GENETICS · STUDY GUIDE

Exam 3 Study Guide

This guide covers chapters 5 and 6. Chapter 5 shows how bacteria and viruses move genes without meiosis, and how each route of transfer becomes a way to map. Chapter 6 zooms out to whole chromosomes: pieces lost, doubled, flipped or swapped, and whole chromosomes or whole sets gained. Work one chapter at a time. Read what to know, copy the formula table by hand, and try each worked example with the steps covered. Then take the ten item self-test. The cram sheet at the end pulls it all together.

WHAT THIS GUIDE COVERS

- Chapter 5 · nucleoid and plasmids, mutants and replica plating, the fluctuation test, F^+, Hfr and F′, interrupted mating, transformation, transduction, phage crosses, Benzer's rII work, Baltimore classes and retroviruses.
- Chapter 6 · chromosome parts and karyotypes, deletions, duplications, inversions, translocations, aneuploidy and nondisjunction, polyploidy, microdeletions and cancer rearrangements.

CHAPTER 5

Bacterial and Viral Genetics

What to know

Bacteria are fast, haploid and easy to count. Every mutation shows at once, because there is no second copy to hide it. Most bacteria have one circular chromosome packed into a **nucleoid** by supercoiling and nucleoid proteins. The *E. coli* chromosome is about 4.6 Mb with about 4,300 genes, and replication starts at one origin, *oriC*.

Plasmids replicate on their own. The F factor (about 100 kb) makes a cell a donor. R plasmids carry resistance genes, often on transposons. Col plasmids carry colicins, and virulence plasmids carry toxins. An **episome** can also integrate into the chromosome. Genotypes are lowercase italic (*leu*$^-$); phenotypes are roman with a capital (Leu$^-$).

A **prototroph** grows on minimal medium. An **auxotroph** needs a supplement. Resistance mutants are found by direct selection. Auxotrophs are found by **replica plating**: a velvet pad prints the master plate onto other media, and a colony missing from minimal medium is an auxotroph. The Luria and Delbrück **fluctuation test** showed that mutations arise at random before selection: independent cultures gave many zeros and a few jackpots.

In conjugation, an $F^+ \times F^-$ mating makes two F^+ cells but moves almost no chromosome. In an **Hfr** cell, F has integrated, so chromosome follows *oriT* in a fixed order. The pair usually breaks apart, so the recipient stays F^-. An **F′** carries a piece of chromosome and makes a partial diploid. Donor DNA must enter the recipient chromosome by an even number of crossovers, which needs RecA.

Interrupted mating maps genes by time of entry. The *E. coli* map runs 0 to 100 minutes, about 46 kb per minute. Later genes reach lower plateaus because pairs break apart over time. Overlapping orders from several Hfr strains only fit a circle.

Transformation is uptake of free DNA and is blocked by DNase. Avery, MacLeod and McCarty used it to show that DNA is the genetic material. Genes close together are cotransformed. In **generalized transduction**, phages such as P1 package random host fragments by headful packaging, about 2 minutes of map. In **specialized transduction**, faulty excision of lambda moves only *gal* or *bio*. Cotransduction frequency measures closeness, and the rarest class in a three factor cross marks the middle gene.

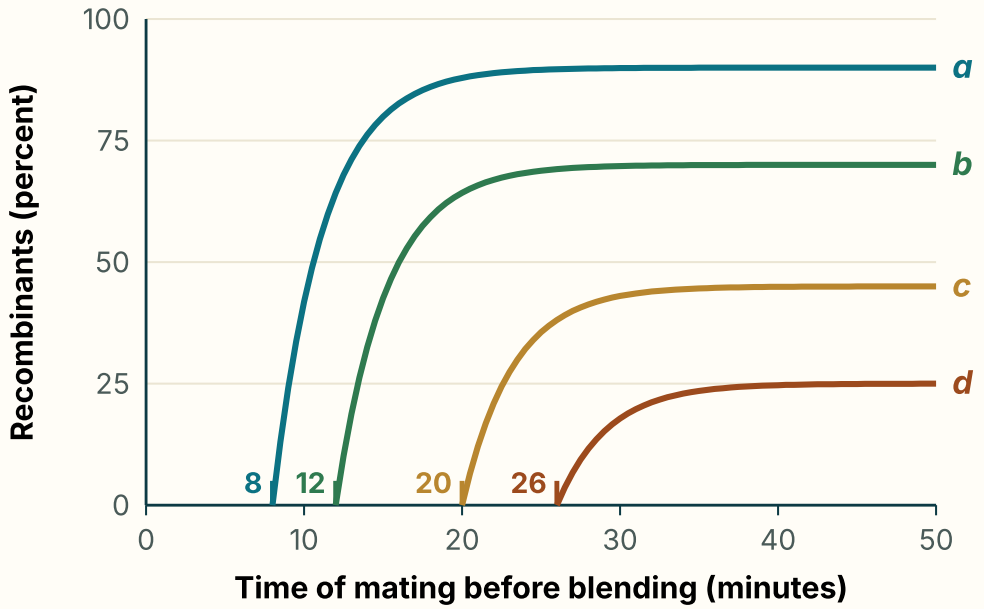
Lytic phages burst the cell. A temperate phage like lambda can instead integrate as a **prophage**, held silent by the cI repressor, until DNA damage triggers induction. Phage crosses are scored by plaque type. Benzer's rII system kept two questions apart: the **complementation test** (in K12(λ)) asks whether two mutations are in the same gene, and the **recombination test** asks whether they are at different sites. Deletion mapping and 2,000 mutations at about 300 sites showed a gene is a line of separable sites.

