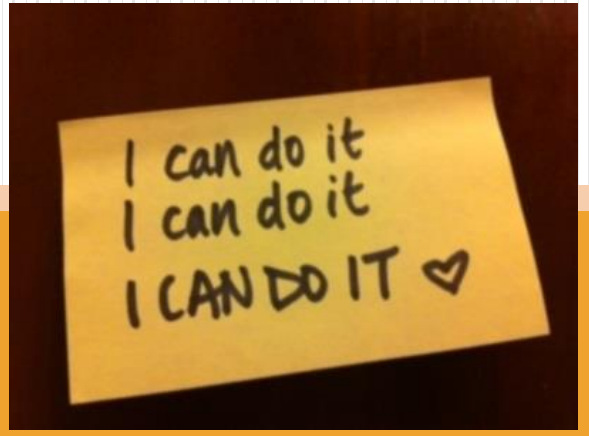
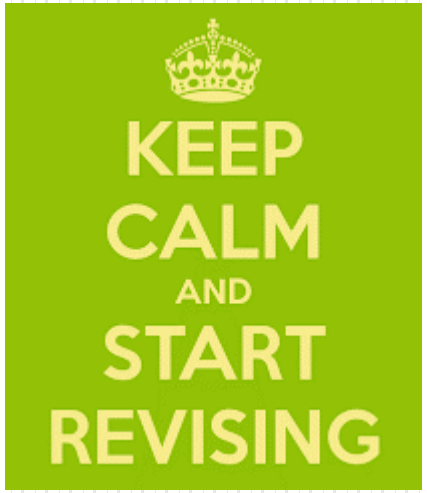


Revision Mid tri test

Class 11

SLE254 Genetics and Genomics

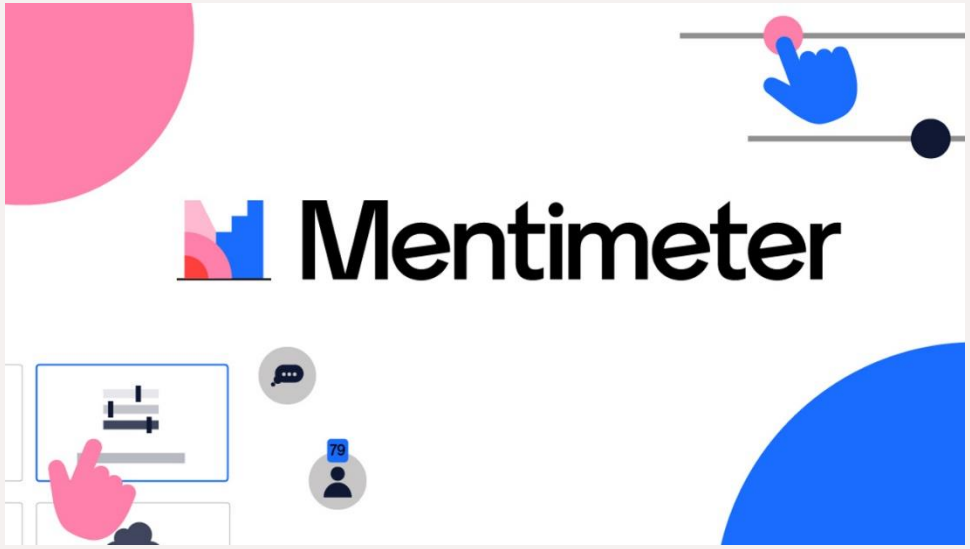
Concepts of Genetics (12th ed)



What’s Going on in Pass

Thursday 7pm Week 4- Karyotypes and Chromosomal Abnormalities

Monday 1pm Week 5- Special Online revision session (Practice questions and Quizzes)



SLE254 Mid-semester class test 1

- Online test
- Opens at 9AM on the 7th of August
- Closes at 9AM on the 8th of August
- 30 MCA, covers material from week 1- 4
- 50 minutes to complete
- Students can log in any time during that 24hr period
- Once you logged in, you will have to complete the test (time will be ticking away in the background even if you log out)
- You can go back and forth between the questions
- No 5pm class on the 7th of August. Students can use the extra hour to study or to complete the test.

Feeling under pressure and tempted to take short cuts?

- **Think again!**
- **Using gAI and not citing your program, prompt or quality control or gAI quiz breaker is cheating on an MCQ test in SLE254…**

However, it's important to understand that ChatGPT or any other AI tool should not replace your own critical thinking and analysis. There are known issues with how up to date and accurate the information provided by AI tools is, and inappropriate use may constitute cheating.

Guidelines for use

If you want to explore using ChatGPT or other AI tools to assist with your assignments or research, you should:

1. Use AI as a tool to assist you in your research and writing, but not as a replacement for your critical thinking and analysis.
2. Ensure that you appropriately cite and reference any text or output generated by AI in your assignment, along with any other sources you use. You should clearly indicate where in your assessment task you have used AI-generated material.
3. Understand the AI tool's limitations and use it in conjunction with other sources to ensure the credibility and reliability of the information you present. You need to check the accuracy of all information generated by AI tools.
4. Be aware of the University's student academic integrity policy and ensure that you follow it.
5. Make sure that the final product is your own work, and not just copied from an AI generator. You can use the generated text as a prompt for inspiration or guidance, but the final submitted assessment must be your work, creation, and analysis.

By following these guidelines, you can use generative AI as a valuable tool to assist you in your research and writing. Understanding how to use these tools correctly is essential to avoiding breaches that could impact your successful course progression and possibly even your graduation.

For more information, visit the Academic Integrity webpage.

The mid-tri test

TIPS

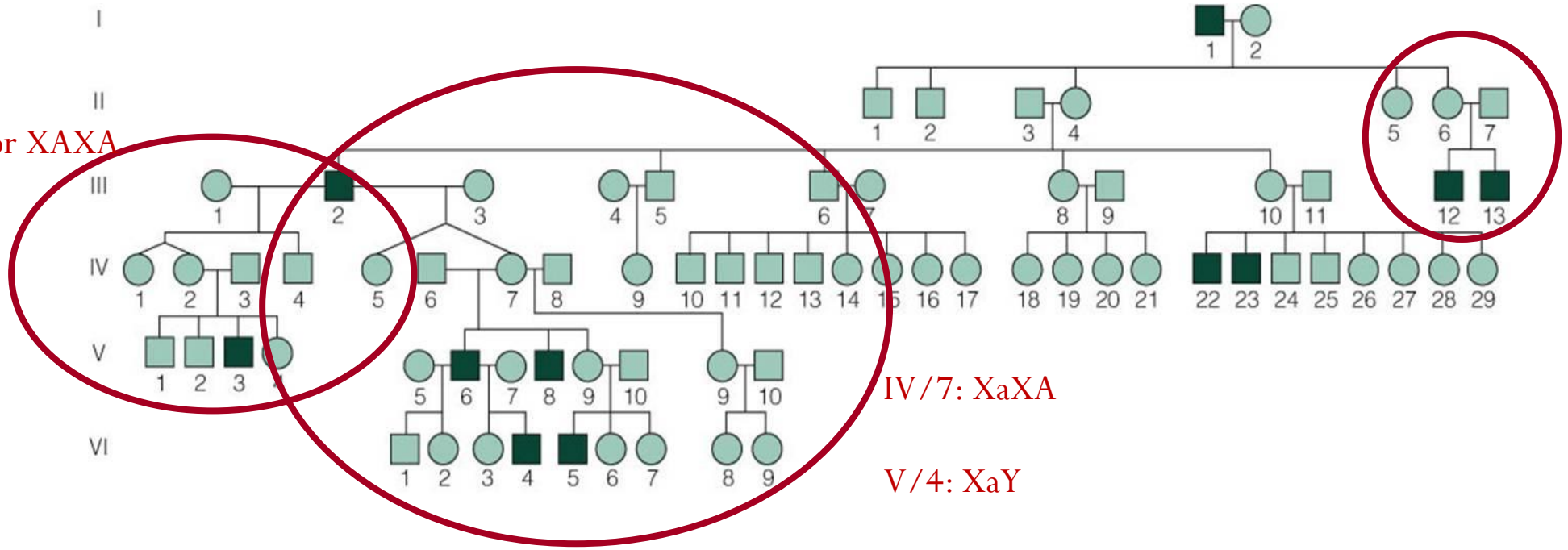
1.Chi square problems: get your punnett squares ready- work out genotypes based on the progeny ratios

