

8. Interpret results from molecular analyses to determine the inheritance patterns and identities of human genes that can mutate.

Crime scene

- White blood cells have DNA in blood, also saliva has DNA and proteins
- ? Semen
- Version of X vs Y chromosome that codes for teeth enamel different

a. Interpret pedigree information to determine the suitability of a DNA marker for tracking a disease trait in a family.

The Family:
A kindred of 17 total: 13 with CAD, 9 of living had acute MI

Based on the pedigree alone what is the mode of inheritance?

A. Autosomal dominant
B. Autosomal recessive
C. X-linked dominant
D. X-linked recessive
E. More than one of the above are possible

ANS: More than one

b. Explain the method of SNP mapping and interpret SNP mapping data to pinpoint the chromosomal location of a human disease gene.

- Variant forms of DNA sequence (polymorphisms) are used to map gene locations
- Polymorphisms include single nucleotide polymorphisms and length polymorphisms
- Alleles of polymorphic sites show mendelian inheritance
- Alleles of polymorphic sites can be detected using methods including DNA hybridization, PCR and gel electrophoresis

Single Nucleotide polymorphisms

- In population, one nucleotide different
- Can be in the gene
- Can be detected using probes, northern/southern analysis, sequencing RFLP analysis
- May affect phenotype
- 1 in every 300-1000bp

SNP Genotype Identification

ASO for normal DNA sequence in region of $\Delta 508$ mutation in cystic fibrosis
5'-CACCAAAGATGATATTTTC-3'
Region deleted in $\Delta 508$

ASO for mutant DNA sequence in region around $\Delta 508$ deletion
5'-CACCAATGATATTTTC-3'

Diagnostics – Cystic Fibrosis

Normal ASO
 $\Delta 508$ ASO

Heterozygous Heterozygous CF Heterozygous normal

Probe: Detecting SNP

- Probe needs to attach to PERFECT MATCH. If not perfect match probe will fall off.
- Can make 4 different probes, each one with a nucleotide polymorphism
- Whichever probe binds is the correct probe.
- Intensity of dots indicated # of alleles

**Detecting the CF deletion allele with ASO
(it's autosomal recessive)**

1	2	3	4	5	6	
●	●	●	●		●	Normal ASO
●		●	●	●	●	Deletion ASO

A Individuals 1, 2, 3, 4, and 6 definitely do not have cystic fibrosis.

B Individual 5 is the only individual that can have cystic fibrosis.

C Individual 2 definitely has two copies of the wild type CFTR gene.

D all of the above

E none of the above

D, all of the above

Molecular Markers

5 conditions that characterize a suitable

- Must be polymorphic
- Codominance inheritance
 - Not expressing but one hide another
- Randomly and frequently distributed throughout the genome
 - Marker on every single chromosome and independently assorting
- Easy and cheap to detect
- Reproducible

Polymorphic markers

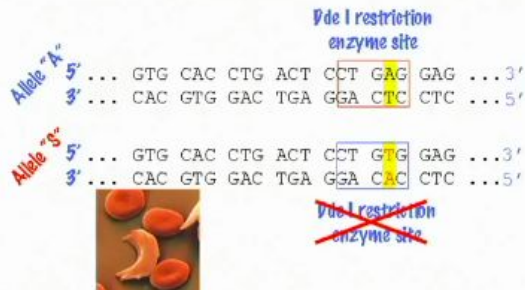
1. Single nucleotide Polymorphisms
 - Small stretch of DNA up to 10 nucleotides long
 - In population, one nucleotide different
 - Can detect with probes or if a site that has a restriction enzyme can cut it there
2. Insertion/Deletions (Indels)
 - Locus longer or shorter depending on insertion / deletion
 - Can be detected with probes
3. Variable Number of Tandem Repeats
 - Can have a lot of repeats or little → making locus longer or shorter
4. Restriction Fragment Length Polymorphisms (RFLP's)
 - Cut DNA with enzyme and profile of the cut (unique to an individual)
 - Hemoglobin

- i. normal = there is restriction site, can cut = 2 fragments
- ii. Mutation = no restriction site, will not cut = 1 fragment.