The **Baltimore classes** sort viruses by their route to mRNA. Plus strand RNA can act as mRNA. Minus strand and double stranded RNA viruses must carry their own polymerase. RNA viruses mutate fast because their polymerases do not proofread. Influenza's eight segments can **reassort**,

which causes antigenic shift. **Retroviruses** carry two plus strand RNA copies with *gag*, *pol* and *env*. Reverse transcriptase makes DNA flanked by LTRs, and integrase inserts it as a permanent **provirus**.

LOCK THESE IN

- Haploid means every mutation shows. Replica plating finds auxotrophs.
- Fluctuation test: mutations come first, selection only reveals them.
- F^+ gives F only. Hfr gives chromosome in order. F′ gives a partial diploid.
- Time of entry gives map position in minutes. 100 minutes total.
- DNase blocks transformation only. Conjugation needs contact.
- Generalized: any gene, about 2 minutes per particle. Specialized: only genes beside the prophage.
- The rarest class in a three factor transduction marks the middle gene.
- Benzer RF = 2 × titer on K12(λ) ÷ titer on B.



Entry time = map position
a 8, b 12, c 20, d 26 min

Order of entry gives
gene order: a b c d

Differences give distance:
b to c = 8 minutes,
about 370 kb
(1 minute ≈ 46 kb)

Later genes reach lower
plateaus: pairs break apart
(gradient of transfer)

Mapping by interrupted mating. Each gene first appears at its time of entry. Order of entry is gene order, and the gaps between entry times are distances in minutes.

Formulas and rules for microbes

Quantity	Formula or rule	Note
Mutation rate, P_0 method	$m = -\ln P_0$; rate ≈ m ÷ final cell number	counts events, not mutant cells
Map distance by conjugation	difference in times of entry	minutes; 1 min ≈ 46 kb
Missing interval	100 − sum of known intervals	the circle is 100 minutes
Cotransduction frequency	transductants with both markers ÷ selected transductants	higher means closer
Wu formula (P1)	$C = (1 - d/L)^3$, L = 2 min	solve $d = L(1 - C^{1/3})$
Linkage by cotransformation	observed double >> product of single rates	means one fragment carried both
Phage titer	plaques ÷ (mL plated × dilution)	plaque forming units per mL
Phage cross RF	recombinant plaques ÷ total plaques	parentals are the largest classes
Benzer rII RF	2 × (titer on K12(λ)) ÷ (titer on B)	doubles the hidden double mutant
Reassortment genotypes	$2^{\text{number of segments}}$	influenza: $2^8 = 256$
Mutations per genome	genome length × error rate	$P(\text{none}) = e^{-\text{mean}}$

Worked examples

WORKED EXAMPLE 1 · A CIRCULAR MAP FROM THREE HFR STRAINS

Three Hfr strains give these times of entry in minutes. Hfr A: *p* 4, *q* 18, *r* 26, *s* 43. Hfr B: *t* 6, *s* 20, *r* 37, *q* 45. Hfr C: *u* 5, *p* 33, *q* 47. Build the circle.

STEP 1 · SUBTRACT WITHIN EACH STRAIN

Hfr A: *p* to *q* 14, *q* to *r* 8, *r* to *s* 17. Hfr B: *t* to *s* 14, *s* to *r* 17, *r* to *q* 8. Hfr C: *u* to *p* 28, *p* to *q* 14.

