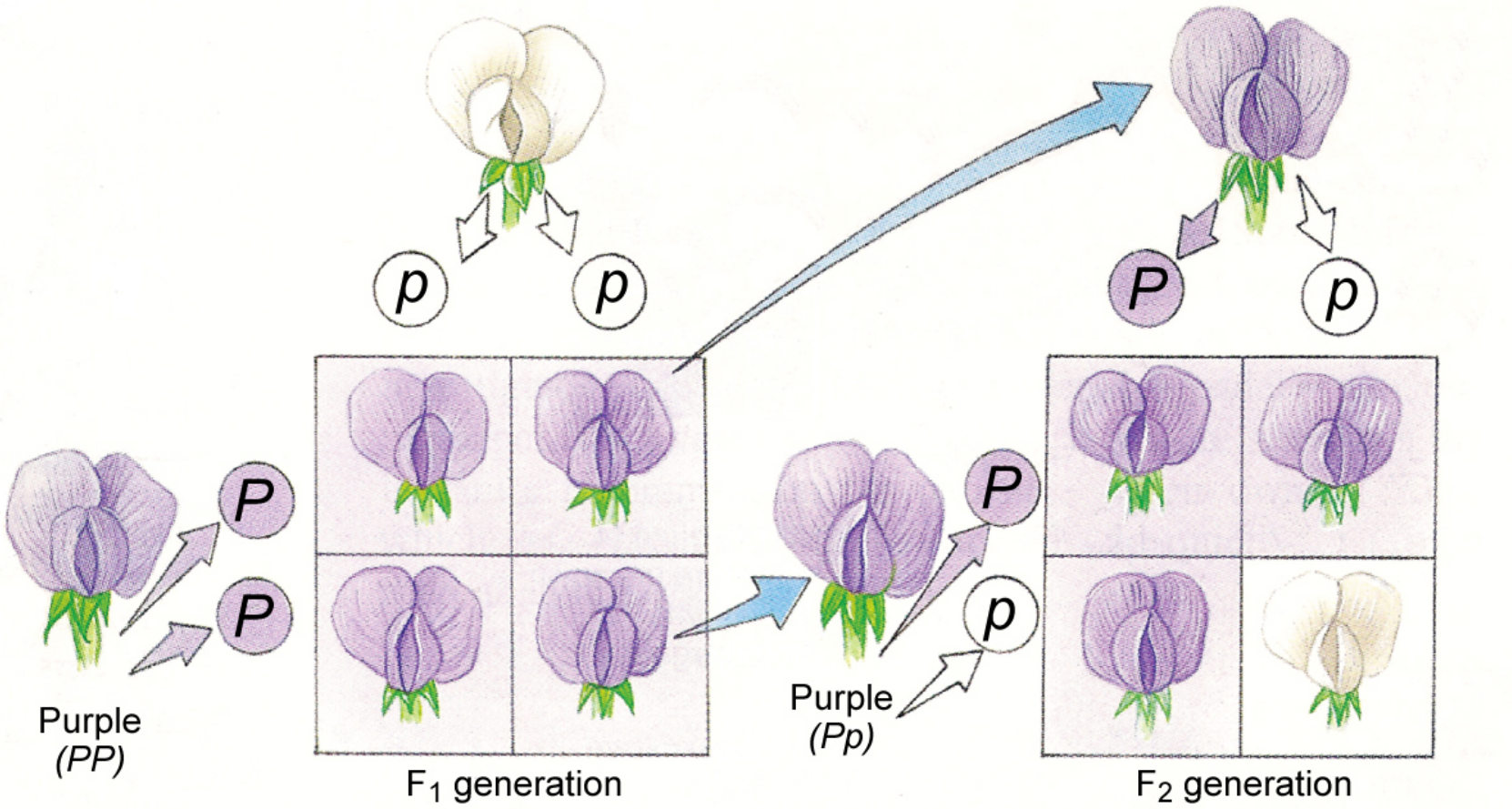
# Father of genetics




Gregor Mendel studies the inheritance of seven phenotypic characters by conducting several matings (crosses)