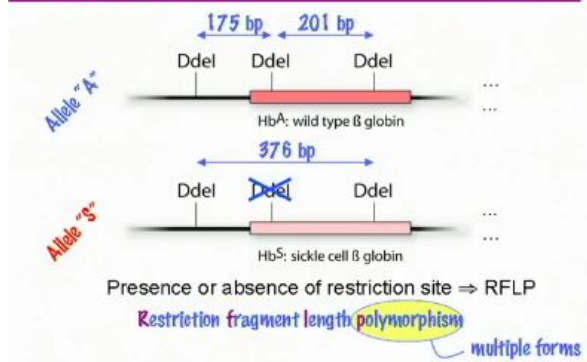
Restriction Fragment Length Polymorphisms (RFLPs)

• Differences in DNA fragment lengths after cutting with one or more restriction endonucleases *Dde*I 5'...CTNAG...3' 3'...GANTC...5'

• An example from hemoglobin B



Identifying Hb Genotype

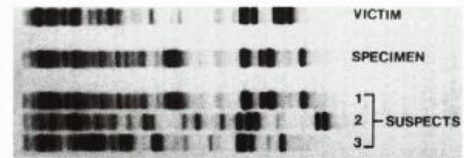


- After restriction digest can
 1. Nothing, just count the number of fragments
 2. hybridization with a labeled probe (southern blot)
 - a. Detect a specific band in that series
 3. PCR
 - a. Amplify specific regions of the segments
 - b. Can detect specific regions on southern blot

RFLP and Nucleic acid blotting

Restriction Fragment Length Polymorphism (RFLP analysis)

- Restriction digests (alleles = specific digest pattern)
- Presence/absence of restriction sites generates fragments of variable lengths of DNA fragments
- Hybridize with probes (Southern analysis)
- alleles (SNPs, VNTRs, STRs, genes) have unique patterns

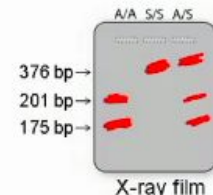
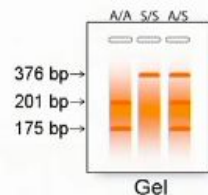
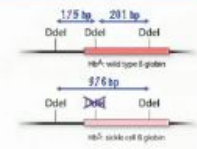


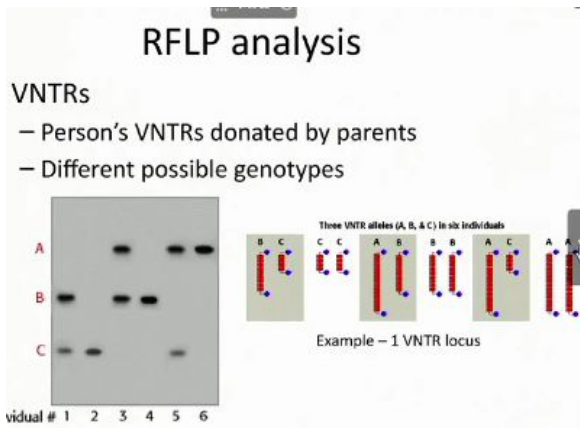
- Through probe for STR etc, can see profiles for the condition
- Alleles SNP, VNTR STR have unique patterns

- 2 thick bands = homozygous wild type
 - 2 copies of the homozygous gene and none cut. Thus thick
- 1 thick fragment = homozygous sickle cell
- One thick and 2 thinner = heterozygous
- Know how meiosis goes so can guess bands someone is going to inherit or can

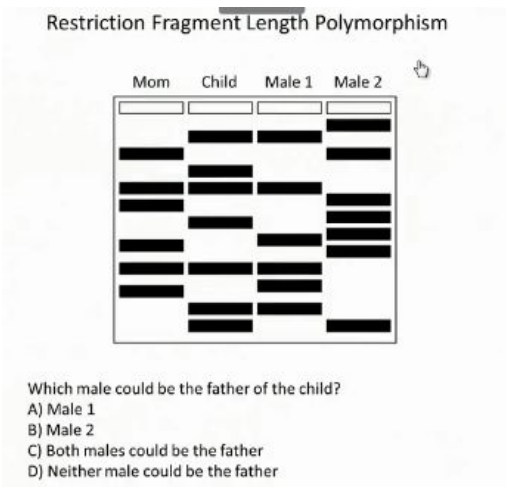
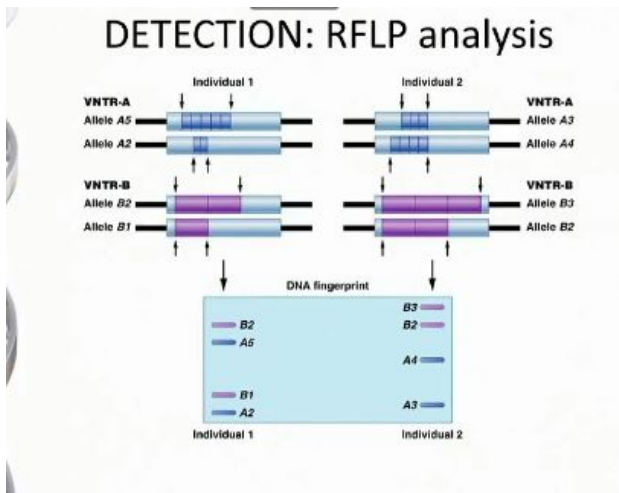
Identifying Hb Genotype by a Southern Blot

