Unit 10 · Mutation and Polyploidy

STUDY NOTES

WHAT NEB'S GRADE 12 EXAM ASKS

- **CDC's grid (2078):** Genetics (Units 7–10 together): 12 of 75 marks; 1 short question (4 marks: understanding 1, application 2, higher ability 1) and 1 long question (8 marks: understanding 2, application 3, higher ability 3)
- **The paper:** Botany 37 + Zoology 38; each part: MCQs × 1 (5 Botany, 6 Zoology) · 4 short × 4 · 2 long × 8; one OR in each short and long group; 3 hours; 1 mark per expected point
- **CDC's model paper:** MCQ: euploidy (whole sets of chromosomes gained or lost)
- **MCQ (1 mark)**
  - sickle-cell anemia: a transversion
  - purine → purine: transition
  - one set: monoploidy; Turner 44 + XO
  - autotetraploid of AA: AAAA
  - which change of a base sequence is a frameshift
- **Short and long (in parts)**
  - mutation; gene vs chromosomal mutation
  - 2n = 14: tetraploid 28, hexaploid 42
  - positive and negative importance of mutation
  - polyploidy: types, significance
  - 45 chromosomes: which mutation, possible outcomes

Key terms

Mutation (de Vries, 1901): sudden, **heritable** change in the genetic material: base sequence, or chromosome structure or number.

Mutant: organism showing a mutation's effect.

Mutagen: outside agent causing mutations: X-rays, gamma rays, UV, chemicals, some viruses.

Gene (point) mutation: change in the base sequence of one gene.

Transition: purine ↔ purine (A ↔ G) or pyrimidine ↔ pyrimidine (C ↔ T).

Transversion: purine ↔ pyrimidine.

Frameshift: insertion or deletion of bases not a multiple of 3; shifts the **reading frame**.

Nonsense mutation: a sense codon becomes a **stop** codon.

Chromosomal (macro) mutation: change in chromosome structure or number.

Non-disjunction: chromosomes or chromatids fail to separate.

Ploidy: number of chromosome sets.

Aneuploidy: single chromosomes lost or gained; **euploidy**: whole sets.

Polyploidy: more than two sets.

Autopolyploidy: extra sets of one species; **allopolyploidy**: a two-species hybrid's sets doubled.

Colchicine: from autumn crocus; stops the spindle → doubles chromosomes.

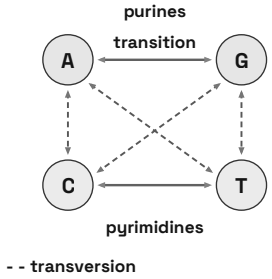
Mutation

- **de Vries** (1886): evening primrose *Oenothera lamarckiana*; *The Mutation Theory* (1901)
- **Morgan**: mutation = change in a gene on a chromosome; **Muller** (1927): X-rays ~150× more mutations
- **spontaneous** (copying mistakes) or **induced** (mutagens)
- body cell: only its descendants (cancer); **germ cell**: inherited
- mostly neutral; some harmful; a few useful
- only source of **new alleles**: raw material of evolution
- ~50–70 new mutations per newborn; ~1 copying mistake per billion

	Gene mutation	Chromosomal mutation
scale	one gene, often 1 bp	long stretch, many genes
change	base sequence	structure or number
cause	replication error missed	breaks; non-disjunction
seen under microscope	no	often (karyotype)
example	sickle-cell anemia	Down syndrome

Gene mutations

a) Substitution: one base replaced



Purines and pyrimidines.

- **transition**: A ↔ G, C ↔ T; ~2× as common as transversions (though half as many possible)
- **transversion**: A or G ↔ C or T
- effects: **silent** (CAT, CAC both His), **missense** (different amino acid), **nonsense** (stop)
- **sickle-cell anemia**: β-globin codon 6, GAG (Glu) → **GTG** (Val): A → T, a **transversion**, missense; hemoglobin fibers → sickle cells

b) Frameshift: insertion or deletion of 1, 2, 4… bases

	DNA	Amino acids
original	ATG CAT GTA TGA	Met–His–Val–Stop
A inserted	ATG ACA TGT ATG A…	Met–Thr–Cys–Met…
C deleted	ATG ATG TAT GA…	Met–Met–Tyr…

- every codon after it changed; stop lost or early → useless protein
- **3 bases** (multiple of 3): **in frame**; whole amino acid lost or added (cystic fibrosis, Phe 508)
- Huntington's: CAG repeat expansion; Tay–Sachs: 4 extra bases, a true frameshift

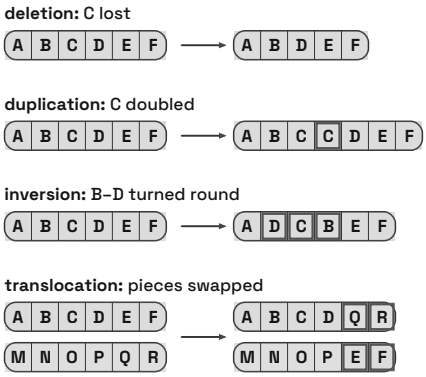
c) Nonsense: sense codon → **UAA, UAG, UGA** → **premature termination**

- template GCT → ACT: mRNA CGA (Arg) → **UGA** (stop); Met–Phe–Stop
- some Duchenne muscular dystrophy (dystrophin cut short)
- one base deleted near the start does the most harm

Chromosomal mutations

Structural (breaks wrongly rejoined)