STEP 2 · CHECK AGREEMENT

The shared intervals match: *q* to *r* is 8 and *r* to *s* is 17 in both A and B, and *p* to *q* is 14 in both A and C.

STEP 3 · STRING THE GENES

The order is *p*, *q*, *r*, *s*, *t*, *u* and back to *p*. Hfr B runs the other way, so its F sits in the opposite orientation.

STEP 4 · FILL THE GAP

Known intervals: 14 + 8 + 17 + 14 + 28 = 81 minutes. So *t* to *u* = 100 − 81 = **19 minutes**.

WORKED EXAMPLE 2 · THREE FACTOR TRANSDUCTION

P1 grown on an $a^+ b^+ c^+$ donor transduces an $a^- b^- c^-$ recipient. Of 500 selected a^+ transductants: $b^+ c^+$ 60, $b^+ c^-$ 150, $b^- c^-$ 286, $b^- c^+$ 4. Find the cotransduction frequencies, the order and the *a* to *b* distance.

STEP 1 · A WITH B

Classes with b^+: 60 + 150 = 210. So 210 ÷ 500 = **0.42**.

STEP 2 · A WITH C

Classes with c^+: 60 + 4 = 64. So 64 ÷ 500 = **0.128**.

STEP 3 · THE ORDER

b travels with *a* more often, so *b* is closer. The rarest class, $b^- c^+$ (4), needs four crossovers if *b* is in the middle. So the order is ***a, b, c***.

STEP 4 · WU FORMULA

$0.42 = (1 - d/2)^3$. The cube root of 0.42 is 0.749, so d/2 = 0.251 and d = **0.50 minute**.

WORKED EXAMPLE 3 · A BENZER RECOMBINATION TEST

Two rII mutants are crossed in *E. coli* B. The lysate is diluted 10^{-7} and 0.1 mL on B gives 250 plaques. Diluted 10^{-3}, 0.1 mL on K12(λ) gives 40 plaques. Find the recombination frequency.

STEP 1 · TITER ON B

$250 \div (0.1 \times 10^{-7}) = 2.5 \times 10^{10}$ per mL. This is all progeny.

STEP 2 · TITER ON K12(Λ)

$40 \div (0.1 \times 10^{-3}) = 4 \times 10^5$ per mL. This is wild type recombinants only.

STEP 3 · FRACTION WILD TYPE

$4 \times 10^5 \div 2.5 \times 10^{10} = 1.6 \times 10^{-5}$.

STEP 4 · DOUBLE IT

The reciprocal double mutant is invisible on K12(λ), so RF = $\mathbf{3.2 \times 10^{-5}}$. The two sites are very close.

WORKED EXAMPLE 4 · A MUTATION RATE FROM ZEROS

Thirty independent cultures each grow to 1×10^8 cells. Twelve give no resistant colonies when plated on phage. Estimate the mutation rate.

STEP 1 · FRACTION WITH NONE

$P_0 = 12 \div 30 = 0.40$.

STEP 2 · MEAN EVENTS PER CULTURE

$P_0 = e^{-m}$, so $m = -\ln 0.40 = 0.916$.

STEP 3 · PER DIVISION

Rate $\approx 0.916 \div 1 \times 10^8 =$ **9.2×10^{-9} per cell per division**.

WHY ZEROS

A jackpot culture holds a huge clone from one early event. Counting zeros counts events, not mutant cells.

TRAPS PEOPLE FALL INTO

- **Thinking F^+ cells move chromosome genes well.** They pass F itself. Hfr cells move chromosome.
- **Expecting Hfr recipients to become donors.** The rest of F enters last, so recipients nearly always stay F^-.
- **Mixing up which transfer DNase blocks.** Only transformation uses naked DNA.
- **Calling specialized transduction a mapping tool.** It moves only genes beside the prophage site.
- **Forgetting the factor of 2 in Benzer's RF.** The double mutant reciprocal does not grow on K12(λ).
- **Blending complementation with recombination.** Complementation: same gene? Recombination: same site?
- **Saying a single crossover brings donor DNA in.** A single crossover opens the circle. It takes an even number.
- **Assuming RNA viruses proofread.** Most do not. Coronaviruses are the exception.

Chapter 5 self-test · ten items

1. A colony grows on complete medium and on minimal plus histidine, but not on minimal alone. What is it?

- **A** A streptomycin resistant mutant
- **B** A temperature sensitive mutant
- **C** A histidine prototroph
- **D** A histidine auxotroph

D. A histidine auxotroph. It needs histidine supplied, so it cannot make its own. A prototroph would grow on minimal medium.