1. Digest human DNA sample with *Dde*I
2. Run gel
3. Blot to filter, hybridize with probe
4. Wash off excess probe, expose film



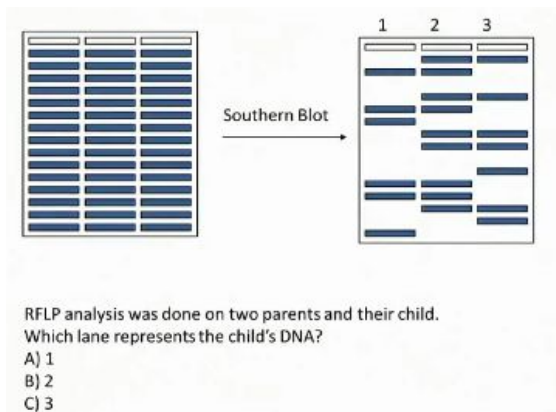


- Multiple different markers
- Use 2 different enzymes to cut DNA then 2 codes to light them up



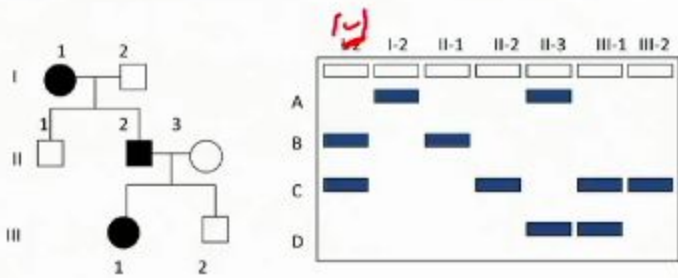
ANS: D, Neither

- must inherit something from mom and something from dad Male 1 could give some stuff but male 2 could give some stuff. They all cannot be the father.
- the child has band 2 but mom and both males do not. Must come from a different male.



ANS: Lane 2 ! Only lane whereby only something in common with one of the parents.

What is the mode of inheritance?



- A autosomal dominant 19
- B autosomal recessive 32
- C X-linked dominant 45
- D X-linked recessive 12

- Based on the pedigree can be anything
- Based on the blott, only males have one band and females have 2
- If you are female and have 1 C you are afflicted so dominant and X linked
- ? But, for III-2 how can you have the C allele but be normal??
 - We need this marker to be unlinked from disease
 - If marker went from X to Y chromosome so now when dad gave the y to his kid the marker was there not the allele causing the gene
 - In mom could have has another recombinant effect so the kid would not get it.

Suppose that MEF2A gene was found to be 5 cM away from marker D15S120. If you were to perform lineage analysis within you own family,

- A The MEF2A-D15S120 distance should also be 5 cM in your family.
- B The MEF2A-D15S120 distance should be at a distance other than 5 cM in your family.

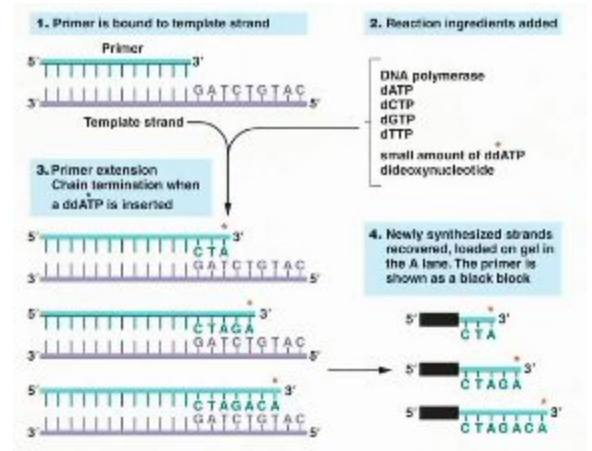
YES, that's how we can predict outcomes of crosses because consistent throughout species.

c. Explain how a gene that mutates to cause a disease can be molecularly identified.