# Mendel's Particle Inheritance

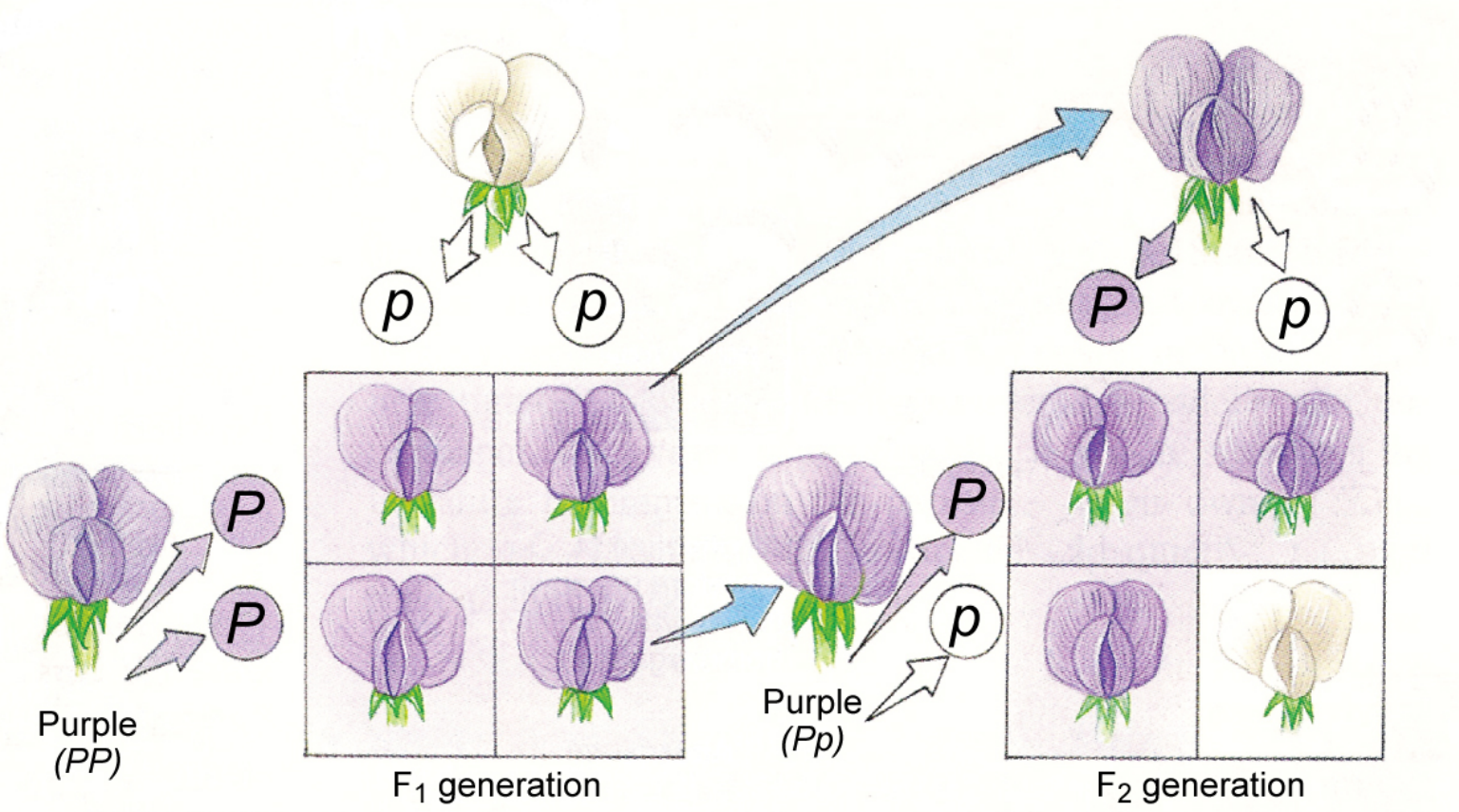
A particle that determines how an organism looks is passed from from parents to offspring and the ratios of phenotypes can be predicted



# Mendel's Particle Inheritance

## Mendel's first law: Segregation of alleles

The units (particles) that makes a flower purple will segregate (separate) in formation of gametes (pollen and eggs)



# Take home message

**The phenotype of an organism is controlled by particles that get passed by parents to offspring**

This may seem very simple for us now when we know all about DNA and genetics but at the time (mid 1800) nothing was yet known

# Fly genetics

Fly room in Morgan's lab were studying genetics on the fruit (vinegar) fly *Drosophila melanogaster.*

Faced difficulties because their flies were all looking the same.

# Fly genetics

Normal flies have red eyes.

One day they found one fly with white eyes in the bottles where they are growing the flies.

# Phenotypic mutants

- White-eyed fly appeared **abnormal** compared with the normal red-eyed flies.
- Thus white-eyed fly is considered a **mutant.**

# Phenotypic mutants

**What is a mutant?**

An organism showing a phenotype that differs from the wild-type.

# Phenotypic mutants

**So can you define the process that makes mutants?**

# Mutation

- **Mutation (old definition):** is the process that generates a phenotype differs from the normal one.

**What is the definition after we know a lot about inheritance, genetics, and DNA?**

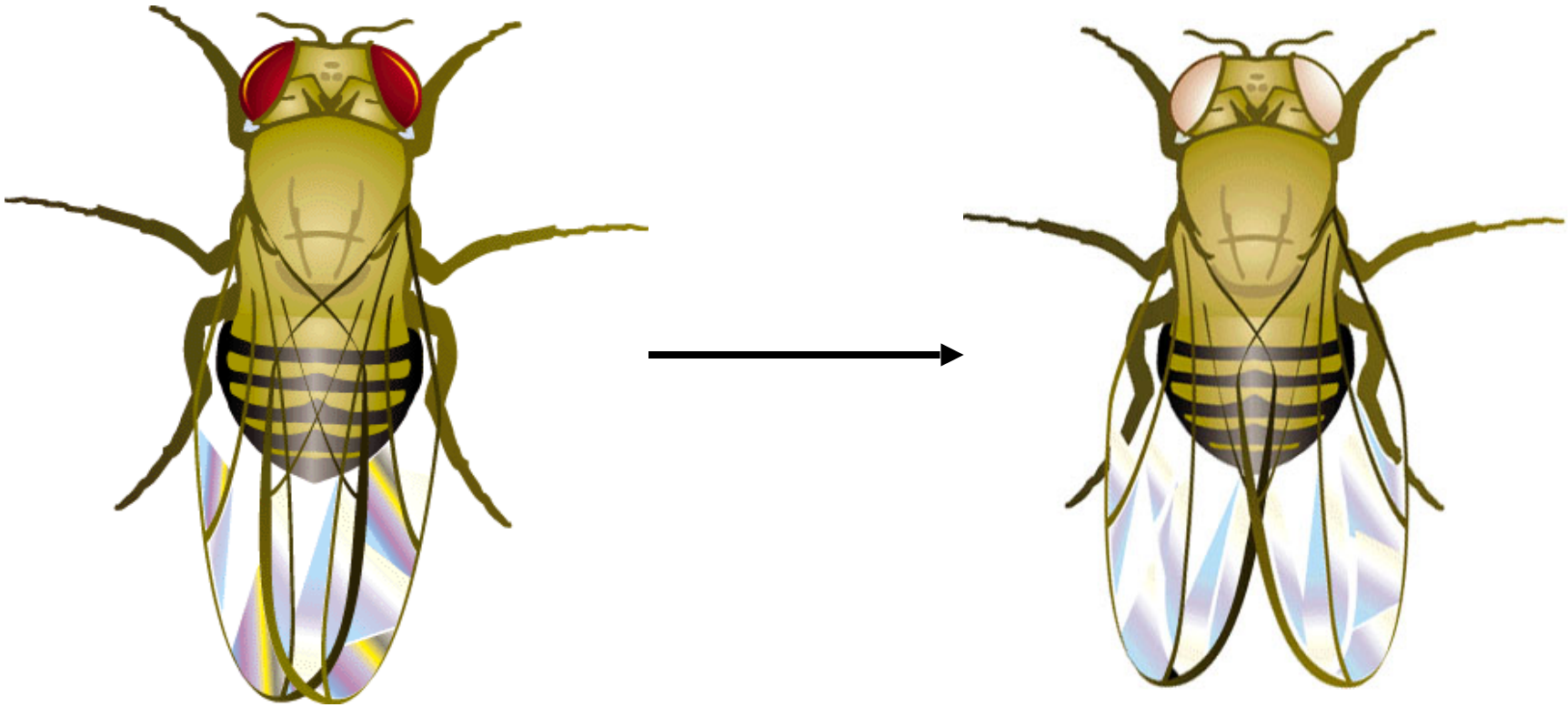
- **Mutation (new) definition:** is the process that results in changes in the sequence of the DNA.