2. What did the Luria and Delbrück fluctuation test show?

- **A** The phage causes resistance in the cells it happens to touch
- **B** Resistance is passed to neighbors through cell contact
- **C** Resistant mutants arise at random before the phage is added
- **D** Every culture has about the same number of resistant cells

C. Resistant mutants arise at random before the phage is added. Independent cultures varied hugely, with many zeros and a few jackpots. That only fits mutations arising during growth, before selection.

3. In an Hfr × F⁻ mating, why does the recipient usually stay F⁻?

- **A** The F factor transfers only to other Hfr cells
- **B** The rest of F enters last, after the whole chromosome
- **C** The F factor is destroyed by the recipient's RecA
- **D** Hfr cells carry none of the *tra* genes of their own

B. The rest of F enters last, after the whole chromosome. Transfer starts in the middle of the integrated F, so the remaining F sequences come last, about 100 minutes later. Mating pairs nearly always break first.

4. Why do genes that enter late in interrupted mating reach lower plateaus?

A. Mating pairs break apart more often as time passes

B. Late genes are farther from the recipient's origin

C. Late genes recombine by a single crossover only

D. Late genes are blocked by DNase in the medium

A. Mating pairs break apart more often as time passes. This is the gradient of transfer. The longer transfer runs, the more pairs have separated on their own.

5. DNA from a donor is added to recipient cells, and DNase in the medium blocks gene transfer. Which process was it?

A. Transformation

B. Transduction

C. Conjugation

D. Lysogenic conversion

A. Transformation. Only transformation uses free DNA. Conjugation is protected inside the mating pair, and transduction inside a phage head.

6. A lambda lysate transduces *gal* at high frequency but never transduces *trp*. Why?

A. It is generalized transduction by headful packaging

B. It is specialized transduction from the site beside *gal*

C. Lambda can transduce only genes in the F factor

D. The *trp* genes are far too large to fit in a phage head

B. It is specialized transduction from the site beside *gal*. Lambda integrates between *gal* and *bio*. Faulty excision picks up only those neighbors.

7. Phages $h^-\ r^+$ and $h^+\ r^-$ coinfect cells. Of 1,000 plaques, 150 are $h^+\ r^+$ or $h^-\ r^-$. What is the RF?

A. 7.5 percent

B. 30 percent

C. 15 percent

D. 85 percent

C. 15 percent. The input types are parental, so the other two classes are recombinant. 150 ÷ 1,000 = 0.15.

8. Two rII mutants coinfect K12(λ) and the cells lyse. What does that tell you?

A. The mutations are both in *rIIA*

B. The mutations are at the very same site

C. The mutations are dominant to wild type

D. The mutations are in different cistrons

D. The mutations are in different cistrons. Lysis in this trans test means each phage supplied what the other lacked. That is complementation, so the mutations are in rIIA and rIIB, one each.

9. Which virus class can use its own genome directly as mRNA?

A. Plus strand RNA viruses, such as poliovirus

B. Double stranded RNA viruses, such as rotavirus

C. Minus strand RNA viruses, such as influenza

D. Retroviruses, such as HIV, after reverse transcription

A. Plus strand RNA viruses, such as poliovirus. A plus strand genome can be read by host ribosomes at once, which is why the naked RNA is infectious. Minus strand and dsRNA viruses carry a polymerase.

10. HIV has about 9,700 nucleotides and makes about 3×10^{-5} errors per nucleotide per cycle. About how many new mutations does one genome get per cycle?

A About 0.003

B About 0.3

C About 30

D About 3

B. About 0.3. $9{,}700 \times 3 \times 10^{-5} = 0.29$. With many virions per day, every single base change arises often, which is why drugs are combined.

Read the full chapter: Chapter 5 · Bacterial and Viral Genetics

CHAPTER 6

Chromosome Mutations

What to know

A working chromosome needs many replication origins, one centromere and two telomeres. Human centromeres sit on alpha satellite DNA but are marked by the histone variant CENP-A. Human telomeres are TTAGGG repeats bound by shelterin. An acentric piece is lost at division, a dicentric can break, and a broken end without a telomere tends to fuse.

The short arm is p and the long arm is q. By centromere position a chromosome is metacentric, submetacentric, acrocentric or telocentric. Human 13, 14, 15, 21 and 22 are acrocentric, and their short arms carry repeated rRNA genes. A **karyotype** uses metaphase cells and G-banding. Bands are numbered outward from the centromere, so 7q31.2 is chromosome 7, long arm, region 3, band 1, sub-band 2. Routine bands are about 5 to 10 Mb. FISH and chromosomal microarray see much smaller changes, but a microarray misses balanced rearrangements.