- Using restriction endonucleases to check for differences in sequences
- Or probes which bind to very specific sequences.

DNA analysis - DNA sequencing

- Most common method of DNA sequencing is dideoxy chain termination sequencing developed by Sanger
- Template = any DNA fragment from PCR, plasmid with insert ETC
- Like a PCR reaction but with one primer. Primer bound to template strand and when ddATP added, stops the strand from continuing making fragments
- Everytime a fragment ends, ends with a ddATP or ddCTP etc
- Primer 5' to 3'
- Bottom of gel 5' to 3'



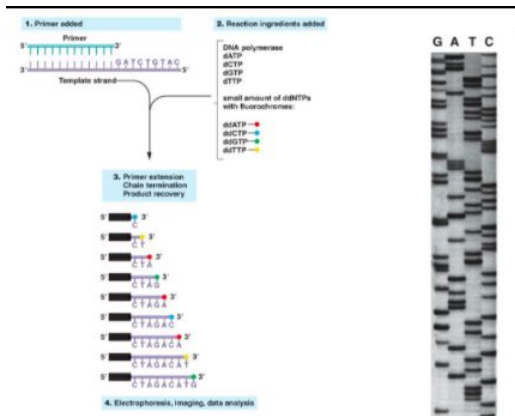
DNA analysis – DNA sequencing

What is the sequence of the first 15 nucleotides of the DNA displayed on the gel?

- 1) 3' CAGACGCTGTCACTG 5'
- 2) 5' CAGACGCTGTCACTG 3'
- 3) 3' CGCTTTCATGTCAGC 5'
- 4) 5' CGCTTTCATGTCAGC 3'



ANS 4.



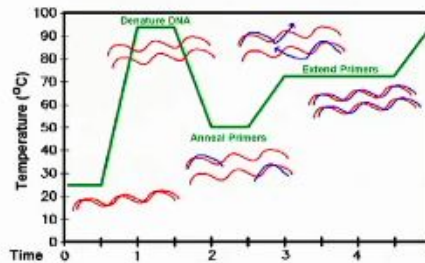
- Can add fluorescence to ddNTP's.
- Flash gel through laser and can get to approx 3000 bp of

Microsatellite/ VNTR Genotyping

- PCR, polymerase chain reaction
- Measure size of DNA you have made by gel electrophoresis
 - The brighter the band the > DNA there
- Makes DNA copies without host cells

- The PCR copies specific sequences through in vitro reactions that can amplify target DNA sequences present in very small quantities.
- PCR requires 2 oligonucleotide primers, one complementary to the 3' end of the DNA to be amplified and one complementary to the 3' end of the other strand
- No longer need probes: amplify genes directly from genomic DNA
- Need
 - Two primers
 - Excess nucleotided dATP, dTTP, dGTP, dCTP
 - Taq polymerase
 - Buffer with MgCl₂
 - Thermocycler

Polymerase Chain Reactions (PCRs)



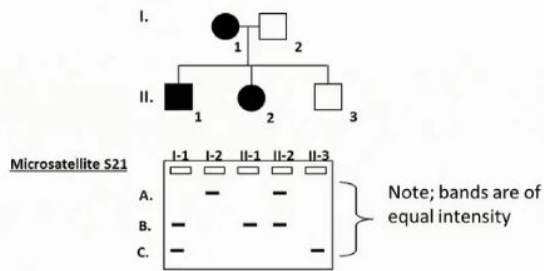
- 1 - Denaturation at 94°C
- 2 - Primer annealing at 37°C to 65°C
- 3 - Extension at 72°C

5' - CCCTGGGCTCTGTAAATGTTTCTAAGTG - 3'
 3' - GGGACCCGAGACATTTACAAAGATTCAC - 5'

- A 3'-ACTGTTAGA-5'
- B 3'-AAATTGGC-5'
- C 3'-ATGCTT1GA-5'
- D 5'-GGGACCCGA-3'
- E 5'-CCCTGGGCT-3'

ansE.

Considering both the pedigree and the microsatellite data, what is the mode of inheritance for CMT?



A	Autosomal dominant	46
B	Autosomal recessive	11
C	X-linked dominant	68
D	X-linked recessive	0
E	More than one of the above is possible	23

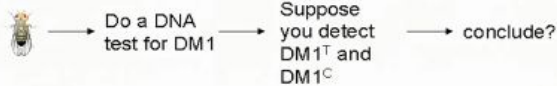
C. If female has 2 bands and male has one likely X lined.

d. Interpret bioinformatics data to compare homologous genes in different species and infer relative degrees of evolutionary relatedness.