2. Get your chi square obs/exp table ready

3. Be ready for the Binomial equation. You will be given all the information you need

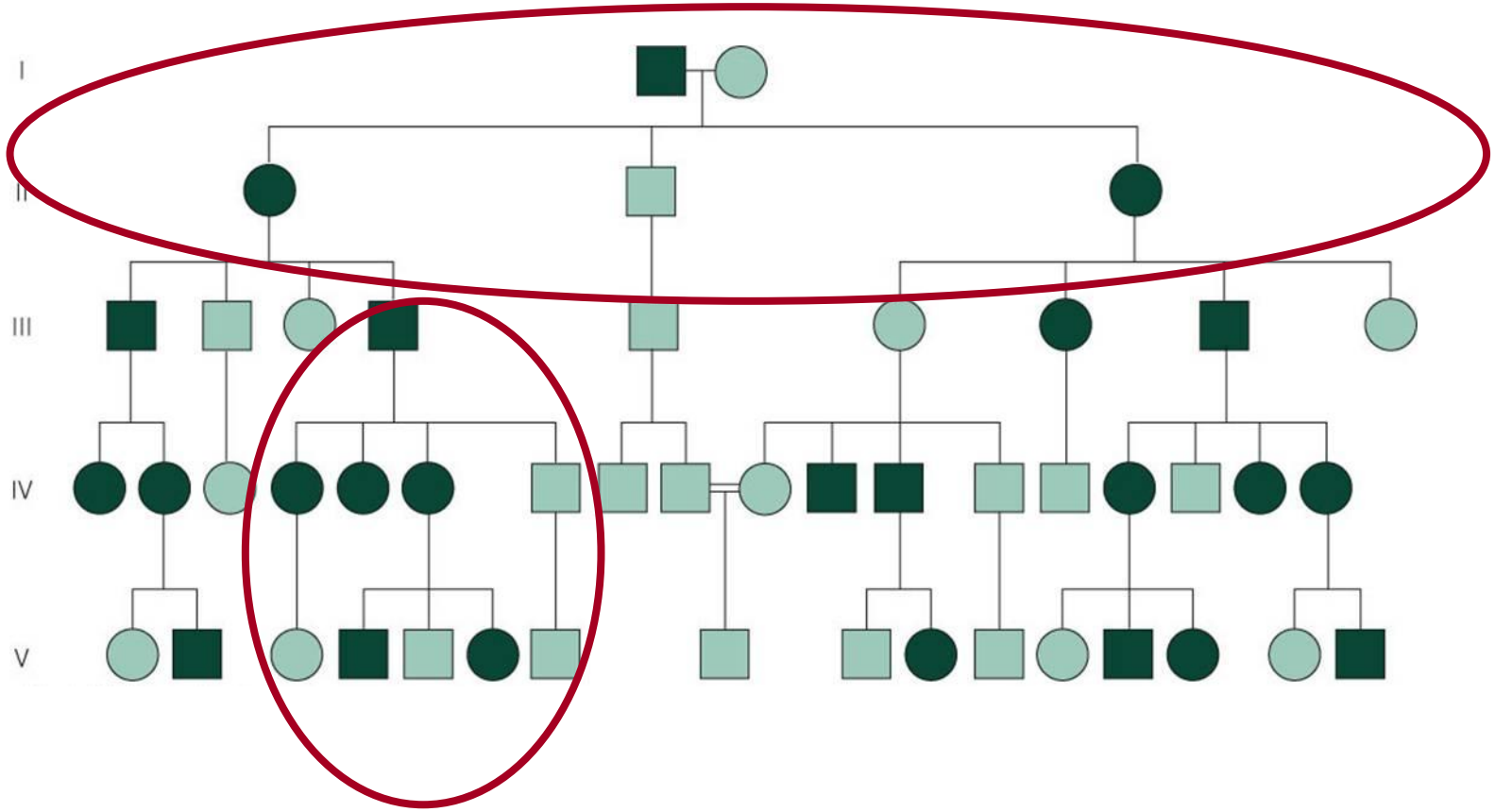
4. Get a Punnett square ready for a dihybrid cross

X-linked recessive pedigree



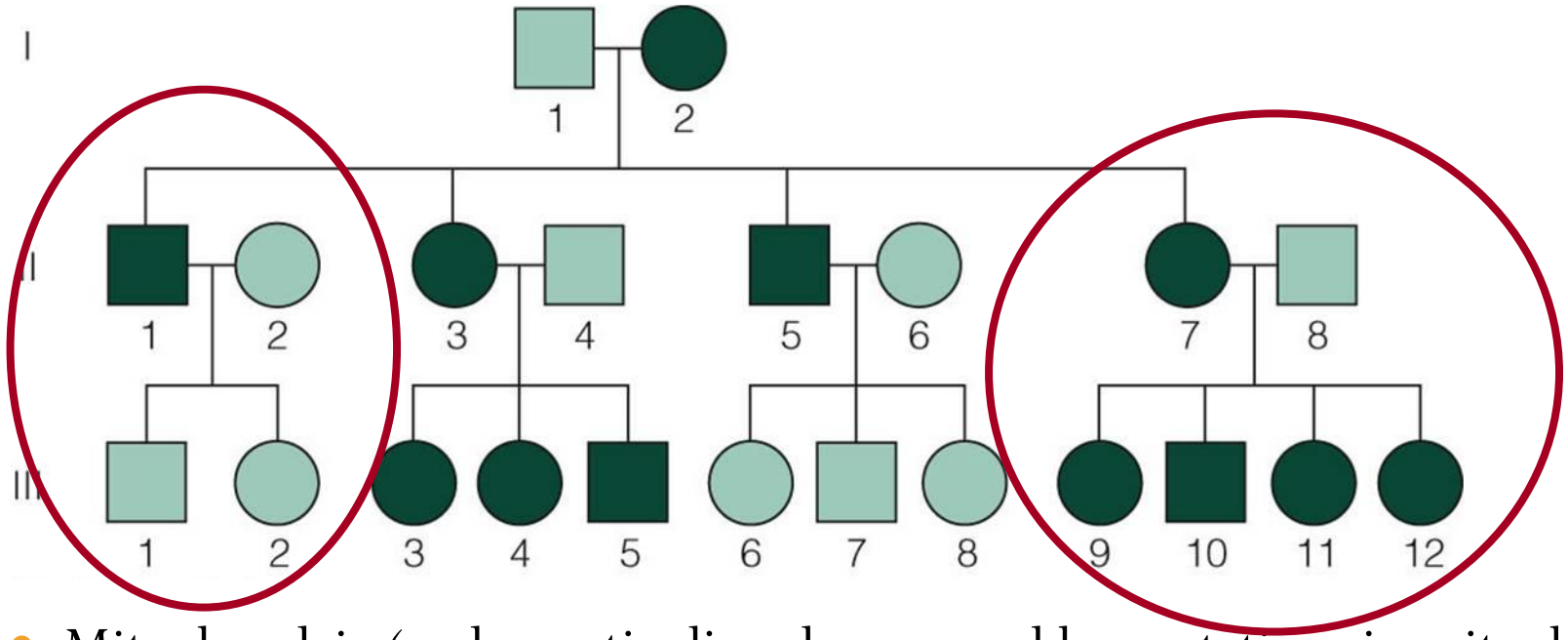
- Hemizygous males are affected and transmit the mutant allele to all their daughters who become carriers
- Phenotypic expression much more common in males

X-linked dominant pedigree



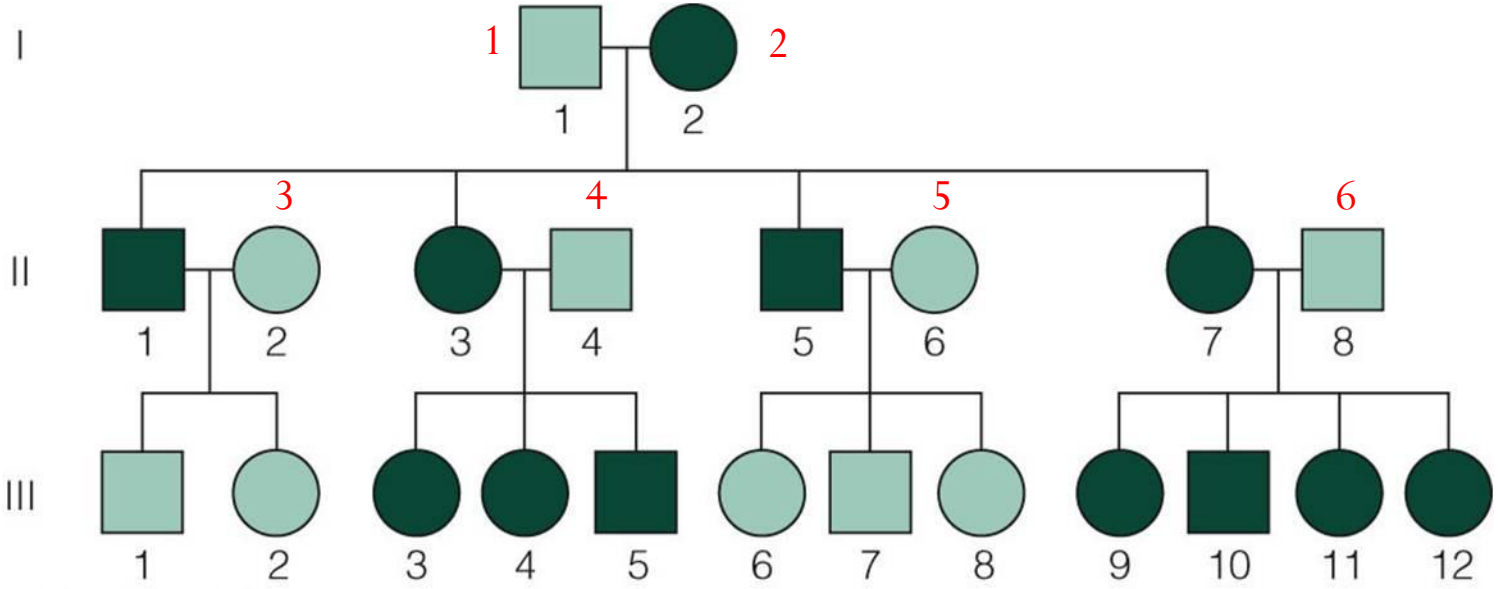
- Affected males produce all affected daughters and no affected sons.
- Affected females transmit the trait to about half their children.
- About twice as many females affected as males.

Mitochondrial gene pedigree



- Mitochondria (and genetic disorders caused by mutations in mitochondrial genes) are maternally inherited
- Both males and females can be affected, but only affected females pass on the trait
- How many mitochondrial genotypes do we have in this pedigree?

