Unit 3 · Genetics

STUDY NOTES

Key terms

Dihybrid cross: pure parents differing in two pairs of characters.

Recombinants: new combinations, in neither grandparent.

Test cross: with a parent recessive for the genes studied.

Linked genes: close on the same chromosome; go into gametes together.

Nucleotide: phosphate + pentose sugar + nitrogen base.

Nucleoside: sugar + base.

Purines: two rings; A, G. **Pyrimidines**: one ring; C, T, U.

Complementary pairing: A–T, G–C; one strand fixes the other.

Antiparallel: strands run in opposite directions.

Codon: three mRNA bases for one amino acid. **Anticodon**: three tRNA bases that pair with it.

Replication: DNA copying. **Semi-conservative**: each new molecule keeps one old strand.

Transcription: DNA → mRNA. **Translation**: mRNA → protein at a ribosome.

Carrier: one recessive allele, no sign, can pass it on.

Hemizygous: only one copy of a gene (a man's X-linked genes).

Criss-cross inheritance: affected man → carrier daughter → her son.

Holandric: Y-linked; father to every son.

Pseudoautosomal regions: short stretches shared by X and Y tips.

Numbers and rules

- 9 : 3 : 3 : 1 = (3 : 1) × (3 : 1)
- 2 × 2 = 4 gametes; 4 × 4 = 16 boxes; 3 × 3 = 9 genotypes; 4 phenotypes
- **product rule**: work each gene alone, multiply; e.g. RrYy × Rryy: 3 : 3 : 1 : 1
- wrinkled yellow = 3/16; of them ⅓ breed true; pure for both = 4/16
- length = base pairs × 0.34 nm; turns = base pairs ÷ 10
- H bonds = 2 × (A–T pairs) + 3 × (G–C pairs)
- A = T, G = C; A + G = T + C = 50%
- nucleotides = (amino acids + 1 stop) × 3
- X with the allele share p:
  - affected men p
  - carriers $2p(1-p)$
  - affected women $p \times p$
- $4^3 = 64$ codons: 61 for 20 amino acids; 3 stop (UAA, UAG, UGA); AUG = methionine + start

Dihybrid cross

- P: RRYY (round yellow) × rryy (wrinkled green); gametes RY, ry
- F_1: all RrYy, round yellow; gametes RY, Ry, rY, ry, a quarter each
- F_2 (Mendel, 556): 315 : 108 : 101 : 32 (expected 312.75 : 104.25 : 104.25 : 34.75)

♂ / ♀	RY	Ry	rY	ry
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

Phenotype	Genotypes (boxes)	Total
round yellow	RRYY 1, RRYy 2, RrYY 2, RrYy 4	9
round green	RRyy 1, Rryy 2	3
wrinkled yellow	rrYY 1, rrYy 2	3
wrinkled green	rryy 1	1

Law of independent assortment: **When two pairs of contrasting characters are followed together, the alleles of one pair separate into the gametes independently of the alleles of the other pair.**

- why: each homologous pair lines up by chance at metaphase I, separately (genes on **different** chromosomes)
- each character alone ≈ 3 : 1 (423 : 133; 416 : 140)

Verification

- **test cross** RrYy × rryy: 1 : 1 : 1 : 1 (31 : 26 : 27 : 26 and 24 : 25 : 22 : 27, both ways round)
- **F_2 grown on**: pure-for-both : hybrid-for-one : hybrid-for-both = 1 : 2 : 4 (33 : 65 : 138)

Structure of DNA and RNA

Base	Kind	Formula	In
adenine	purine	$C_5H_5N_5$	both
guanine	purine	$C_5H_5N_5O$	both
cytosine	pyrimidine	$C_4H_5N_3O$	both
thymine	pyrimidine	$C_5H_6N_2O_2$	DNA
uracil	pyrimidine	$C_4H_4N_2O_2$	RNA

Sugar: **ribose** $C_5H_{10}O_5$ (–OH on 2′); **deoxyribose** $C_5H_{10}O_4$ (H on 2′).