DNA polymorphisms are Genomic Landmarks

- Mile markers throughout the genome
- If we can show our trait is linked to a DNA polymorphism, you know where the gene is located
- You do test cross to link one of the polymorphisms with a gene

DNA Marker Genotypes



Conclude:

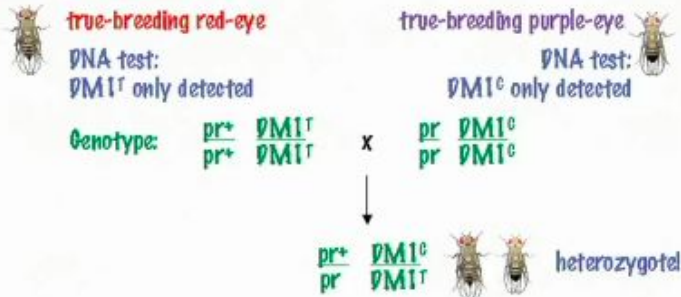
One homologue had $DM1^T$ allele, one homologue had $DM1^C$ allele...

this fly is **heterozygous** for this DNA marker



Testing for linkage

Step 1. Generate the heterozygous flies.



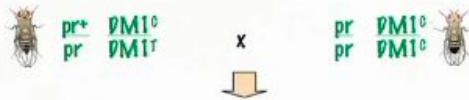
When the heterozygote makes gametes... what would you consider the parental types among these gametes?

$pr^+ DM1^T$ and $pr DM1^C$

Recombinants → Pr+Dm1c and PrDM1T

Testing for linkage (cont'd)

Step 2. Do a testcross.



Step 3. Score the progeny—

For each progeny fly: what eye color?
which allele(s) at DM1?

Sample results...

gamete?	P/NP?	phenotype:	# of progeny
$pr^+ DM1^T$	P	red, DM1 ^T & DM1 ^C	322
$pr DM1^C$	P	purple, DM1 ^C & DM1 ^C	318
$pr^+ DM1^C$	NP	red, DM1 ^C & DM1 ^C	78
$pr DM1^T$	NP	purple, DM1 ^T & DM1 ^C	82

↑
progeny genotype?

Testing for linkage (cont'd)

Step 4. Interpret the results.

Conclusion? The eye color gene is linked to the DM1 locus

$$\text{Map distance} = \frac{78 + 82}{322 + 318 + 78 + 82} = 20 \text{ cM}$$



9. Apply the results from molecular genetics studies in model organisms to understand aspects of human genetics and genetic diseases.

1) Describe molecular genetic approaches used for studying genes, gene functions (knock-outs and gene targeting technologies), and genomes to determine inheritance patterns and identities of genes that can mutate.

2) Describe the benefits and limitations of using model organisms to study human genes and human genetic diseases.

Benefits:

- cheap and easy to maintain
- Smaller in size
- Much easier to understand human genes and genetic diseases because they breed at a faster rate compared to humans
- Can use forward and reverse genetics

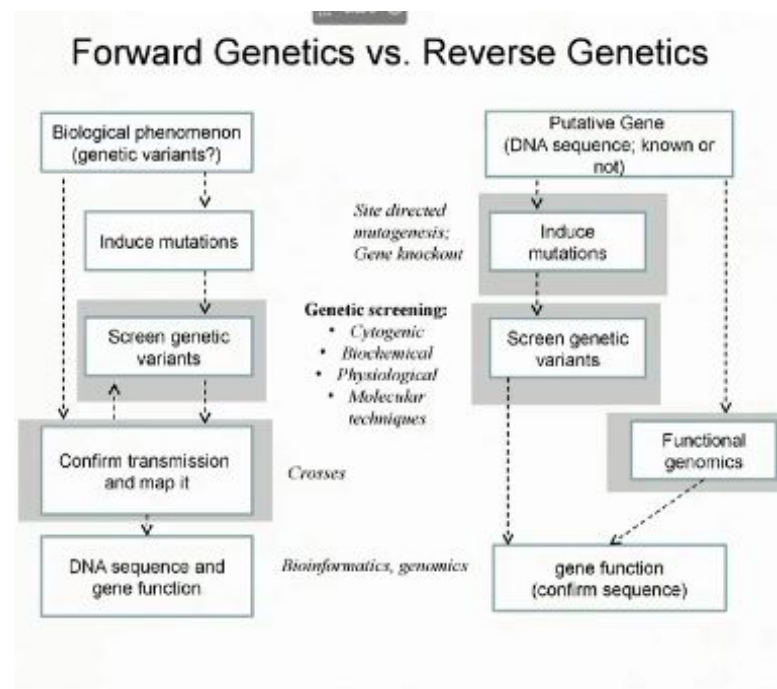
Limitations:

- Different physiological systems than humans
-

3) Explain why information on functions of human genes can often be acquired through studies of simple organisms such as yeast, nematode worms, and fruit flies

- Genes can be inserted into yeasts / fruit flies via plasmids using restriction endonucleases
- These expression of these genes can be induced in these small molecules and the proteins created can be studied in control environments.