A **deletion** removes a segment. It can uncover a recessive allele on the other homolog (**pseudodominance**) and can expose **haploinsufficiency**. Loss of part of 5p causes cri du chat

syndrome. A **duplication** adds a segment, often by unequal crossing over. Bar in flies shows a position effect. Duplication of *PMP22* causes Charcot Marie Tooth type 1A, and its reciprocal deletion causes a different neuropathy.

An **inversion** flips a segment. Paracentric inversions leave out the centromere; pericentric ones include it. Heterozygotes pair by forming a loop. A crossover in a paracentric loop gives a dicentric bridge and an acentric fragment. In a pericentric loop it gives duplication plus deficiency products. Only the two chromatids in the crossover are unbalanced, so inversions suppress recombinant offspring.

A **reciprocal translocation** swaps ends between nonhomologous chromosomes. Carriers are usually healthy but form a cross shaped quadrivalent. Alternate segregation gives balanced gametes. Adjacent segregation gives unbalanced ones, so carriers are often semisterile. A **Robertsonian translocation** fuses two acrocentric long arms. The carrier has 45 chromosomes and is healthy, but some children may have translocation Down syndrome.

Aneuploidy adds or removes single chromosomes and upsets gene balance. Nondisjunction in meiosis I puts both homologs together and makes all four products abnormal. In meiosis II it puts two sister copies together and leaves half normal. Markers near the centromere tell you which division failed. Trisomies 21, 18 and 13 and the sex chromosome aneuploidies survive to birth. Free trisomy 21 risk rises with maternal age, from about 1 in 1,250 at 25 to 1 in 30 at 45. Mitotic errors give mosaics, and trisomy rescue can give uniparental disomy.

Polyploids have extra whole sets, which keeps gene balance. An **autopolyploid** has sets from one species. Selfing an *AAaa* autotetraploid gives 35:1, and triploids are nearly sterile, which is why bananas and some watermelons are seedless. An **allopolyploid** combines sets from two species. Doubling a sterile hybrid gives a fertile new species, as in bread wheat (AABBDD, 2n = 6x = 42).

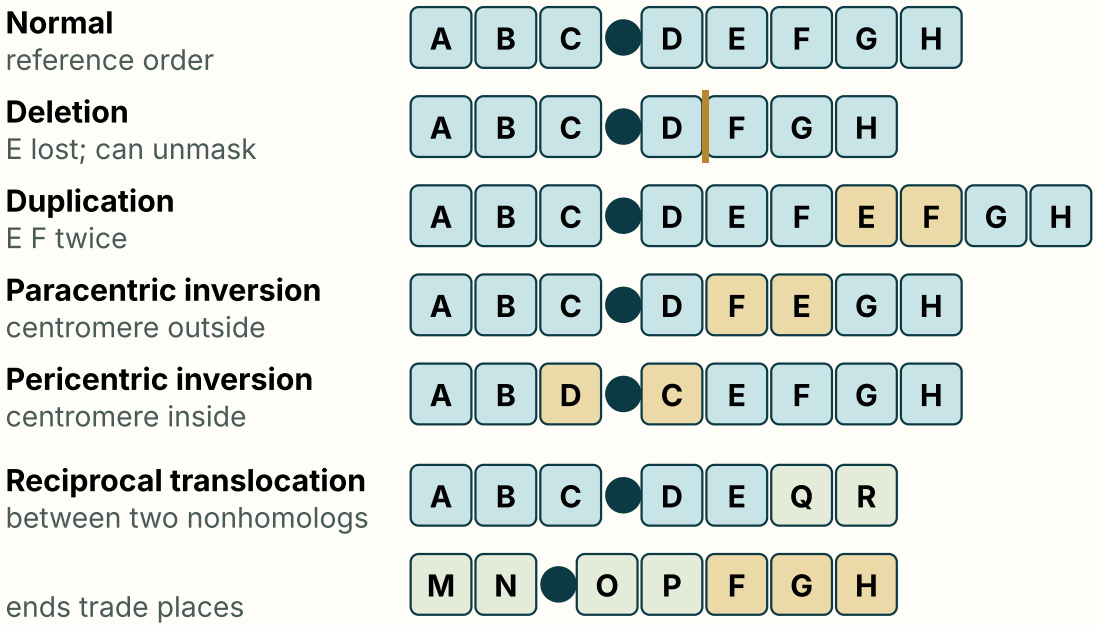
About half of first trimester losses are chromosomally abnormal, and about 1 in 150 newborns has a chromosome change. Microdeletions flanked by repeats cause syndromes such as 22q11.2 deletion and Williams syndrome. In cancer, t(9;22) makes the **Philadelphia chromosome** and the BCR-ABL1 kinase of chronic myeloid leukemia, which imatinib blocks. Other changes place an