# Mutation

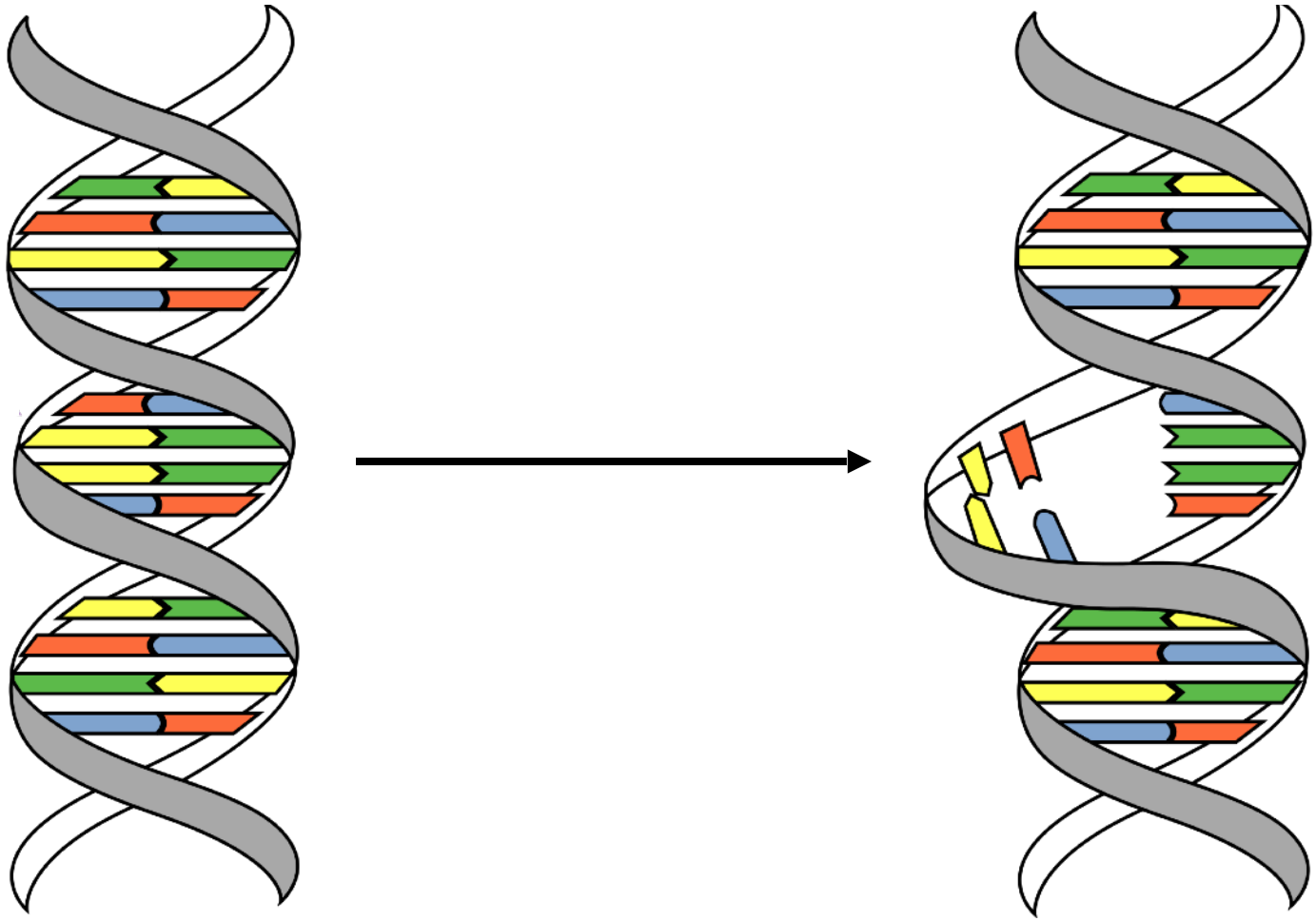
- **The changes of DNA can be:**

    1. Changes to the bases of the DNA
    2. Changes to the chromosome

# Mutation



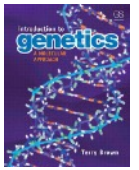
# Mutation



# Introduction

- **Mutations** refer to changes in DNA sequence
- Causes of Mutations;
  - Errors in DNA replication
  - Chemical or physical **mutagen**s, e.g.;
    - Heat
    - Ultraviolet radiation
- **DNA repair** mechanisms correct most of the mutations
- Only mutations that escape repair during meiosis can become permanent (inherited)