Bonds

1. **N-glycosidic**: base → sugar's 1′ carbon (N9 of purine, N1 of pyrimidine); strong, covalent
2. **Phosphodiester**: 5′ carbon of one → 3′ carbon of next, through a phosphate; the backbone; strong
3. **Hydrogen**: base to base across strands; A–T **two**, G–C **three**; weak, opened in copying

Watson and Crick, 1953

1. two strands; right-handed double helix
2. backbone outside, bases inside
3. antiparallel (5′ → 3′ read)

4. A with T, G with C; purine with pyrimidine, so width is constant
5. about 2 nm wide; 0.34 nm per base pair; about 10 base pairs (3.4 nm) a turn
6. major and minor grooves

Chargaff, 1950: A = T, G = C (human: about 31% A, 29% T, 20% G, 20% C).

G–C-rich DNA separates at a higher temperature.

DNA	RNA
deoxyribose	ribose
A, G, C, T	A, G, C, U
double helix	single strand, folded in places
A = T, G = C	need not be equal
mostly in the nucleus	mostly cytoplasm, ribosomes
stores information	uses it to make proteins

Types of RNA

	mRNA	tRNA	rRNA
share	few percent	about 1⁄6	about 4⁄5
shape	straight strand	clover leaf (L)	folded with proteins
size (nucleotides)	hundreds to thousands	76–90	120–5,000
job	carries the code	brings amino acids	builds ribosome, joins amino acids
special	codons	anticodon, CCA end	–

tRNA arms: acceptor (3′ end CCA, amino acid on the last A), anticodon, D, T.

- anticodon runs opposite: AUG read by 3′-UAC-5′

Role of DNA

1. **Stores information**:
   - base order
   - gene = stretch coding for a protein or RNA
   - about 3.1 billion base pairs, about 20,000 protein genes per set
2. **Replication**: unzip → each strand a template → DNA polymerase; 2 molecules, one old and one new strand (Meselson and Stahl, 1958; two generations: half halfway, half light)
3. **Controls characters**: **DNA → RNA → protein**; wrinkled pea: r has an extra 800 base pairs, no starch-branching enzyme
4. **Variation**: mutation (new alleles), recombination (independent assortment, crossing over)
5. **Passes on**: half from each parent

Evidence for DNA:

- Griffith 1928 (transformation)
- Avery, MacLeod and McCarty 1944 (only DNA transformed)
- Hershey and Chase 1952 (only DNA entered)

Some viruses: RNA.

Sex-linked inheritance

X: about 800 protein genes; Y: a few dozen.

Kind	Where	Example
X-linked	X, no partner on Y	color vision, hemophilia, dystrophin, G6PD
Y-linked (holandric)	Y, no partner on X	SRY
XY-linked	pseudoautosomal	SHOX

- hairy ears only once *thought* Y-linked; give SRY

X-linked recessive:

- more men affected
- no father-to-son
- affected father → all daughters carriers
- affected woman needs an affected father

Mother × father	Sons	Daughters
XX × X^cY	all normal	all carriers
X^cX × XY	½ color-blind	½ carriers, ½ normal
X^cX × X^cY	½ color-blind	½ color-blind, ½ carriers
X^cX^c × XY	all color-blind	all carriers

Color blindness:

- red and green pigment genes side by side on the X
- about 1 man in 12, 1 woman in 200 (N. European ancestry)
- Dalton lacked the green pigment gene

Hemophilia: clotting factor missing.

- **A**: factor VIII, 1 in 4,000–5,000 boys
- **B** (Christmas disease): factor IX, 1 in 20,000 boys
- treated by injecting the factor
- Queen Victoria a carrier; Alexei had B (read 2009)
- hemophilic woman very rare, not impossible

COMMON MISTAKES

✗ A and T "double bond", G and C "triple bond"
✓ **two** and **three hydrogen bonds**.

✗ RrYy makes 16 kinds of gamete
✓ **four**; 16 is the meetings.

✗ Chargaff's rules on one strand
✓ only for two strands together.

✗ Y-linked example: hairy ears
✓ **SRY**.

✗ XY-linked example: total color blindness
✓ **SHOX**; that gene is on an autosome.

✗ A father passes color blindness to his son
✓ son gets his **Y**.

✗ 0.34 nm × turns
✓ × base pairs; one turn is 3.4 nm.