oncogene by a strong enhancer, as with MYC in Burkitt lymphoma, amplify an oncogene, as with *ERBB2*, or delete a tumor suppressor.

LOCK THESE IN

- p is short, q is long. Bands count outward from the centromere.
- Microarray finds gains and losses, never balanced rearrangements.
- Paracentric loop crossover: dicentric bridge plus acentric fragment. Pericentric: duplication plus deficiency.
- Only half the products of a crossover meiosis in an inversion loop are unbalanced.
- Alternate segregation is balanced. Adjacent segregation is not.
- Meiosis I nondisjunction: both homologs. Meiosis II: two identical sisters.
- *AAaa* autotetraploid gametes are 1 : 4 : 1; selfing gives 35 : 1.
- Triploid balanced gametes = $2 \times (1/2)^{x}$.

Letters are gene blocks and the dark dot is the centromere. Gold marks the changed part.



Translocation start: A B C • D E F G H and M N • O P Q R. F G H and Q R trade places.
Duplications often arise by unequal crossing over. A deletion can unmask a recessive allele.

Rearrangements, block by block. Ask two questions of any rearrangement: is genetic material lost or gained, and does the changed segment include the centromere?

Formulas and rules for chromosomes

Quantity	Formula or rule	Note
Arm ratio	q length ÷ p length	1.0 to 1.7 metacentric; 1.7 to 3.0 submetacentric; above 3.0 acrocentric
Centromeric index	p length ÷ total length	about 0.5 for metacentric
Unbalanced products, inversion	½ × fraction of meioses with a loop crossover	same half rule as RF
Aneuploid names	2n − 2, 2n − 1, 2n + 1, 2n + 2	nullisomy, monosomy, trisomy, tetrasomy
Meiosis I nondisjunction	two n + 1 and two n − 1	all four abnormal
Meiosis II nondisjunction	one n + 1, one n − 1, two n	half abnormal
Trisomic *AAa* gametes	1 *AA* : 2 *Aa* : 2 *A* : 1 *a*	random pairs of three
Autotetraploid *AAaa* gametes	1 *AA* : 4 *Aa* : 1 *aa*	choose 2 of 4 = 6 ways
Selfing *AAaa*	35 dominant : 1 recessive	$(1/6)^2$ = 1/36 recessive
Triploid balanced gametes	2 × $(1/2)^x$	x = chromosomes in one set
Polyploid notation	2n = number in body cell; n = gamete; x = basic set	bread wheat 2n = 6x = 42, n = 21
Robertsonian carrier	45 chromosomes; six gamete types; three viable outcomes	der(21;21): every viable child has Down syndrome

Worked examples

WORKED EXAMPLE 1 · READING A KARYOTYPE AND AN ARM RATIO

Read 46,XX,inv(9)(p12q13). Then classify a chromosome with p = 1.8 µm and q = 6.2 µm.

STEP 1 · THE KARYOTYPE

46 chromosomes, female, with an inversion in chromosome 9 whose breakpoints are on both arms. A segment that spans both arms holds the centromere, so this is a **pericentric inversion**.

STEP 2 · ARM RATIO

q/p = 6.2 ÷ 1.8 = 3.44.

STEP 3 · CENTROMERIC INDEX

p/total = 1.8 ÷ 8.0 = 0.225.

STEP 4 · CLASSIFY

An arm ratio above 3.0 means **acrocentric**.

WORKED EXAMPLE 2 · POLLEN LOSS FROM AN INVERSION

A plant is heterozygous for a pericentric inversion. One crossover falls inside the loop in 40 percent of meioses, and none in the rest. What fraction of its pollen is unbalanced?

STEP 1 · CROSSOVER MEIOSES

Two of the four chromatids took part in the crossover. They carry a duplication and a deficiency. So half the products are unbalanced.

STEP 2 · OTHER MEIOSES

With no crossover in the loop, all four products are balanced.