# An Error in Replication

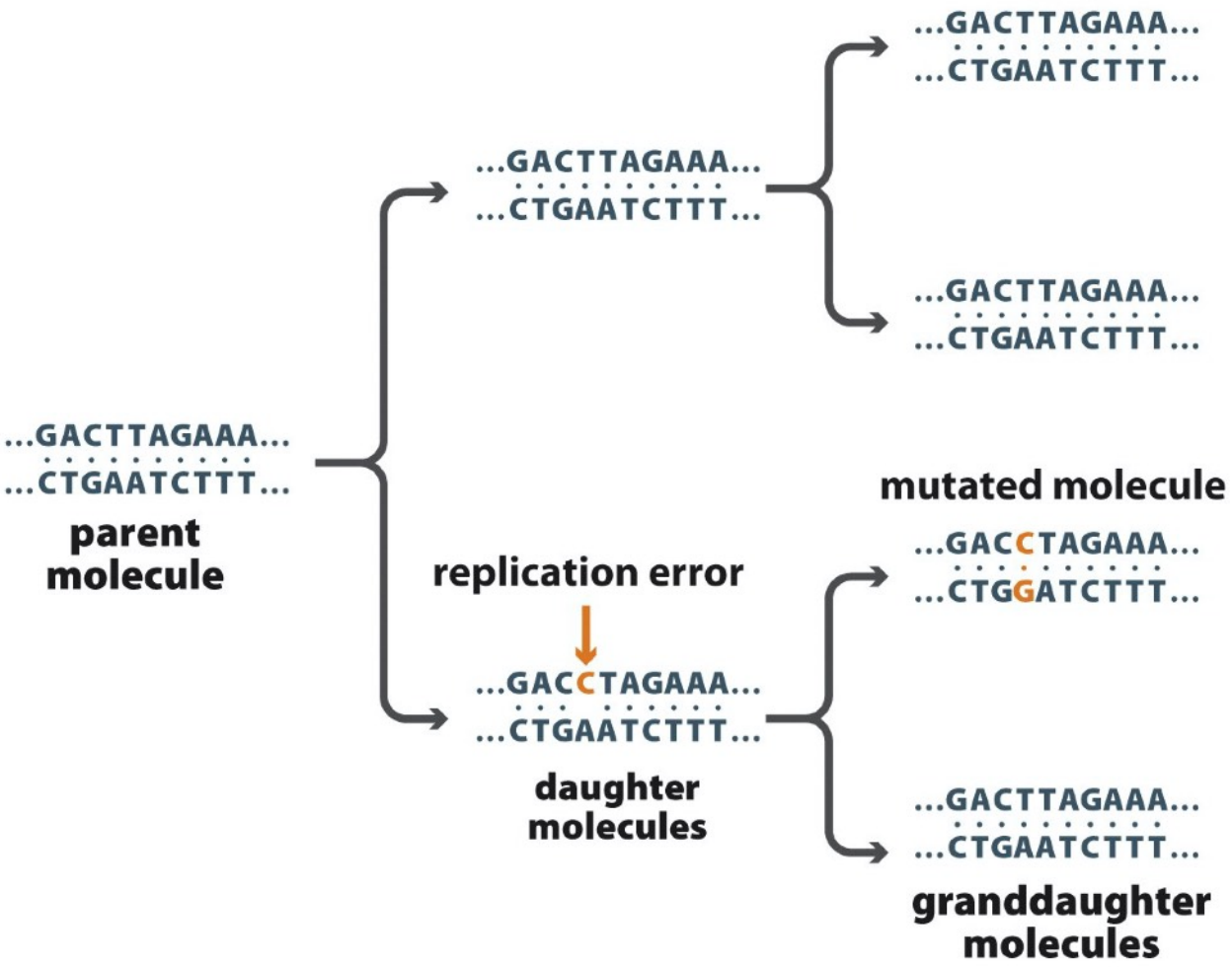
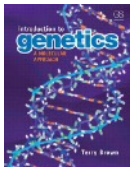



Figure 16.1a Introduction to Genetics (© Garland Science 2012)





# One Possible Effect of a Mutagen

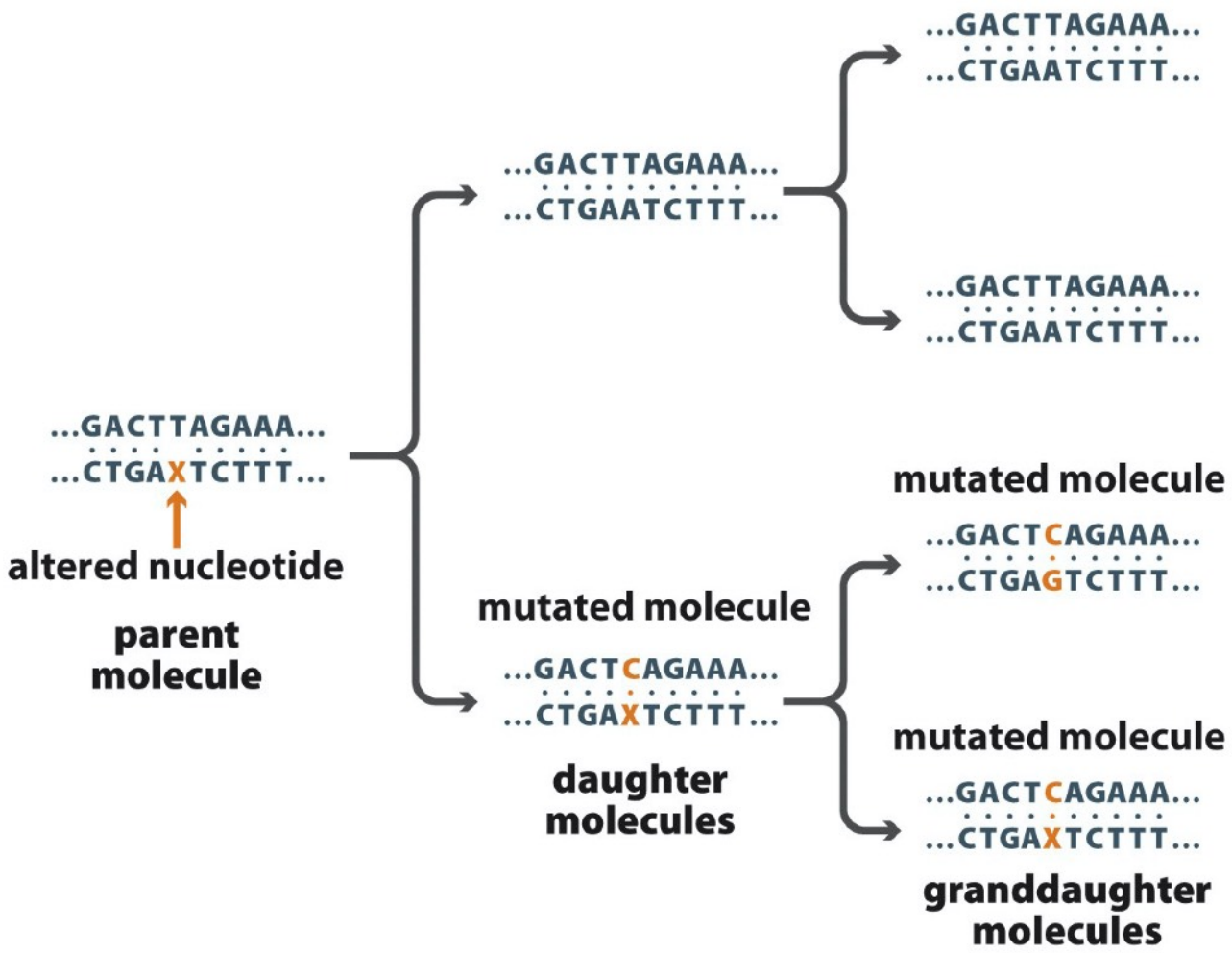
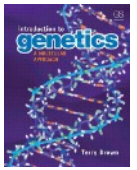