Mitochondrial gene pedigree



- Mitochondria (and genetic disorders caused by mutations in mitochondrial genes) are maternally inherited
- Both males and females can be affected, but only affected females pass on the trait
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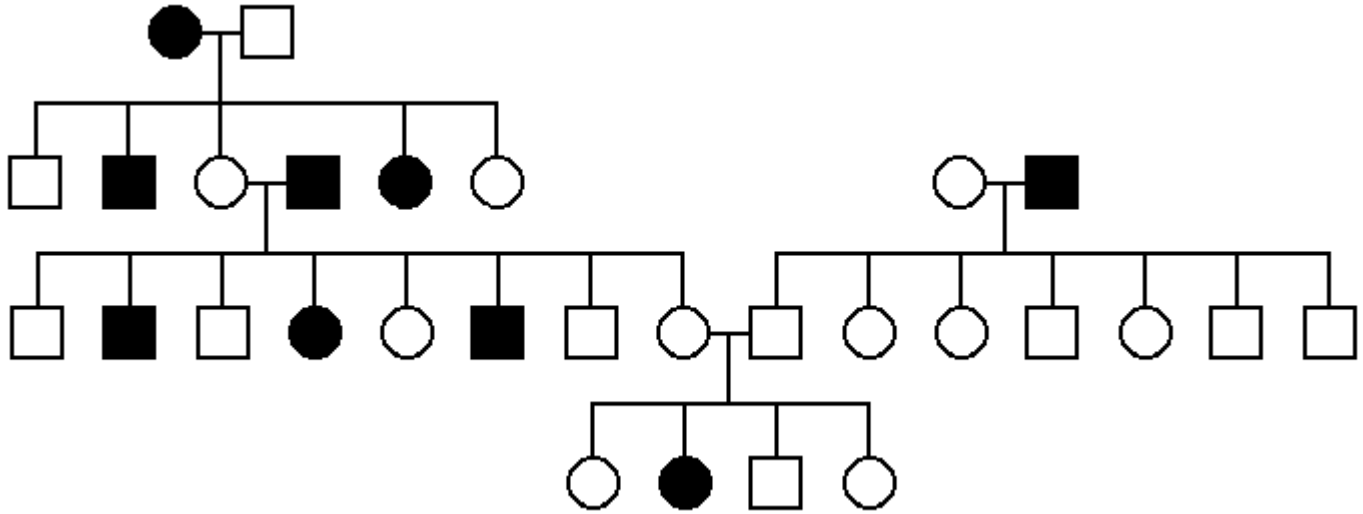
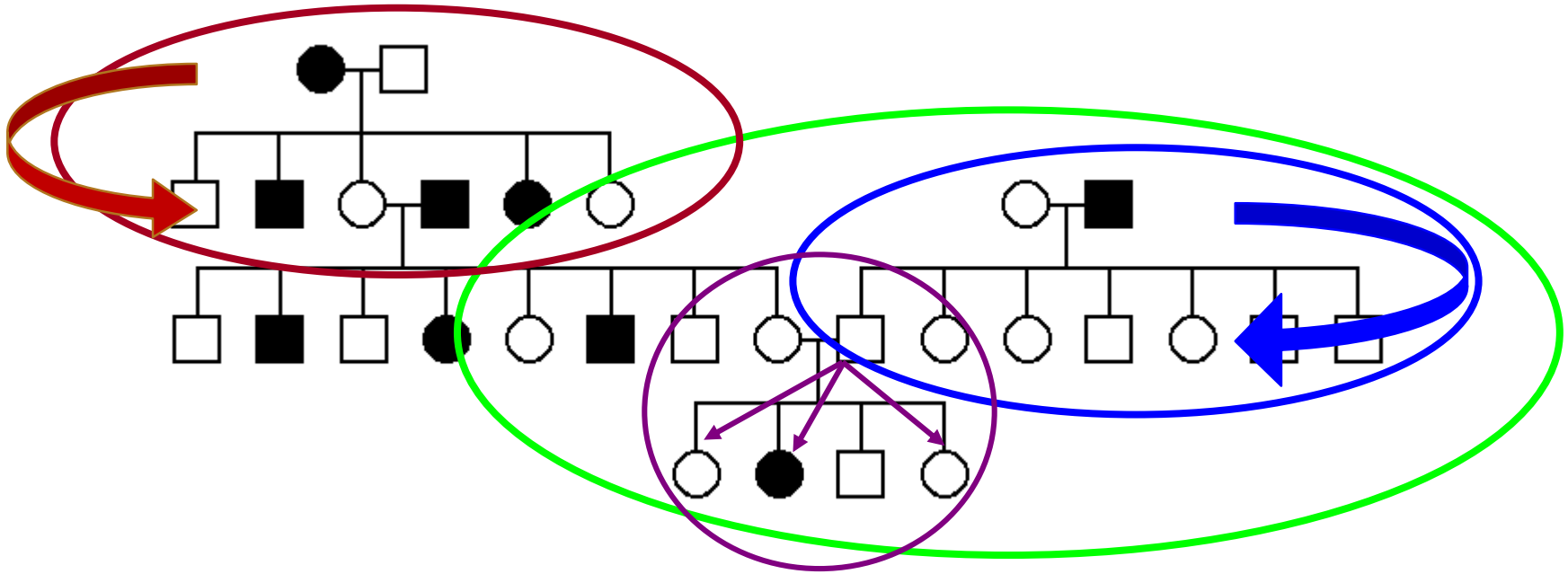
Now try these



- Which best describes the genetics of the afflicting allele in the pedigree?

- autosomal dominant
- autosomal recessive
- X-linked dominant
- X-linked recessive
- Y-linked dominant
- Y-linked recessive

Now try these



- Which best describes the genetics of the afflicting allele in the pedigree?

- autosomal dominant X
- autosomal recessive
- X-linked dominant X
- X-linked recessive X
- Y-linked dominant X
- Y-linked recessive X

Questions below are all based on the information provided below:

Assume that you have a garden and in that garden some pea plants have solid leaves and others have striped leaves. You conduct a series of crosses [(a) through (e)] and obtain the results given in the table.

cross		progeny	
		solid	striped
(a)	Solid X striped	0	57
(b)	Striped X striped	39	111
(c)	Striped X striped	45	150
(d)	Solid X solid	76	0
(e)	Solid X striped	107	93

Based on the observations in the table

A) Solid is the dominant allele
B) Striped is the dominant allele
C) These alleles are codominant
D) These alleles do not display Mendelian patterns of inheritance

$$\chi^2 = \Sigma \frac{(o - e)^2}{e}$$

Questions below are all based on the information provided below:

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$$\chi^2 = \Sigma \frac{(o - e)^2}{e}$$

MONOHYBRID CROSS

st = striped

so= solid

You can use:

S= striped s= solid

Punnett Square

A

	so	so
st	stso	stso
st	stso	stso

Punnett Square

B+ C

	st	so
st	stst	stso
so	stso	soso

Punnett Square

D

	so	so
so	so	so
so	so	so

Punnett Square

E

	so	st
so	soso	stso
so	soso	stso

If you were to conduct a chi square test on some of the crosses from this data set the expected ratios for cross (c) would be:

A. 3 solid: 1 striped
B. 1 solid: 1 striped
C. 4 solid: 0 striped
D. 3 striped: 1 solid

cross		progeny	
		solid	striped
(a)	Solid X striped	0	57
(b)	Striped X striped	39	111
(c)	Striped X striped	45	150
(d)	Solid X solid	76	0
(e)	Solid X striped	107	93

Using the formula below the table, the critical chi square value for data obtained from cross (c) would be (NOTE all values were rounded to 4 significant figures for this calculation):

A. 0.98
B. 97.00
C. 102.2
D. 0.3846 ⟸

$$\chi^2 = \Sigma \frac{(o-e)^2}{e}$$

cross		progeny	
		solid	striped
(a)	Solid X striped	0	57
(b)	Striped X striped	39	111
(c)	Striped X striped	45	150
(d)	Solid X solid	76	0
(e)	Solid X striped	107	93

You are looking at cross C which has 45 solid and 150 striped

Total number is 195
And it's a heterozygote cross (as we have two parents both showing dominant phenotype but producing recessive offspring) so we expect 3:1 ratio (0.75 : 0.25)

So expected numbers are:
195x0.75 = 146.25
195x0.25 = 48.75

So the calculation is:
(150-146.25)^2/ 146.25 = 0.0962 (to 4 significant figures)
(45 – 48.75.5)^2/48.75 = 0.2885 (to 4 significant figures)
0.0962 + 0.2885 = 0.3846 which is D

	Expected ratio in a monohybrid cross	Cross expected ratio	Observed (o)	Expected	(o-e)	Deviation(d^2)	d^2/e
SS+Ss (striped)	3 in 4	0.75	150	146.25	3.7500	14.0625	0.0962
ss (solid)	1 in 4	0.25	45	48.75	-3.7500	14.0625	0.2885
			195				
							0.3846

Your chi square value is: 0.0962 + 0.2885 = 0.3846
Your df = 1

Critical chi square value at p = 0.05 is 3.84

You chi square value is < than the critical chi square value

	Probability (p)					
	0.90	0.50	0.20	0.05	0.01	0.001
1	0.02	0.46	1.64	3.84	6.64	10.83
2	0.21	1.39	3.22	5.99	9.21	13.82
3	0.58	2.37	4.64	7.82	11.35	16.27
4	1.06	3.36	5.99	9.49	13.28	18.47
5	1.61	4.35	7.29	11.07	15.09	20.52
6	2.20	5.35	8.56	12.59	16.81	22.46
7	2.83	6.35	9.80	14.07	18.48	24.32
8	3.49	7.34	11.03	15.51	20.09	26.13
9	4.17	8.34	12.24	16.92	21.67	27.88
10	4.87	9.34	13.44	18.31	23.21	29.59
15	8.55	14.34	19.31	25.00	30.58	37.30
25	16.47	24.34	30.68	37.65	44.31	52.62
50	37.69	49.34	58.16	67.51	76.15	86.60

χ^2 values

By comparing the chi square value you calculated to the critical chi square value table above, which statement would be true in its entirety?

A) The chi square value is less than the critical chi square value and therefore you **accept** the null hypothesis and any differences between observed and expected numbers are due to chance.

B) The chi square value is less than the critical chi square value and therefore you **accept** the null hypothesis and any differences between observed and expected numbers are **NOT** due to chance.