- Have sequence and through some procedures can create mutations within the sequence. Then can put it into an organism and see how you disrupted the function. If you disrupt the function you can figure out the function.
- Mouse and human protein for FOXP2 differ by only 3 AA and folded up the same way / acted the same way. How would using the human FOXP2 gene affect development?
- **Reverse Genetic Analysis.** Have the DNA sequence but unsure exactly what mutation it does. They concluded that affect jaws in mouse so likely have something to do with jaw formation / speech.



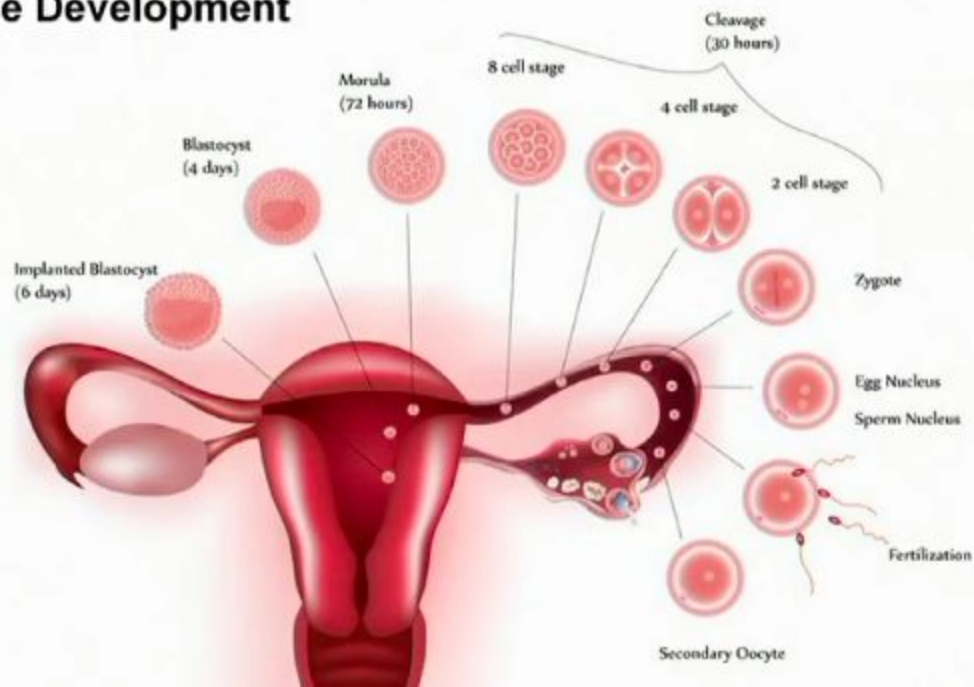
Based on available information about FOXP2 protein, which one of the following statements makes the most sense?

- A If its FoxP2 gene is removed, the mouse might "talk." 7
- B Feeding mice the three amino acids that differ between human and mouse might enable the mice to "talk." 10
- C Replacing the mutated human FOXP2 gene with a mouse FoxP2 gene is a way to cure speech disorders. 90
- D Putting the human version of FOXP2 gene into a mouse might enable it to "talk." 27

If mouse wild type protein works the same, so if you replace the mutated human version with that of a mouse might behave regularly.

- Eggs arrested in meiosis 1. Once fertilized finishes meiosis II and fuses

Mouse Development



- Take a transgene solution, inject something inside the nucleus, the nuclei will fuse then implant cell into pseudopregnant female.
- Pseudopregnant female → cross with a sterile male. Mouse thinks its pregnant so prepares hormonally but inject the fertilized egg into mouse.

How to make a humanized FOXP2 transgenic mouse?

Human FOXP2 DNA in plasmid → mouse zygote → transgenic mouse

Group Activity 2: Draw a flowchart using *some* of the following items (1-7) to illustrate how you could make a “humanized” FOXP2 transgenic mouse and label where FOXP2 is going to be at different developmental stages (from the zygote stage to the baby mouse stage.)