Figure 16.1b Introduction to Genetics (© Garland Science 2012)





# Mutations that Escape Repair in Gametes are Inherited

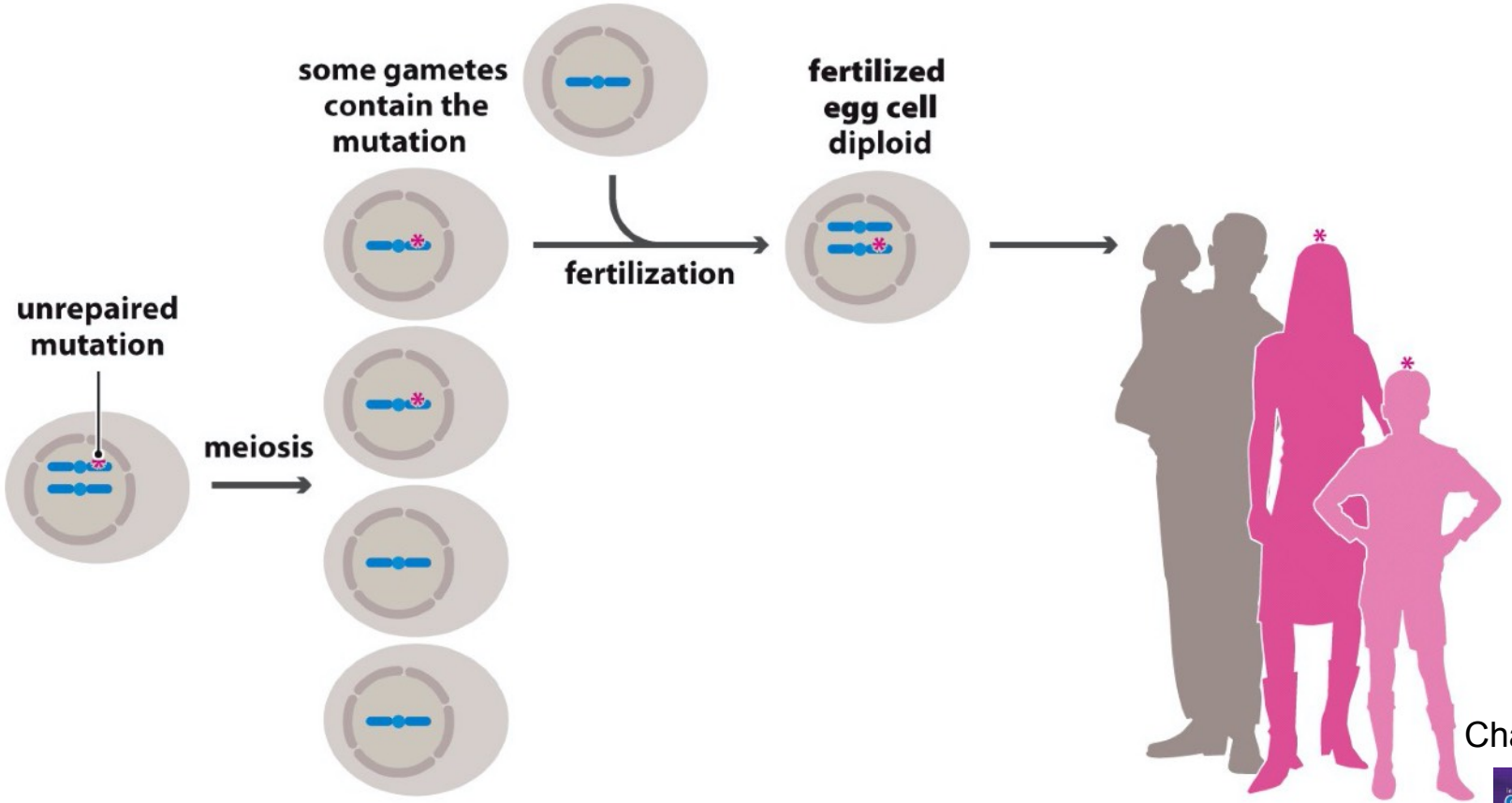
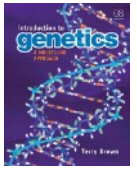



Figure 16.2 Introduction to Genetics (© Garland Science 2012)