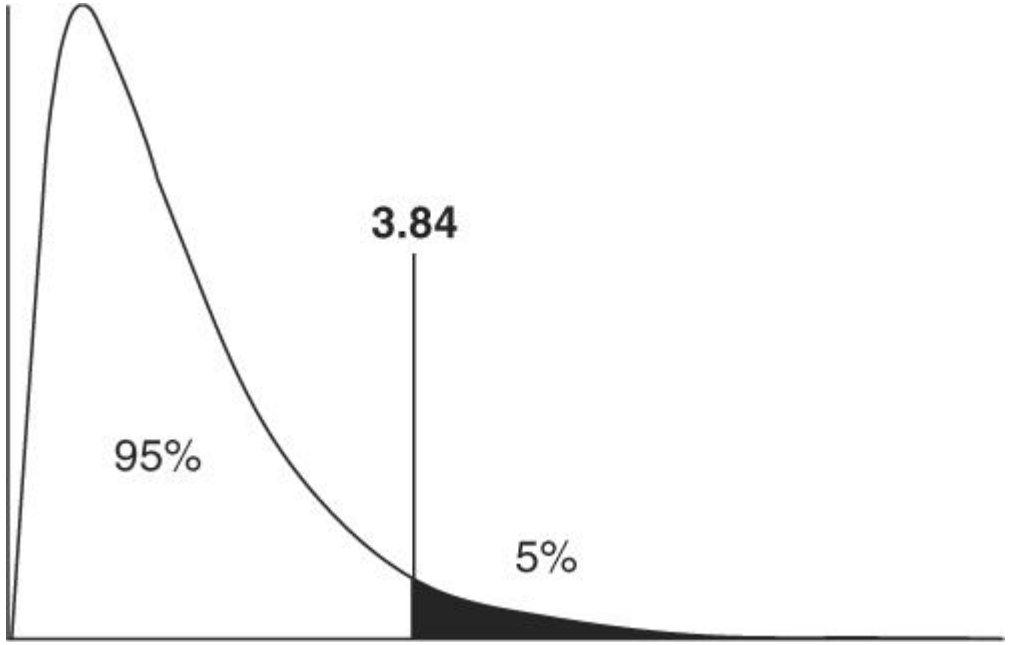
C) The chi square value is less than the critical chi square value and therefore you **reject** the null hypothesis and any differences between observed and expected numbers are due to chance.

D) The chi square value is less than the critical chi square value and therefore you **reject** the null hypothesis and any differences between observed and expected are **NOT** due to chance.

WHAT IS THE CHI SQUARE PROBABILITY TABLE TELLING ME?

Your chi square value is: 0.0962 + 0.2885 = **0.3846**, Your df = 1

Chi Square distribution 1 df
Critical chi square value at p = 0.05 is **3.84**



<3.84 Accept Null Hypothesis **=>3.84 Reject Null Hypothesis**

Probability that you would get your X2 value if the null hypothesis was true is <5%

	Probability (p)					
	0.90	0.50	0.20	**0.05**	**0.01**	**0.001**
1	0.02	0.46	1.64	3.84	6.64	10.83
2	0.21	1.39	3.22	5.99	9.21	13.82
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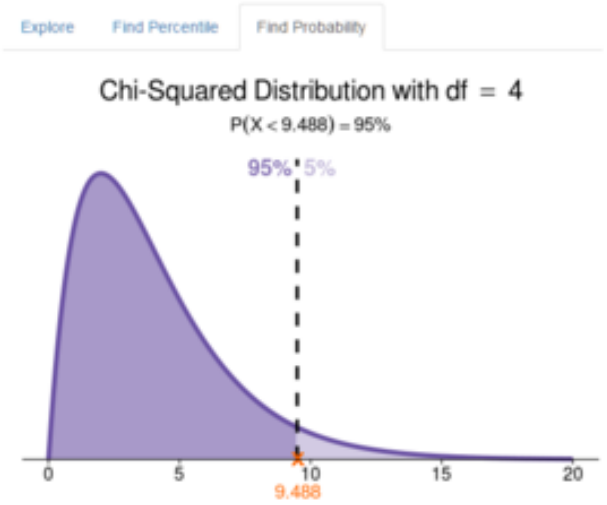
χ^2 values

You chi square value is < than the critical chi square value



The probability distribution varies with the df

the degrees of freedom are number of classes -1

	Probability (p)					
	0.90	0.50	0.20	0.05	0.01	0.001
1	0.02	0.46	1.64	3.84	6.64	10.83
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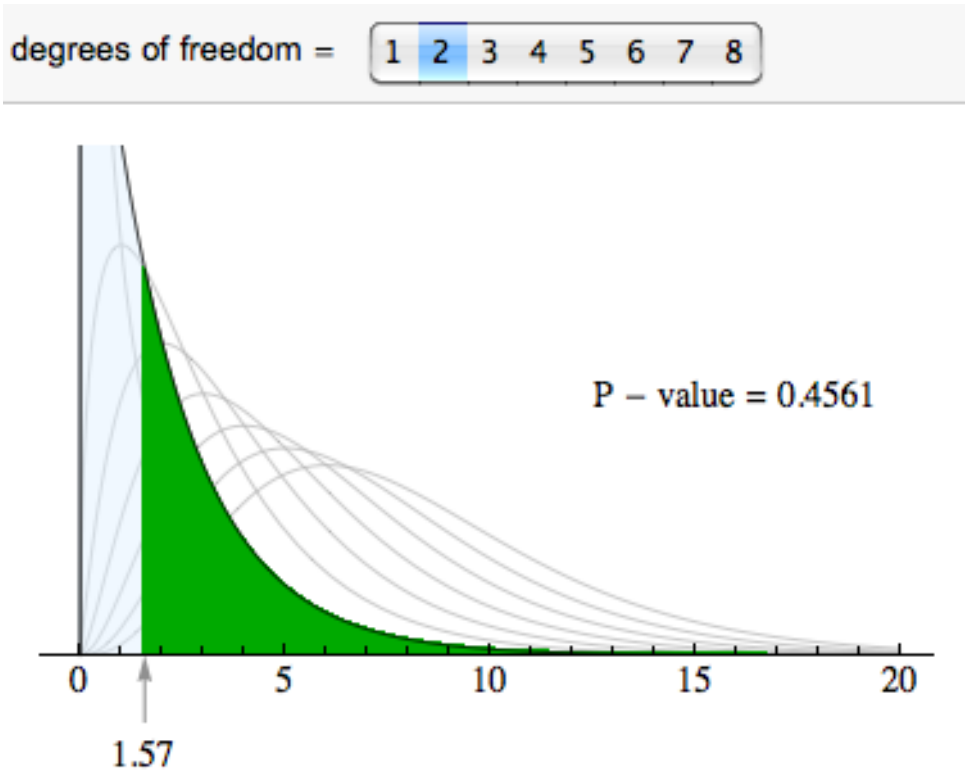
χ^2 values



Practice Test - Answers

1. At which stage of mitosis are chromosomes usually photographed in the preparation of a karyotype?

A) prophase

B) metaphase ← Because this is when the chromosomes are most condensed

C) anaphase

D) interphase

2. A triploid cell contains sets of three homologous chromosomes. If a cell of a usually diploid species with 42 chromosomes is triploid, this cell would expect to have which of the following?

A) 63 chromosomes in 31 & 1/2 pairs

B) 14 chromosomes, each with three chromatids

C) 21 chromosomes pairs and 21 unique chromosomes

→ D) 63 chromosomes in 3 sets of 21

Because the haploid (n) has 21 and the triploid is 3n = 3x21 = 63

3. How do cells at the completion of meiosis compare with cells that are in prophase of meiosis 1? They have____.

A) half the number of chromosomes and half the amount of DNA.

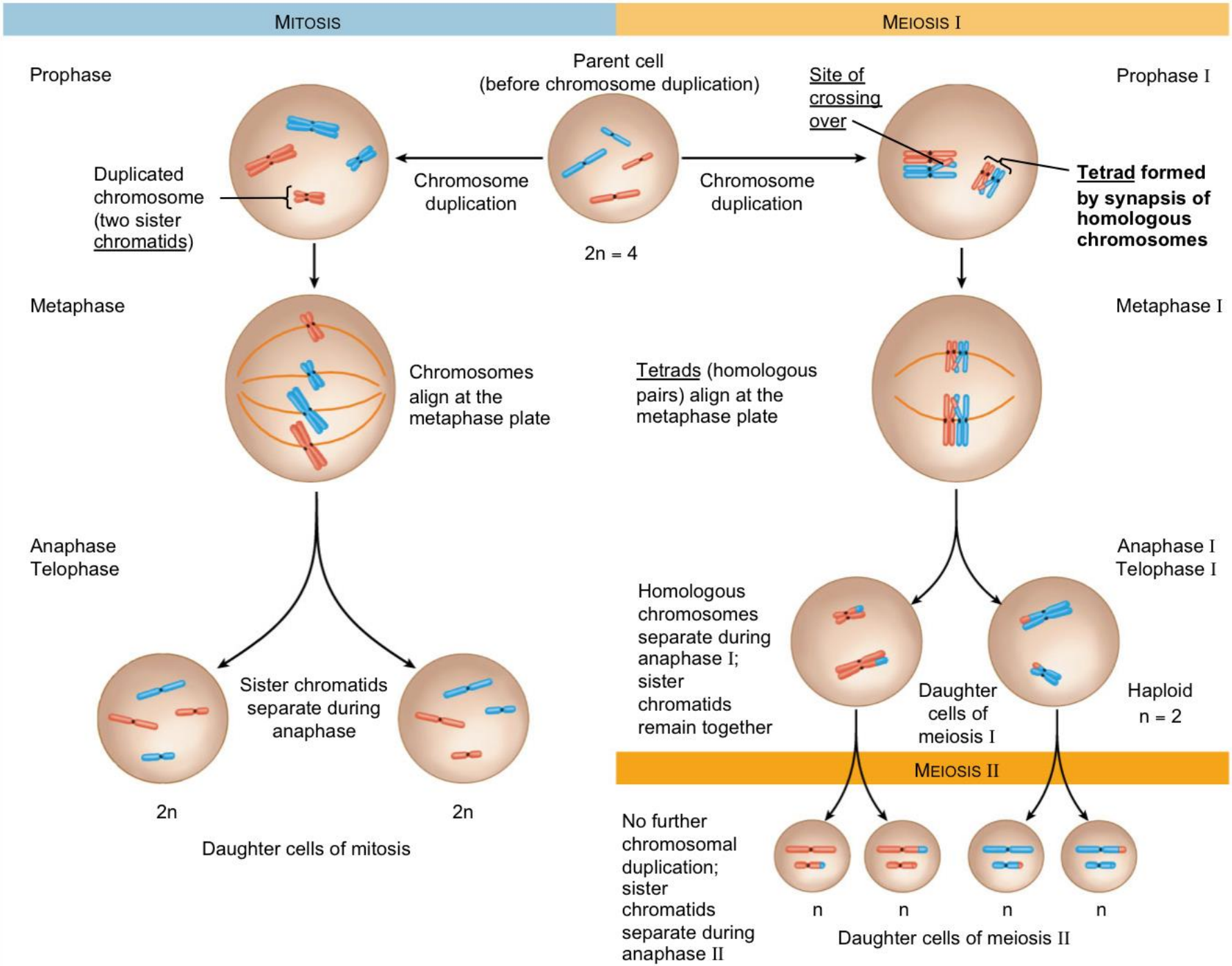
B) the same number of chromosomes and half the amount of DNA.