1. Pseudopregnant female mouse
2. Mouse blastocyst
3. Mouse zygote cell
4. Human *FOXP2* DNA
5. Human *FOXP2* RNA
6. Human FOXP2 protein
7. Mouse *FoxP2* DNA

4,3,1.

Below are some alternative methods for making a “humanized” FOXP2 mouse that has the transgene in every single cell. Which one would work best?

Below are some alternative methods for making a “humanized” FOXP2 mouse that has the transgene in every single cell. Which one would work best?

- A. Inject the human FOXP2 **PROTEIN** into both cells at the **2-cell stage**.
- B. Inject the human *FOXP2* **RNA** into the zygote.
- C. Inject the human *FOXP2* **DNA** into a **fertilized egg**.
- D. Inject the human *FOXP2* **RNA** into the **blastocyst cavity**.

A	Inject the human FOXP2 PROTEIN into both cells at the 2-cell stage.	14
B	Inject the human FOXP2 RNA into the zygote.	26
C	Inject the human FOXP2 DNA into a fertilized egg.	82
D	Inject the human FOXP2 RNA into the blastocyst cavity.	13

- We want every cell to have DNA, so if DNA crosses over into cell in exchange for mouse FOXP2 just like in meiosis
 - Proteins and RNA, can only be expressed for little time and may not even be used during these stages of the development
 - If injected in blastocyst, have to go through very many divisions and DNA must survive all the divisions
 - If you go early in development where the nuclei has not fused yet then best chance (zygote)

Recombinant DNA Technology

- DNA transport → Plasmid
- Replicate independently of the chromosomal DNA

- Carry antibiotic resistance genes (genetic marker, can select with)
- Polylinker region
 - Full of restriction sites allow insertion of DNA fragments
- Easily manipulated.

How to use recombinant RNA technology ?

1. Amplify your DNA of interest using PCR.
 2. Digest DNA of interest by restriction enzymes.
 - a. Cut double stranded DNA at specific nucleotide sequence, produce sticky ends
 3. Ligate of DNA.
 - a. Dna molecule with compatible ends can be joined together by DNA ligase
 4. Transformation and Selection
 - a. Propagation of recombinant dna
 - b. Only bacteria containing recombinant DNA grow on medium + Abx
 - c. Ones that don't grow didn't get the plasmid.
 - d. How to verify if they actually have your inserted gene?
 5. Extract plasmid and purify
 - a. Can do PCR on it, amplify region and see if it's there
 - b. Can try to add restriction enzymes to take it out (digest it and see if its there)
- Now to insert it into an organism, isolate plasmid with the insert and make a pure solution of that
 - Inject the solution into a fertilized cell before the 2 nuclei merge together. Inject inside female nucleus. Hope that DNA fragment crosses over into nucleus in the right spot. Will take ++ years.
 - Put that in pseudopregnant female
 - You do it right before it divides so every new cell has the gene
 - You make sure you get a heterozygous individual because u ensured the gene was put into only the female, if you added it in after could end up in both.
 - Identify those who have the mutation by screening the genome
 - To make a homozygous line, cross the mice together or back crosses.