Chapter 16



# Mutation

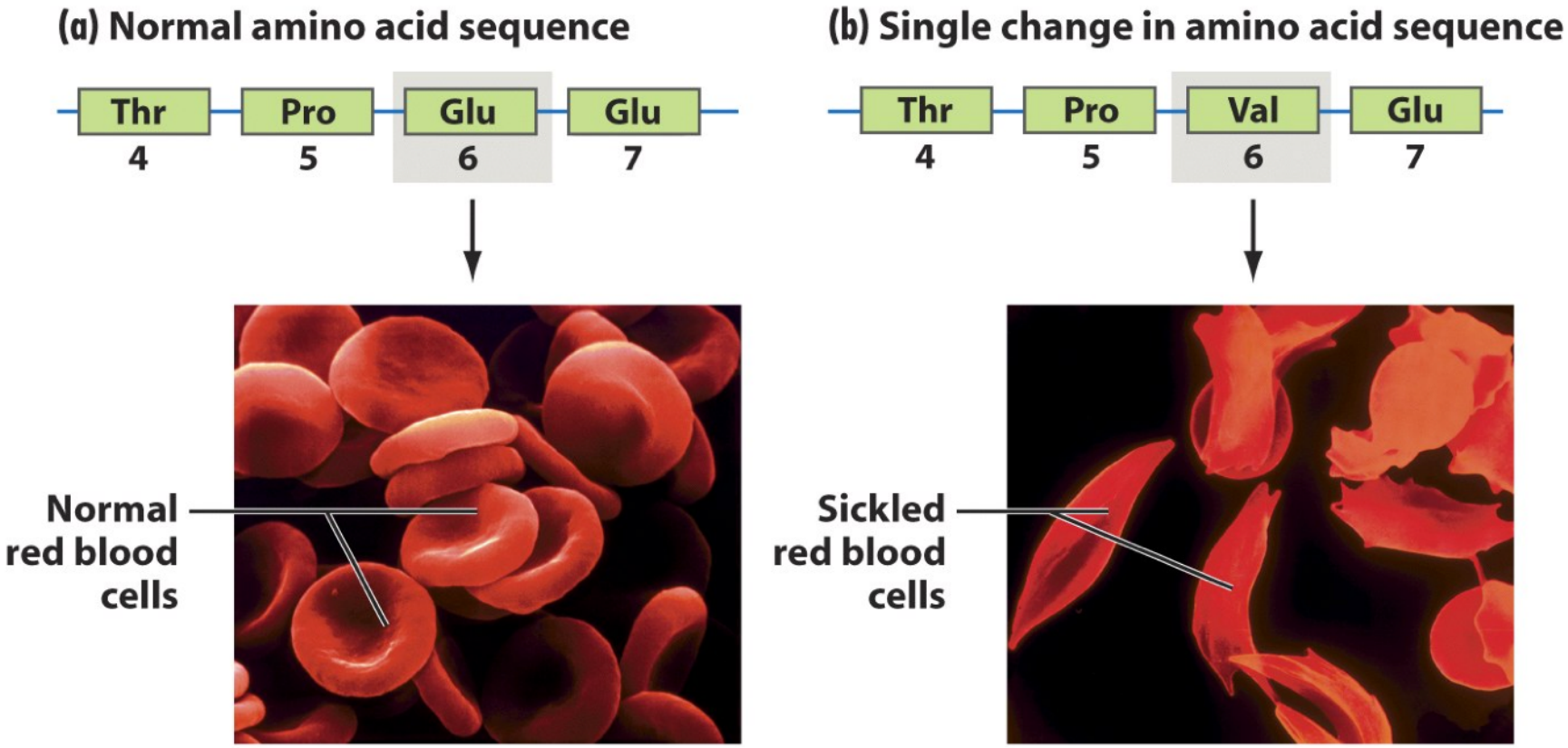
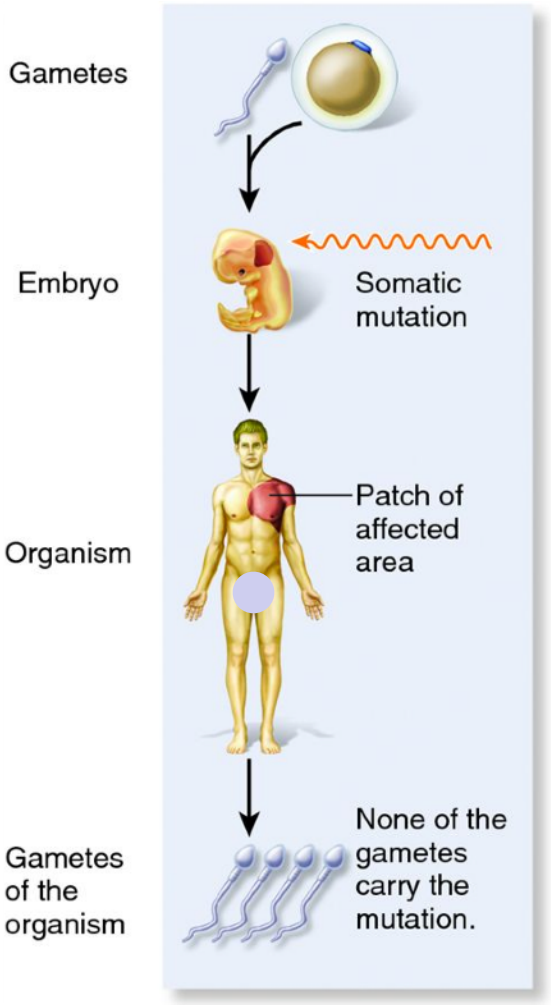



Figure 3-13 Biological Science, 2/e

# Mutation location

1. **Mutations in somatic cells:**
   - Occur in cells other than gametes.
   - Can not be inherited (passed on to future generations).
   - Example: tumor cells mutations.