C) half the amount of cytoplasm and twice the amount of DNA.

→ D) half the number of chromosomes and one-fourth the amount of DNA.

After Meiosis, the daughter cells each have 1 copy of each chromosome as a single chromatid.

Whereas in prophase of meiosis 1, the cell has 2 copies of the chromosome and 4 chromatids.



4. What following statement is the most correct about Heterochromatin?

A) tightly packed DNA that facilitates transcription during cellular mitosis.

B) loosely packed DNA that allows for transcription during cellular interphase.

C) only occurs during mitosis and facilitates transcription.

→ D) areas of DNA that stain with Giemsa the most in karyotyping.

Remember Heterochromatin is tightly packed and stains darker in G-Band staining.
Where as Euchromatin is loosely packed and stains lightly (if at all) in G-Band Staining.
The more tightly packed = does not allow transcription machinery to have access = less transcription

5. What is a major difference between mitosis and meiosis 1 in a diploid organism?

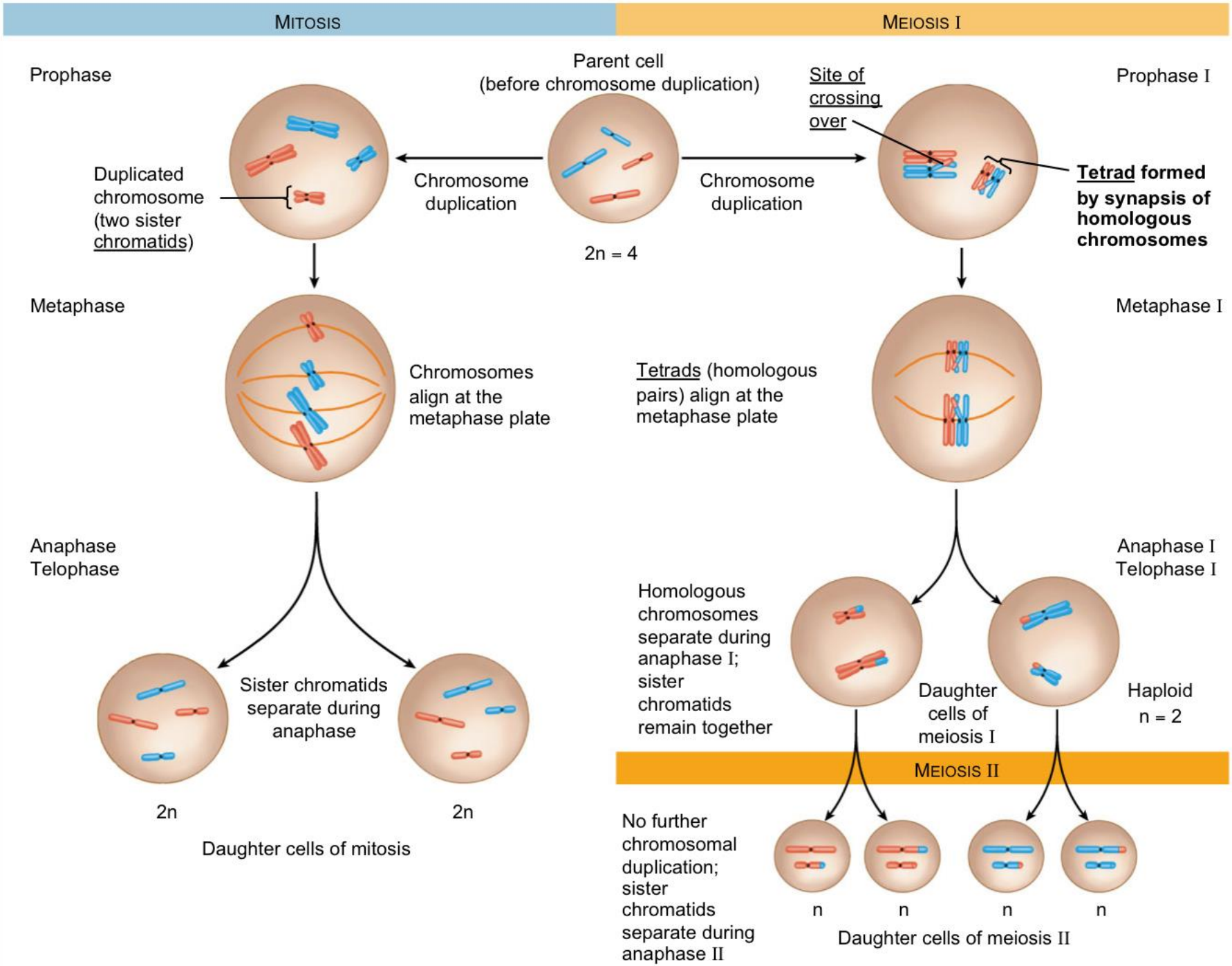
A) Sister chromatids separate in mitosis, while homologous pairs of chromosomes separate in meiosis I.

B) Sister chromatids separate in mitosis, while homologous pairs of chromosomes separate in meiosis II.

C) DNA replication takes place prior to mitosis, but not before meiosis I.

D) Only meiosis I results in daughter cells that contain identical genetic information.

Remember (see pic on next slide)



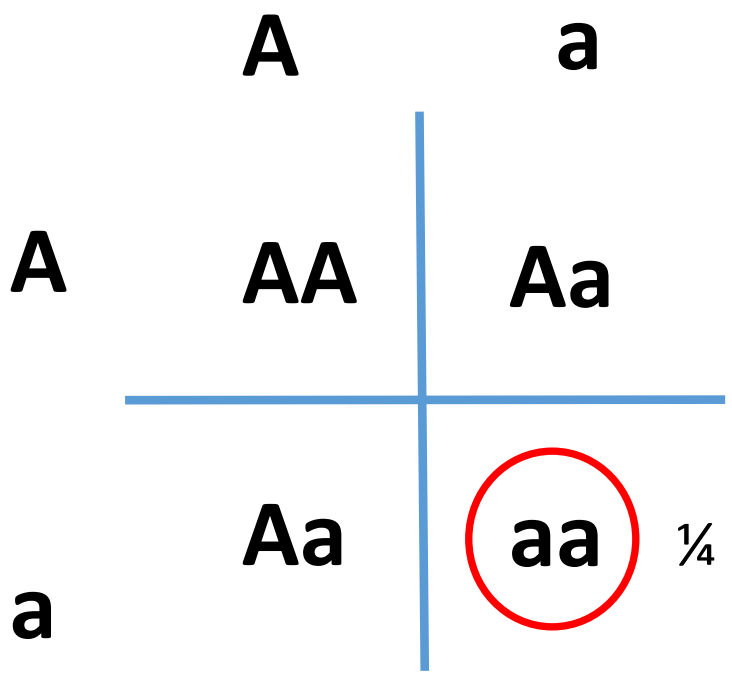
6. If two people who are both heterozygous for Huntington's disease (an autosomal dominant trait) marry, what is the probability that they will have five children, all of which are normal?

A) 0.001

B) 0.125

C) 0.237

D) 0.016



¼ Chance of a Normal Child

7. Gregor Mendel performed a dihybrid cross between two traits in the pea plant; seed colour and seed shape. Yellow (Y) was dominant to green (g) and round (R) was dominant to wrinkled (w). If Mendel crossed two plants that were both heterozygous for both traits (YgRw), what is the probability of getting the following all-dominant genotype 'YYRR'?

YgRw X YgRw

A) 1/2.

B) 1/4.

C) 1/8.

D) 1/16.

	YR	Yw	gR	gw
YR	YYRR	YYRw	YgRR	YgRw
Yw	YYRw	YYww	YgRw	Ygww
gR	YgRR	YgRw	ggRR	ggRw
gw	YgRw	Ygww	ggRw	ggww

8. Gregor Mendel performed a dihybrid cross between two traits in the pea plant; seed colour and seed shape. Yellow (Y) was dominant to green (g) and round (R) was dominant to wrinkled (w). If Mendel crossed two plants that were both heterozygous for both traits (YgRw), what is the probability of getting either the all-dominant genotype 'YYRR' **or** the all-recessive genotype 'ggww'?

YgRw X YgRw

A) 1/32.

B) 1/4.

C) 1/8. ←

D) 1/16.

	YR	Yw	gR	gw
YR	YYRR	YYRw	YgRR	YgRw
Yw	YYRw	YYww	YgRw	Ygww
gR	YgRR	YgRw	ggRR	ggRw
gw	YgRw	Ygww	ggRw	ggww

Mutually exclusive = what rule?