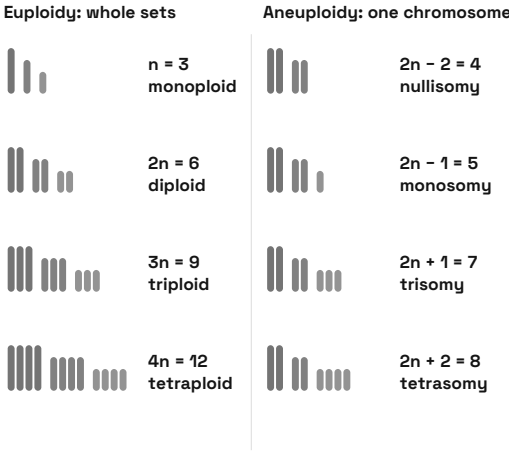
- **deletion**: piece lost; **cri-du-chat** (short arm of 5)
- **duplication**: piece twice; imbalance, or raw material (red, green pigment genes)
- **inversion**: piece turned round; no genes lost; loop in meiosis → **reduced fertility**
- **translocation**: piece to a **non-homologous** chromosome; **reciprocal** or **non-reciprocal**; balanced carriers normal; 14/21 Down; **Philadelphia chromosome** (9, 22) → leukemia
- deletion, duplication, inversion intrachromosomal; translocation interchromosomal



Structural changes in chromosomes.

Genomic (number; mostly **non-disjunction**)

- n + 1 or n − 1 gametes → 2n + 1 or 2n − 1 zygotes; whole division fails → 2n gamete → polyploidy



Aneuploidy and euploidy.

Aneuploidy: single chromosomes; genes out of balance

Kind	Number	Example
nullisomy	2n − 2	lethal in animals; bread wheat survives
monosomy	2n − 1	**Turner, 44 + XO** (45,X)
trisomy	2n + 1	**Down** (trisomy 21), Edwards (18), Patau (13), **Klinefelter 44 + XXY**
tetrasomy	2n + 2	tetrasomy X, 48,XXXX
pentasomy	2n + 3	49,XXXXX
hexasomy	2n + 4	hardly known

- hypoploidy: nullisomy, monosomy; hyperploidy: trisomy and up

- Down: 45 + XX or 45 + XY (47); 45 chromosomes → **monosomy**, e.g. Turner (short, no puberty untreated); autosomal monosomy lethal

Euploidy: whole sets; genes stay balanced; common in plants

- **monoploidy** (n): small, weak, sterile; pollen-grown, doubled → pure lines; drones of bees haploid by nature
- **polyploidy**: 3n, 4n, 5n, 6n…; 2n = 14 → 4n = 28, 6n = 42

Polyploidy

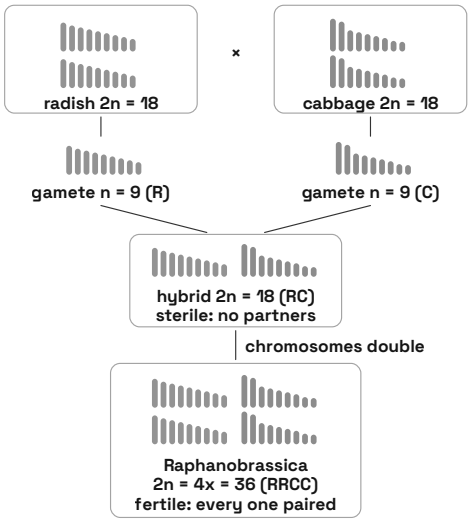
- causes: 2n gametes, two sperm in one egg, doubling without division; **colchicine**
- very common in plants; rare, usually harmful in animals (triploid farm fish, oysters)

Autopolyploidy (AA → AAA, AAAA)

- **banana** (Cavendish AAA): no seeds, grown from suckers; plantains AAB, ABB
- **seedless watermelon**: 4n × 2n → **3n**
- **potato**: autotetraploid, 2n = 4x = 48
- some apples, big-berried grapes

Allopolyploidy (hybrid AB → AABB)

- hybrid AB sterile (no partners in meiosis); doubled → fertile, a **new species**
- *Raphanobrassica* (**Karpechenko**, 1927–28): radish RR (18) × cabbage CC (18) → RC (18, sterile) → **RRCC (36)**, fertile; radish leaves, cabbage root
- **bread wheat** AABBDD, 2n = 6x = 42
- **rayo** AABB (36) = tori AA (20) × black mustard BB (16): gametes 10 + 8 = 18 → 36
- cotton; **triticale** (wheat × rye)



Raphanobrassica.

Significance

- new species in one generation
- larger cells, organs (**gigas** effect)
- **seedless fruits** (triploids)
- vigor, yield, disease resistance
- new crops (triticale); sterile hybrids made fertile
- study of gene dose

Importance of mutation

Positive

- **variation** for evolution; only source of new alleles
- useful characters, adaptation (disease, poison resistance)
- food: radiation-bred rice, barley, wheat
- understanding genes and disease
- disease-resistant, high-yielding varieties

Negative

- genetic disorders (Down, sickle-cell)
- **cancer** (body cells)
- reduced fertility, birth defects
- weak or non-viable organisms
- **drug resistance** in germs (antibiotics)

COMMON MISTAKES

✗ Sickle-cell is a transition
✓ **transversion** (A → T), missense.

✗ A transition swaps a purine for a pyrimidine
✓ that is a **transversion**.

✗ Every insertion or deletion is a frameshift
✓ not when it is a **multiple of 3**.

✗ Turner syndrome is 45 + XO
✓ **44 + XO** (45 chromosomes).

✗ Mutations in skin cells are inherited
✓ only **germ-cell** mutations.

✗ Euploidy is single chromosomes gained or lost
✓ that is **aneuploidy**; euploidy = whole sets.

✗ Banana is allopolyploid
✓ Cavendish is **autotriploid** AAA.

✗ Allopolyploids are sterile
✓ the doubled hybrid is **fertile**; the undoubled hybrid is sterile.