STEP 3 · COMBINE

0.40 × 1/2 + 0.60 × 0 = **0.20**, so 20 percent of pollen is unbalanced.

COMMON SLIP

Do not call all four products of a crossover meiosis unbalanced. Only the two that crossed over are.

WORKED EXAMPLE 3 · A TETRAPLOID TESTCROSS

An autotetraploid *AAaa* plant (gene near the centromere, random chromosome pairing) is crossed to an *aaaa* plant. *A* is fully dominant. What phenotypic ratio do you expect?

STEP 1 · GAMETES OF *AAAA*

Choosing 2 of 4 gives 6 pairs: one *AA*, four *Aa*, one *aa*.

STEP 2 · GAMETES OF *AAAA*

All are *aa*.

STEP 3 · COMBINE

AAaa 1/6, *Aaaa* 4/6, *aaaa* 1/6. The first two show the dominant trait.

STEP 4 · RATIO

5 dominant : 1 recessive, not the 1:1 of a diploid testcross.

WORKED EXAMPLE 4 · WHY TRIPLOIDS ARE STERILE

A triploid plant has x = 9. For each chromosome type, a gamete gets one copy or two with equal chance. What fraction of gametes are balanced?

STEP 1 · ALL SINGLE

P(one copy of all 9) = $(1/2)^9$ = 1/512.

STEP 2 · ALL DOUBLE

P(two copies of all 9) = 1/512.

STEP 3 · ADD

2/512 = **1/256, about 0.4 percent**. Nearly all gametes are unbalanced.

TRAPS PEOPLE FALL INTO

- **Using a microarray to find a balanced translocation.** No DNA is gained or lost, so the array looks normal.
- **Calling an inversion with breaks on p and q paracentric.** If it spans the centromere, it is pericentric.
- **Thinking inversions stop crossing over.** Crossovers happen. The recombinant products just do not survive.
- **Treating a Robertsonian carrier as affected.** The lost short arms carry only redundant rRNA genes.
- **Mixing up the two nondisjunction outcomes.** Two different parental homologs point to meiosis I. Two identical copies point to meiosis II.
- **Thinking polyploidy is worse than aneuploidy.** Whole sets keep gene ratios. Single extra chromosomes do not.
- **Confusing x and n.** x is one basic set. n is what a gamete holds.
- **Calling the Philadelphia chromosome an extra chromosome.** It is a shortened der(22) from a reciprocal t(9;22).

Chapter 6 self-test · ten items

1. What does 46,XY,del(5)(p15.2) describe?

A. A male with a balanced translocation involving 5

B. A male with an extra copy of chromosome 5 in each cell

C. A male with a duplication on the long arm of 5

D. A male missing part of the short arm of chromosome 5

D. A male missing part of the short arm of chromosome 5. del means deletion, (5) is the chromosome and p15.2 is the breakpoint on the short arm. This is the cri du chat deletion.

2. A recessive allele shows its phenotype in a heterozygote because the other homolog has lost that region. What is this called?

A. Haploid rescue

B. A position effect

C. Pseudodominance

D. Codominance

C. Pseudodominance. A deletion uncovers the recessive allele, so it is expressed as if dominant. This is why deletions are good mapping tools.

3. A crossover inside a paracentric inversion loop gives which unbalanced products?

A. One trisomic and one monosomic gamete each time

B. Two chromatids with a duplication and a deficiency

C. Two balanced chromatids with all genes in order

D. A dicentric chromatid and an acentric fragment

D. A dicentric chromatid and an acentric fragment. With the centromere outside the loop, the recombinants have two centromeres or none. The dicentric forms a bridge and breaks, and the acentric is lost.

4. Why are reciprocal translocation carriers often semisterile?

A. Adjacent segregation makes many unbalanced gametes

B. The translocated genes are all deleted in meiosis

C. Carriers make no crossovers on any chromosome

D. Alternate segregation makes only aneuploid gametes

A. Adjacent segregation makes many unbalanced gametes. Alternate segregation gives balanced gametes and adjacent segregation does not. In many plants the two happen about equally, so about half the pollen aborts.

5. A color blind 47,XXY boy has a carrier mother (X^AX^a) and a father with normal vision. Where did nondisjunction most likely occur?

A. The father, in meiosis II

B. The mother, in meiosis II

C. The mother, in meiosis I

D. The father, in meiosis I

B. The mother, in meiosis II. He must be X^aX^aY. The father has no X^a, so both X chromosomes are maternal and identical, which points to sisters failing to separate in meiosis II.