1/16 + 1/16 = 2/16 = 1/8

9. Independent assortment of chromosomes is a result of _____?

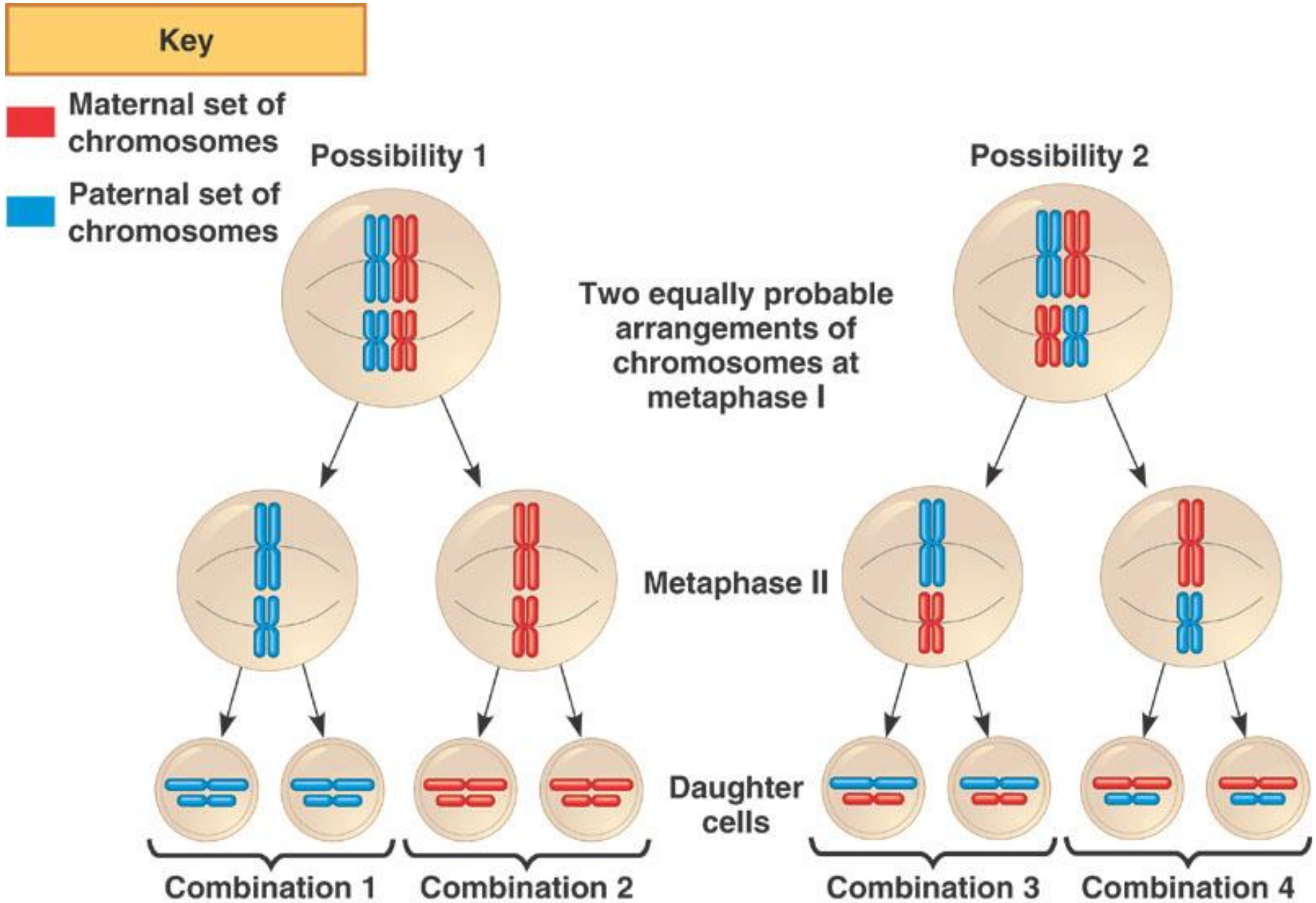
A) the random way each pair of homologous chromosomes lines up at the metaphase plate during meiosis I.

B) the random combinations of eggs and sperm during fertilisation.

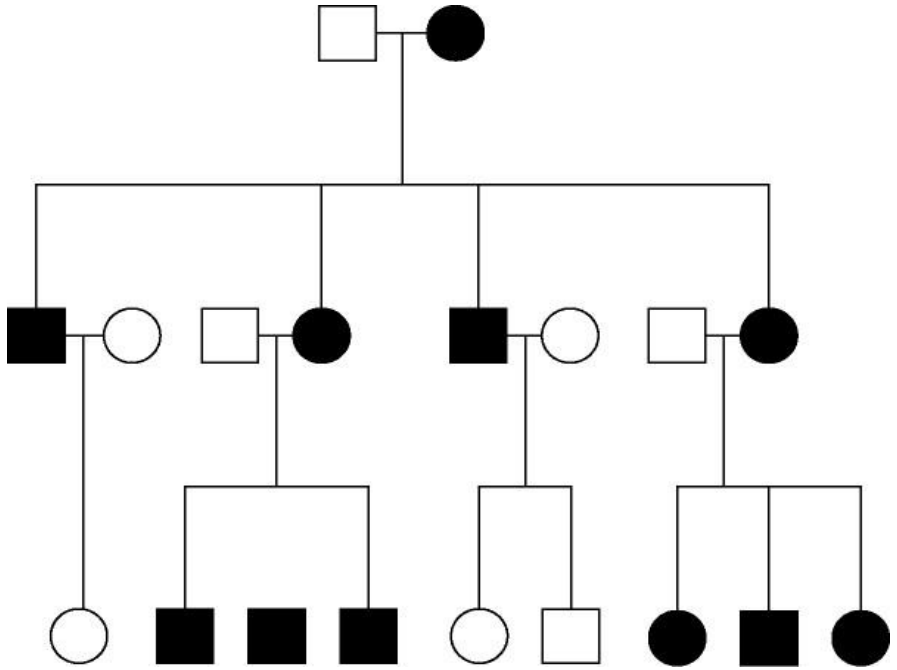
C) the random distribution of the sister chromatids to the two daughter cells during anaphase II.

D) the diverse combination of alleles that may be found within any given chromosome.

Remember (see pic on next slide)


Key
Maternal set of chromosomes
Paternal set of chromosomes
Possibility 1
Possibility 2
Two equally probable arrangements of chromosomes at metaphase I
Metaphase II
Daughter cells
Combination 1
Combination 2
Combination 3
Combination 4

10. The pedigree in the figure below shows the transmission of a trait in a particular family. Based on this pattern of transmission, the trait is most likely _____?



A) mitochondrial
B) sex-linked dominant
C) sex-linked recessive
D) autosomal dominant

Very easy to see that all children from an affected mother are also affected

11. The epistatic allele for the white colouration of squash is represented by a W. The hypostatic allele for green colouration is represented by a G. When squash has all recessive alleles for colouration (wwgg) they are yellow. What is the probability that when a green squash (wwGg) is crossed with a white squash (Wwgg) the offspring are green (wwGg or wwGG)?

A) 0.

B) 1/4.

C) 1/2.

D) 3/16

wwGg X Wwgg

	wG	wg	wG	wg
Wg	WwGg	Wwgg	WwGg	Wwgg
Wg	WwGg	Wwgg	WwGg	Wwgg
wg	wwGg	wwgg	wwGg	wwgg
wg	wwGg	wwgg	wwGg	wwgg

= 4/16
= 1/4

12. The Bombay genotype (mutant *FUT1* gene) prevents the synthesis of the H substance and consequently red blood cells cannot produce functional A and B antigens. If mum had the genotype I^AI^A and dad had I^BI^O, and dad has the Bombay phenotype, what blood groups do the parents have?

A) mum has O, while dad has B.
B) mum has A, while dad has O.
C) mum has A, while dad has B.
D) both parents have O.

Remember the Bombay phenotype doesn't allow either A or B antigens to be present, therefore if the dad has the Bombay phenotype he will present as O.

13. A red flower with the genotype (C^RC^R) crosses with a white flower with the genotype (C^WC^W) and produces a pink flower with the geneptype (C^RC^W). This exception to Mendelian genetics can be account for by _____?

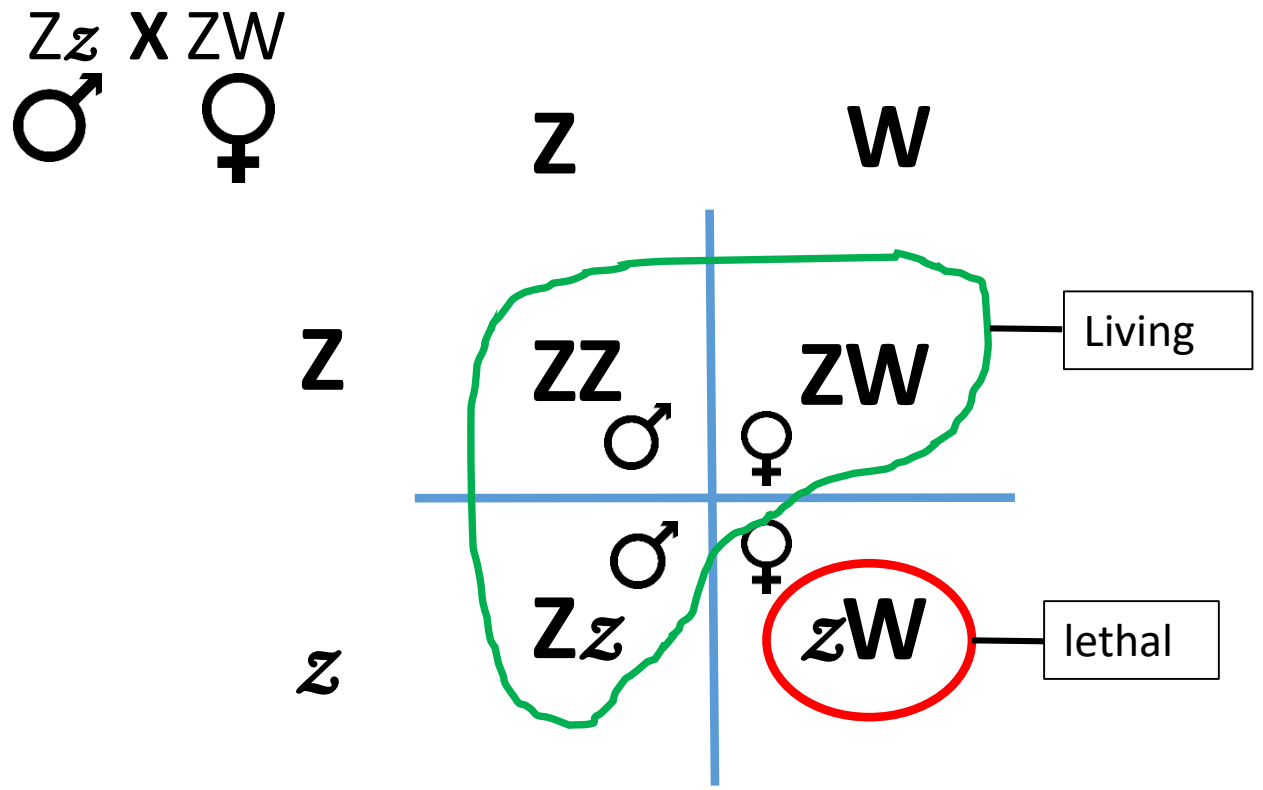
A) codominance.
B) gene linkage.
C) multiple alleles.
D) incomplete dominance. ←

Remember this is where both alleles blend their effects.