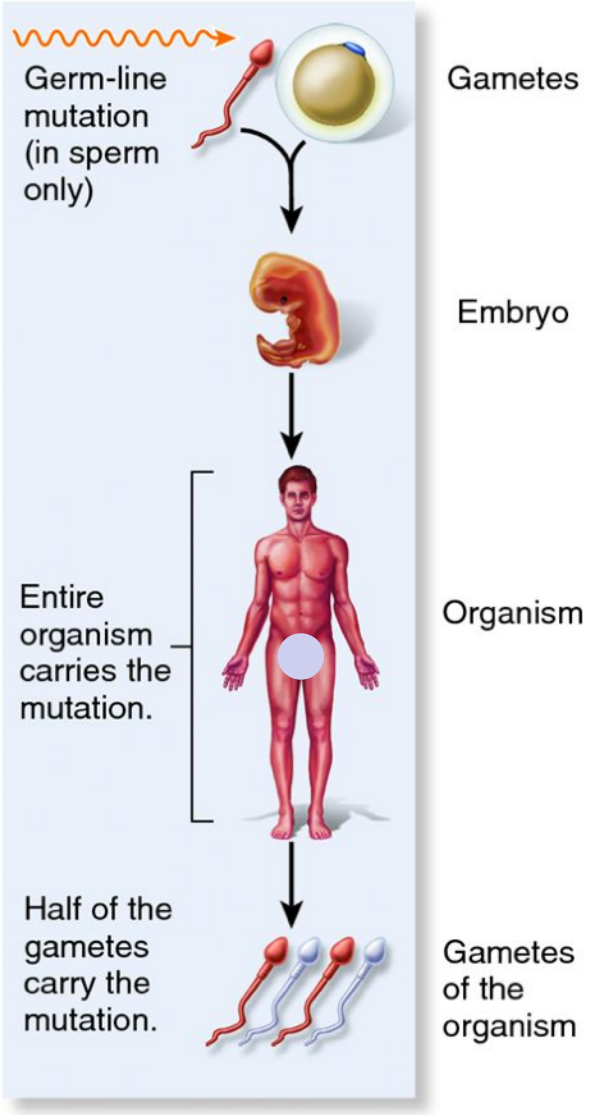



(b) Somatic cell mutation

# Mutation location

2. **Mutation in germ line cells (gametes):**

   - Happens in eggs and sperms.
   - Can be inherited.
   - Example: mutations cause disease without family history.




(a) Germ-line mutation

# Somatic cell mutation

The two eye color cat (or human) is an example of a somatic cell mutation

**What would happen if it was not a somatic cell mutation?**

# Somatic cell mutation

**What would happen if it was not a somatic cell mutation?**

# Germ line mutation

- Normal parents
- Normal siblings
- Only one unfortunate germ mutation.

REAL LIFE

Real-life Thumbelina

A LITTLE MIRACLE

This brave girl has defied her doctors to live a normal life

**Grasping a tiny pencil in her hand, brave Kenadie Jourdin-Bromley proudly draws her name on a sheet of paper. The 10-year-old has a rare condition called primordial dwarfism and stands at just 85cm, the size of an average toddler.**

Despite her brain developing much slower than other children her age, Kenadie has defied doctor's expectations by learning to read and write.

'Kenadie's been reading basic words since last May, and you bet she writes her name on everything!' says mum Brianne Jourdin.

And like most girls her age, Kenadie has a really big obsession.

'She's a huge Justin Bieber fan!' Brianne laughs. 'Every night before bed, in her high-pitched voice, Kenadie can be heard singing: "Baby, baby, baby..."'

Kenadie was born weighing just 1.3kg. Doctors said she was likely to have brain damage and wasn't expected to live for long.

'We baptised her the day she was born. We even made funeral arrangements,' Brianne admits.

'Doctors said we'd be lucky to even see her smile.'

But Kenadie went from strength to strength, says Brianne. 'She was sitting up by 11 months, babbling by one and starting to walk by two.'

Even when Kenadie was eventually diagnosed with primordial dwarfism at 18 months, she showed no signs of becoming ill. 'Her brain, heart and organs are all perfectly healthy,' her mother explains.

'They were saying she might even live to her 20s, or maybe even 30s. But they've kind of stopped predicting how long she might live.'

While Kenadie develops at a slower rate than those around her, she is in the fourth grade at school with others her age and has a special tutor with her at all times.

She's determined to be independent, and despite struggling to even open the fridge door, she makes her breakfast every day at home. Her mum says she's an avid photographer too.

'I bought her this tiny pink digital camera... and some of the pictures she takes are amazing! The world she captures from her perspective is so different, the way people look at her, the angle she views the big world from.'

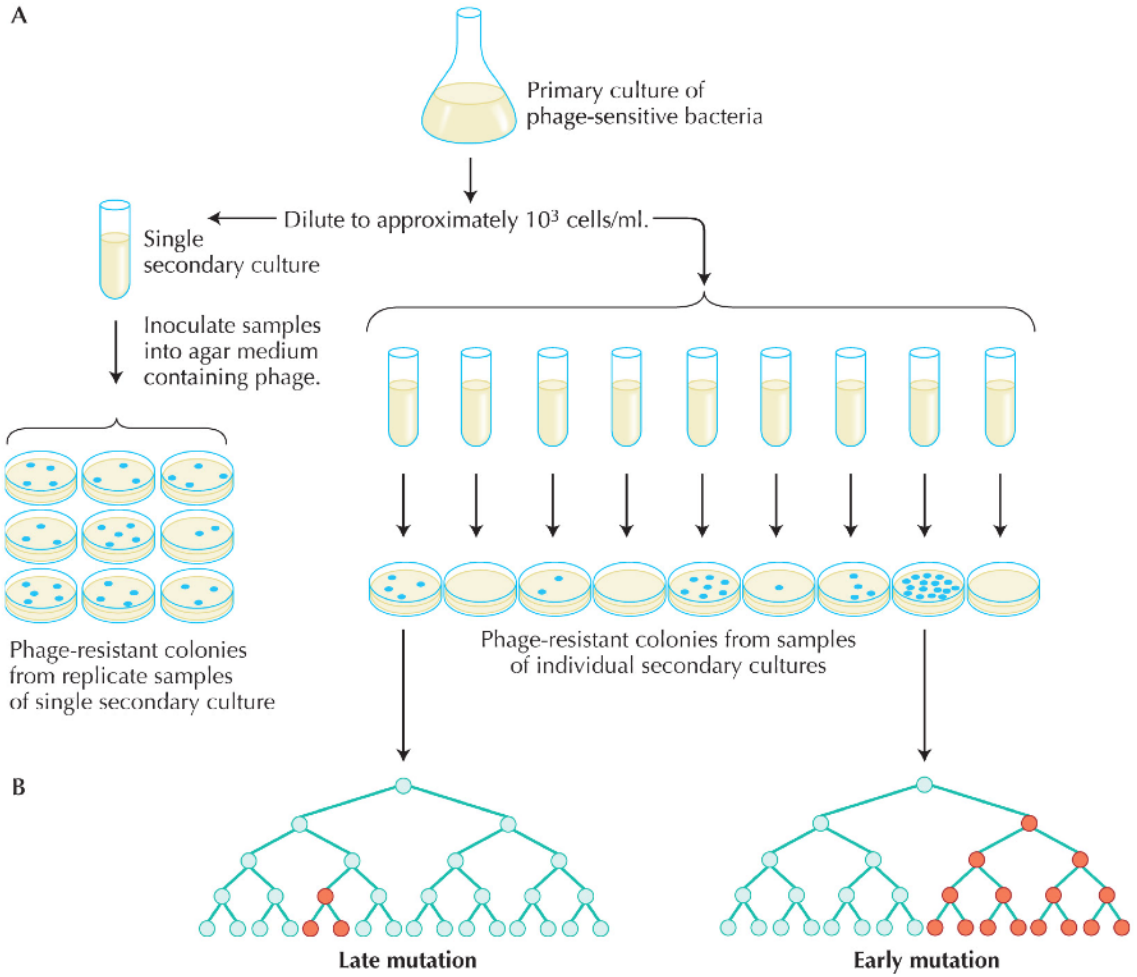
For now, Brianne, from Kitchener, Canada, says: 'I've learnt that every kid is different and every day is a new day. We didn't ask for this but Kenadie has been a blessing.' NI