6. Which autosomal trisomy is the most common single finding in miscarriages but is never seen at birth?

A. Trisomy 18

B. Trisomy 13

C. Trisomy 16

D. Trisomy 21

C. Trisomy 16. Trisomy 16 is the most common trisomy in first trimester losses. Only trisomies 21, 18 and 13 commonly survive to birth.

7. An *AAaa* autotetraploid is selfed. What fraction of offspring show the recessive phenotype?

A 1/6

B 1/4

C 1/16

D 1/36

D. 1/36. Only *aa* × *aa* gametes give *aaaa*, and each *aa* gamete has frequency 1/6. So 1/6 × 1/6 = 1/36.

8. A sterile hybrid of two diploid species doubles its chromosome number and becomes fertile. Why?

A Every chromosome now has an identical partner to pair with

B Doubling makes the hybrid able to self fertilize

C Doubling turns the hybrid into an autopolyploid of one parent

D Doubling removes the chromosomes from one of the parents

A. Every chromosome now has an identical partner to pair with. In the hybrid the two sets cannot pair. After doubling, each chromosome has a homolog, so meiosis works. That is an allopolyploid, a new species in one step.

9. A woman carries der(21;21). What happens in her viable pregnancies?

A None are affected by it

B About one in three has it

C Every one has Down syndrome

D About half of them have it

C. Every one has Down syndrome. Her gametes carry two copies of 21q or none. With a normal sperm, the first gives three copies and the second monosomy 21, which is not viable.

10. How does t(8;14) cause Burkitt lymphoma?

A. It fuses *BCR* and *ABL1* into one kinase gene

B. It places *MYC* next to an immunoglobulin enhancer

C. It deletes the *RB1* tumor suppressor gene entirely

D. It amplifies *ERBB2* into many extra copies

B. It places *MYC* next to an immunoglobulin enhancer. The *MYC* protein is normal but overexpressed by the strong IGH enhancer. BCR-ABL1 comes from t(9;22).

Read the full chapter: Chapter 6 · Chromosome Mutations

ONE PAGE

Cram sheet

Everything from chapters 5 and 6 in one place. Cover the right column and see if you can rebuild each row.

Chapter 5 rules

Topic	Rule
Bacteria	haploid; one circular chromosome in a nucleoid; *E. coli* 4.6 Mb, about 4,300 genes
Mutants	prototroph grows on minimal; auxotroph needs a supplement; replica plating finds them
Fluctuation test	many zeros and jackpots; mutations arise before selection
Mating types	$F^+ \times F^-$ gives two F^+; Hfr moves chromosome in order; F′ gives a merozygote
Interrupted mating	time of entry in minutes; 100 minute circle; gradient of transfer
Transformation	free DNA; DNase sensitive; cotransformation means close
Generalized transduction	P1 or P22; headful; any gene; about 2 minutes; rarest class marks the middle gene
Specialized transduction	lambda; faulty excision; only *gal* or *bio*
Lysogeny	prophage held by cI repressor; DNA damage and RecA trigger induction
Benzer	complementation in K12(λ): same gene? Recombination: same site? RF = 2 × K12 ÷ B
Viruses	Baltimore classes by route to mRNA; RNA viruses mutate fast; segments reassort
Retroviruses	*gag*, *pol*, *env*; reverse transcriptase; LTRs; integrase makes a provirus

Chapter 6 rules

Topic	Rule
Essential parts	origins, one centromere (CENP-A), two telomeres (TTAGGG)
Shapes	metacentric, submetacentric, acrocentric, telocentric; human acrocentrics 13, 14, 15, 21, 22
Detection	G-banding about 5 to 10 Mb; FISH and microarray smaller; microarray misses balanced changes
Deletion and duplication	pseudodominance; haploinsufficiency; unequal crossing over; position effect
Inversions	paracentric: dicentric plus acentric; pericentric: duplication plus deficiency
Translocations	quadrivalent; alternate balanced; adjacent unbalanced; semisterility; pseudolinkage
Robertsonian	45 chromosomes, healthy; risk of translocation Down syndrome
Nondisjunction	MI: all four abnormal; MII: half abnormal
Births	trisomy 21 about 1 in 700; 45,X about 1 in 2,500 females; 47,XXY about 1 in 500 to 1,000 males
Polyploidy	*AAaa* gametes 1:4:1; selfed 35:1; triploids nearly sterile; allopolyploids are new species
Cancer	t(9;22) BCR-ABL1 and imatinib; t(8;14) MYC; t(15;17) PML-RARA; *ERBB2* amplification

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