*refer to lecture 6

14. A recessive lethal allele that causes death of the embryo is sometimes present on the Z chromosome in pigeons. What would the sex ratio be in the living offspring of a cross between a male that is heterozygous for the lethal allele and a normal female?

A) 2:1 male to female ←
B) 1:2 male to female
C) 1:1 male to female
D) 3:1 male to female



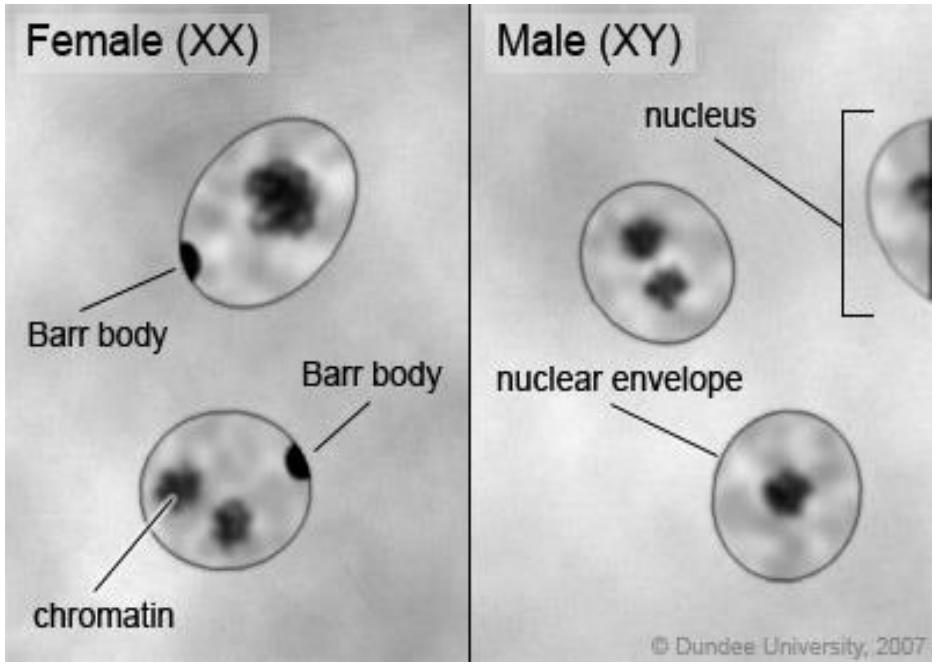
15. Complete the sentence: two or more genes located on the same chromosome that do not show independent assortment _____?

A) are in incomplete dominance and they are inherited together.
B) are called linked genes where they are not inherited together.
C) are called linked genes that tend to be inherited together.
D) are in incomplete dominance and they are not inherited together.

Remember linkage is where genes close together on the same chromosome and are inherited together which means you can treat them as a mono-hybrid cross
*Refer to Lecture 6

16. Human females have two X chromosomes in all somatic cells and regulate the expression of sex-linked genes through X-linked inactivation. Every X-chromosome that is inactivated forms a Barr body within the nucleus as shown in the figure below. If a Male has the following genotype with regards to his X-chromosomes (48,XXXY), how many Barr bodies form in each nucleus?

A) 3.
B) 2.
C) 1.
D) none, X-inactivation only occurs in females.



Remember only 1 X-chromosome is required and all others become Barr bodies

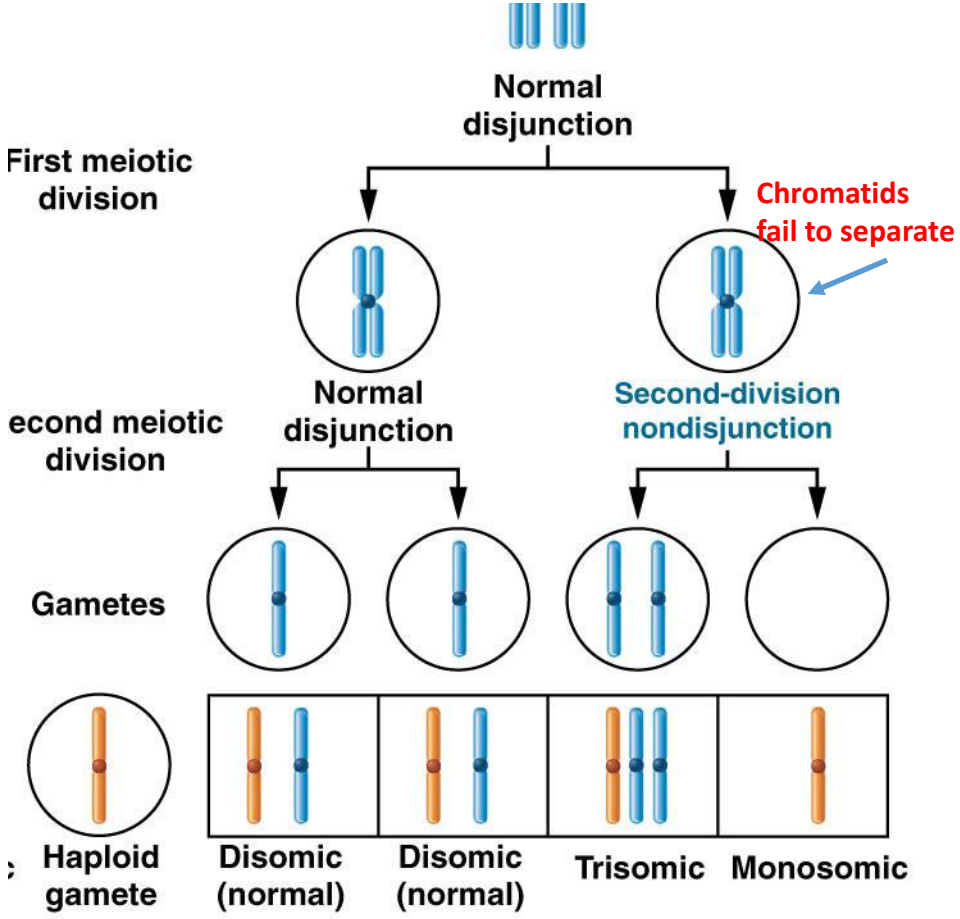
~~X~~ ~~X~~ X Y

These become Barr bodies

17. If cell X enters meiosis, and nondisjunction of one chromosome occurs in one of its daughter cells during meiosis II, what will be the result at the completion of meiosis?

A) All the gametes descended from cell X will be diploid.
B) Half of the gametes descended from cell X will be *n* + 1, and half will be *n* - 1.
C) 1/4 of the gametes descended from cell X will be *n* + 1, 1/4 will be *n* - 1, and 1/2 will be *n*.
D) Two of the four gametes descended from cell X will be haploid, and two will be diploid.

Non-disjunction at Meiosis



Fertilization with a haploid gamete- resulting zygotes

Figure 6.1

18. During meiosis, a defect occurs in a cell that results in the failure of microtubules (spindle fibres) to bind at the kinetochores, a protein structure on chromatids where the spindle fibres attach during cell division to pull sister chromatids apart. Which of the following is the most likely result of such a defect?

A) New microtubules with more effective binding capabilities to kinetochores will be synthesised to compensate for the defect.
B) Excessive cell divisions will occur, resulting in cancerous tumours and an increase in the chromosome numbers known as polyploidy.
C) The defect will be bypassed in order to ensure normal chromosome distribution in the new cells.
→ D) The resulting cells will not receive the correct number of chromosomes in the gametes, a condition known as aneuploidy.

19. Monosomy of a chromosome refers to _____?

A) A single chromosome breakage that results is a unique chromosome
B) A chromosomal number that is a multiple of the normal haploid chromosomal set
C) A condition in which one member of a chromosomal pair is missing; one less than the diploid number (2n – 1)
D) A condition in which one chromosome is present in three copies, and all others are diploid; one more than the diploid number (2n + 1)

Remember monosomy is when you are lacking a copy of a particular chromosome

20. Trisomy of a chromosome refers to _____?

A) A single chromosome breakage that results is a unique chromosome
B) A chromosomal number that is a multiple of the normal haploid chromosomal set
C) A condition in which one member of a chromosomal pair is missing; one less than the diploid number (2n – 1)
→ D) A condition in which one chromosome is present in three copies, and all others are diploid; one more than the diploid number (2n + 1)

Remember trisomy is when you have an extra copy of a particular chromosome.
*Do not confuse with triploid where there are 3 whole sets of chromosomes (3n).

21. Which of the following is not a purine

A) Adenine
B) Guanine
C) Thymine
D) None are purines

This is a pyrimidine!