Like millions of other girls her age, Kenadie has been swept up in Justin Bieber fever! Left: Kenadie, 10, stands with her younger brother Tyran, who is 45cm taller than her.

42 New Idea

# Spontaneous mutations

## Delbruck and Luria experiment of spontaneous mutations



# Spontaneous mutations

**Can mutations occurring randomly be quantified?**

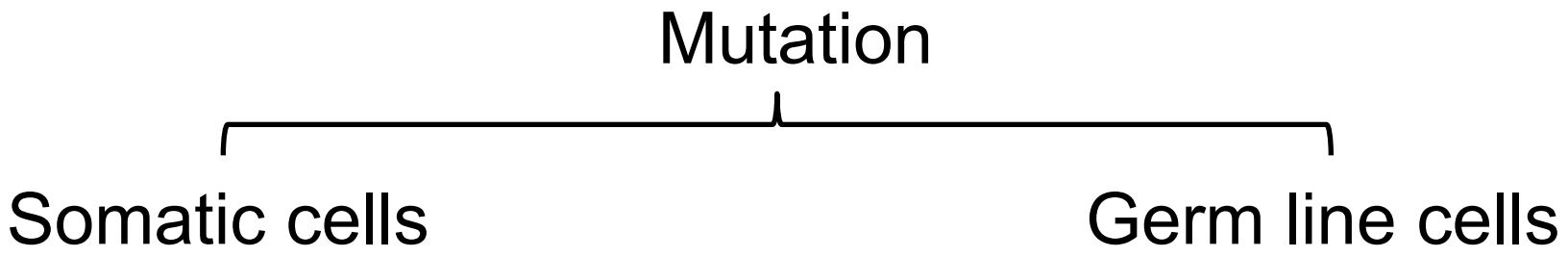
# Spontaneous mutations

• The occurrence of random mutations can be quantified by measuring its probability of happening over a time period. This is called **mutation rate.**

• **Mutation frequency** is a measure of how many individuals carry the mutation. This can give us an idea of when the mutation took place.

# Mutations location

**Where mutations take place in an organism?**

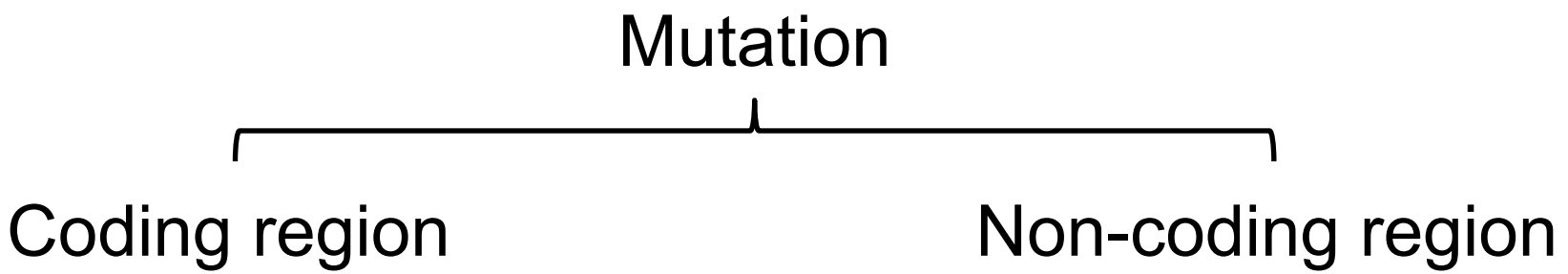



**Do bacteria have germ line cells?**

**Which organisms have germ line cells?**

# Mutations location

**Where mutations take place in a genome?**




**What is a coding region?**

**What is a non-coding region?**

**Any examples?**

# To know

Coding region

Somatic cell mutation

Chromosomal mutation

Mutation frequency

Non-coding region

Germ line mutation

DNA mutation

Mutation rate

# Expectations

- You know the basic inheritance principles and their relation to mutations.
- You know that the study of mutations moved from looking at a phenotype to understanding the DNA changes associated.
- You know where mutations (what cells) mutations have genetic consequences.
- You know that somatic cell mutations have health consequences.

# For a smile


"AS FAR AS I CAN TELL, YOUR DNA CHAIN
WAS MISSING A FEW MINOR LINKS."
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