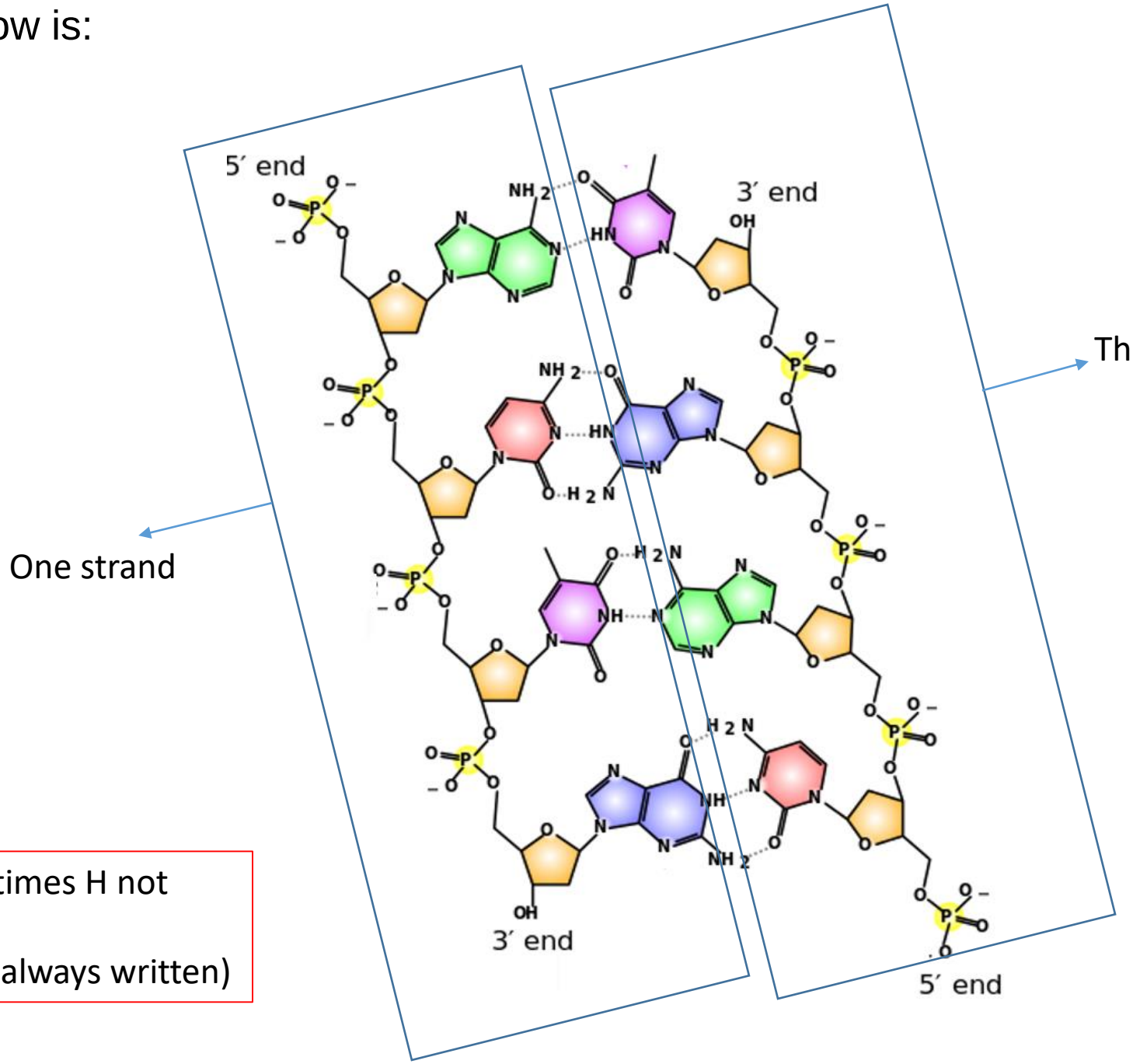
Remember the main pyrimidines in DNA are Thymine and Cytosine each have a ‘Y’ in the name and so does pyrimidine ☺

Adenine – Cytosine – Guanine – Thymine/Uracil

Purine	**Pyrimidine**	**Purine**	**Pyrimidine**
Double	**Single**	**Double**	**Single**

22. The polynucleotide below is:

A) Single stranded DNA
B) Double stranded RNA
→ C) Double stranded DNA
D) Single stranded RNA



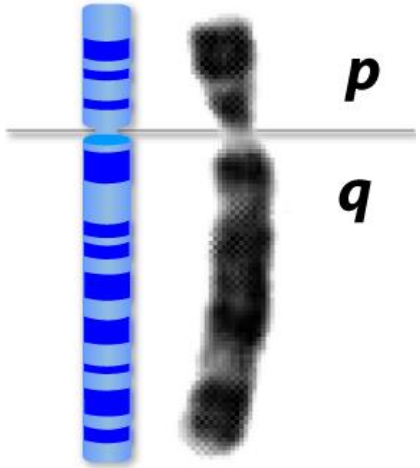
DNA has a ‘H’ at the (sometimes H not written)
RNA has an ‘OH’ (oxygen is always written)

23. The enzyme that releases the tension of supercoiling DNA is:

A) Polymerase
B) Topoisomerase
C) Helicase
D) Exonuclease

Remember this is the enzyme that puts ‘nicks’ in the DNA to release the tension.

24. The chromosome below is a(n) …….. chromosome:

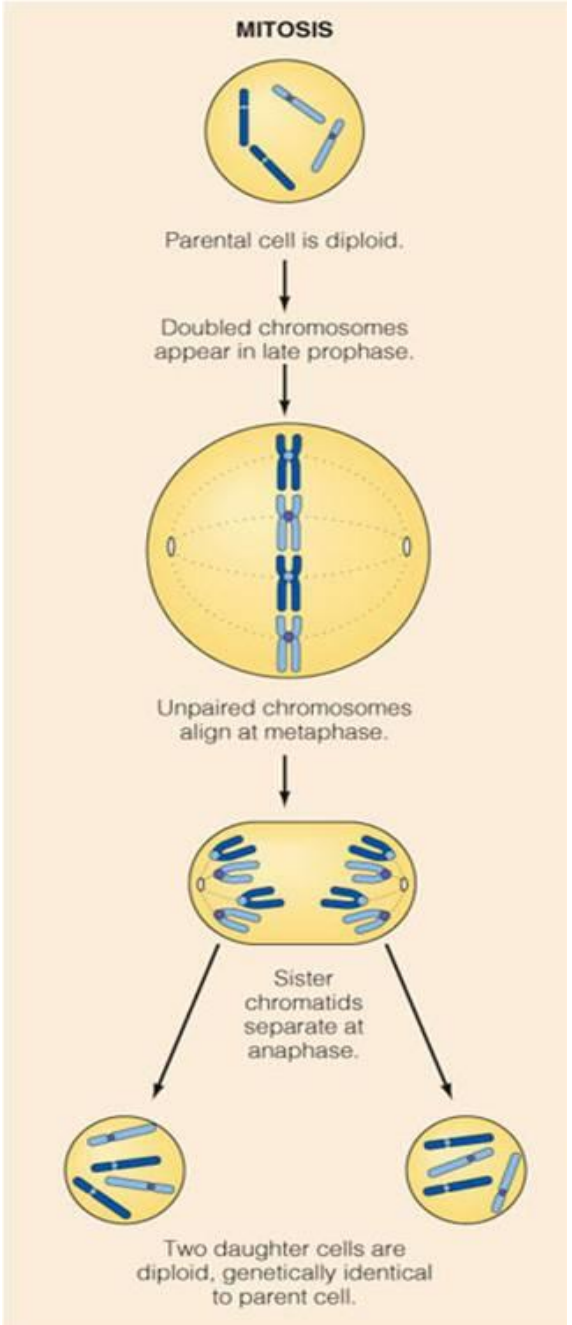


Chromosome 4

A) Acrocentric
B) Metacentric
C) Submetacentric
D) Telocentric

25. How many chromosomes and chromatids are present in the prophase of mitosis in a human somatic cell?

A) 46 chromosomes, 92 chromatids
B) 23 chromosomes, 46 chromatids
C) 92 chromosomes, 46 chromatids
D) 46 chromosomes, 23 chromatids



Phase (Mitosis)	# Chromosomes	# Chromatids
Prophase	46	92
Metaphase	46	92
Anaphase	92	92
Telophase	92	92
End of Mitosis (separated cells)	46	46
Phase (Meiosis I)	# Chromosomes	# Chromatids
Prophase I	46	92
Metaphase I	46	92
Anaphase I	46	92
Telophase I	46	92
End of Meiosis I (separated cells)	23	46
Phase (Meiosis II)	# Chromosomes	# Chromatids
Prophase II	23	46
Metaphase II	23	46
Anaphase II	46	46
Telophase II	46	46
End of Meiosis II (separated cells)	23	23

https://datbootcamp.com/biology-strategy/chromosome-and-chromatid-numbers-during-mitosis-and-meiosis/

26. The Karyotype 46,XY,del(13)(p9) means _____?

A) Female, 46 Chromosomes, deletion of short arm on chromosome 13 at region 9

B) Male, 46 Chromosomes, deletion of long arm on chromosome 13 at region 9

C) Female, 46 Chromosomes, deletion of long arm on chromosome 13 at region 9

D) Male, 46 Chromosomes, deletion of short arm on chromosome 13 at region 9

27. You have to complete a genetics exam but you didn't study at all (How could you? The lecturer makes it so boring!). The proposed exam consists of 20 questions - all multiple choice with 4 options (a, b, c, d). What is the probability that you can pass the genetics exam, score 10/20, by completely guessing every answer (like you are not even going to bother reading the questions)?

Step 1: Calculate the individual probabilities

- P(correct answer) = p = ¼ = 0.25
- P(incorrect answer) = q = ¾ = 0.75

Step 2: Determine the number of events

n = total number of questions = 20

x = number of correct scores = 10

n-x = number of incorrect scores = 10

Step 3: Substitute the values for p, q, x, and n in the binomial expansion equation

$$\frac{n!}{x!\,(n-x)!}\; p^{x} q^{(n-x)}$$

$$P = (20!/10!x10!)(0.25)^{10}(0.75)^{10}$$

$$P = 184756.\ x\ 5.37E^{-08}$$

$$P = 0.0099 = 0.99\%$$

0.99% chance to pass the test without studying!

$$\frac{2.43E^{+18}}{1.31E^{+13}} = 184756$$

$P = (20!/10!x10!)(0.25)^{10}x(0.75)^{10}$

$(0.25)^{10}(0.75)^{10} = 5.37E^{-08}$

$P = 184756 \times (0.25)^{10} \times (0.75)^{10}$

$184756 \times 5.37E^{-08} = 0.0099$

$P = 0.0099 = 0.99\%$

$$\frac{n!}{x!\,(n-x)!}\; p^x q^{(n-x)}$$

0.99% of chance that you will pass the test without studying!

Class test
Week 5, starts at 9AM Wed 7thth of August

- 30 MCA questions, material from week 1 - 4
- Complete online!
- You have 50 minutes from when you start. You have one attempt!
- Test closes Thursday 8t^{h} August 9am

Are you worried about the test? Don't panic, study hard and watch this to have a laugh and relax:

https://www.youtube.com/watch?v=hlvZ_8V6uc4

4.46min