10. Describe the processes that can affect the frequency of phenotypes in a population over time

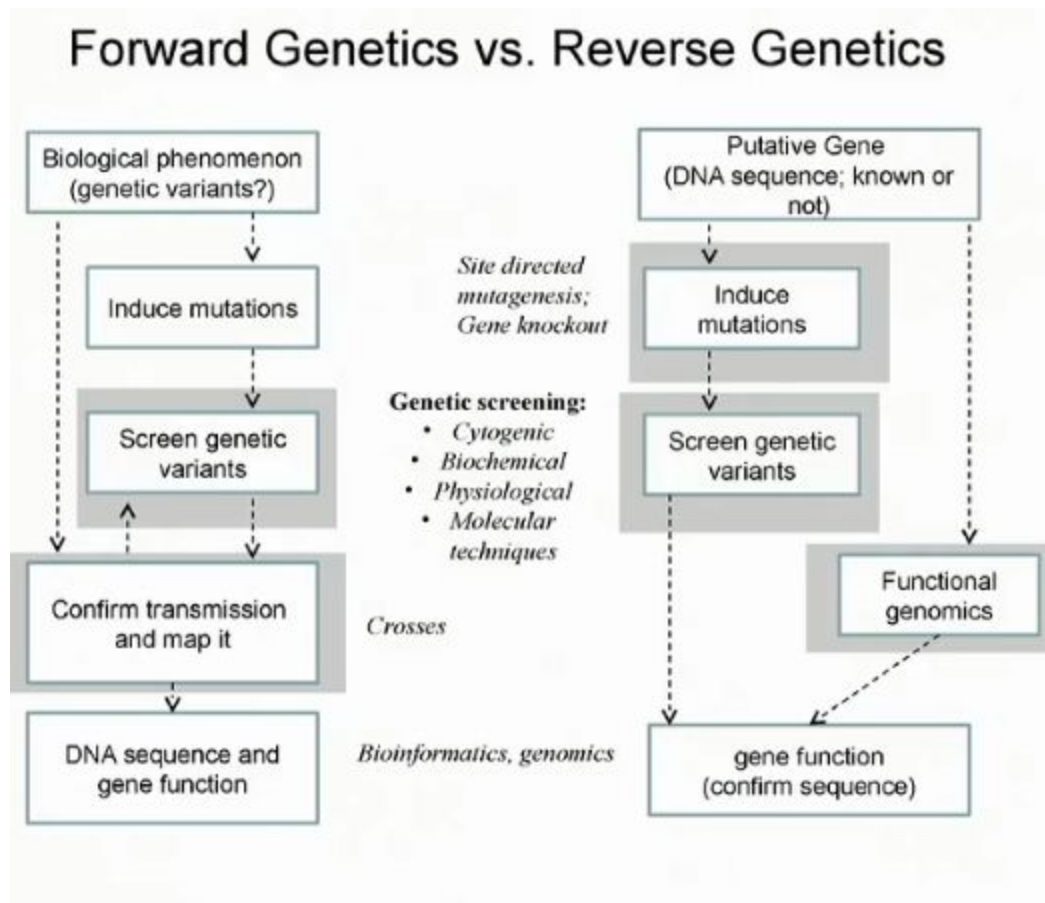
1. Explain how natural selection and genetic drift can affect the elimination or maintenance of alleles in a population

In natural selection, those variations in the genotype that increase an organism's chance of survival will be favoured. Natural selection decreases the frequency of particular genes that decrease fitness and increases the frequency of genes that increase fitness. Genetic drift is the change in frequency of existing alleles in a population. Genetic drift continues until the allele is either lost in the population or until it's the only allele present in the population. This occurs in small populations where the alleles face a greater chance of being lost.

2. Determine allele frequencies based on phenotypic data for a population in equilibrium.
3. Interpret experiments to determine the relative influences of genes and the environment on a given phenotype
4. Use population genetic concepts to interpret DNA forensic analysis from crime scenes.

11. Evaluate the social impacts of genetics in the clinical setting.

1. Name and describe the different types of genetic tests and the purpose for conducting them.



2. Describe the types of results the various types of genetic tests provide

- Newborn screening
- Newborn screening is used just after birth to identify genetic disorders that can be treated early in life. Millions of babies are tested each year in the United States. All states currently test infants for [phenylketonuria](#) (a genetic disorder that causes intellectual disability if left untreated) and [congenital hypothyroidism](#) (a disorder of the thyroid gland). Most states also test for other genetic disorders.

- Diagnostic testing
- Diagnostic testing is used to identify or rule out a specific genetic or chromosomal condition. In many cases, genetic testing is used to confirm a diagnosis when a particular condition is suspected based on physical signs and symptoms. Diagnostic testing can be performed before birth or at any time during a person's life, but is not available for all genes or all genetic conditions. The results of a diagnostic test can influence a person's choices about health care and the management of the disorder.
- Carrier testing
- Carrier testing is used to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder. This type of testing is offered to individuals who have a family history of a genetic disorder and to people in certain ethnic groups with an increased risk of specific genetic conditions. If both parents are tested, the test can provide information about a couple's risk of having a child with a genetic condition.
- Prenatal testing
- Prenatal testing is used to detect changes in a fetus's genes or chromosomes before birth. This type of testing is offered during pregnancy if there is an increased risk that the baby will have a genetic or chromosomal disorder. In some cases, prenatal testing can lessen a couple's uncertainty or help them make decisions about a pregnancy. It cannot identify all possible inherited disorders and birth defects, however.
- Preimplantation testing
- Preimplantation testing, also called preimplantation genetic diagnosis (PGD), is a specialized technique that can reduce the risk of having a child with a particular genetic or chromosomal disorder. It is used to detect genetic changes in embryos that were created using assisted reproductive techniques such as in-vitro fertilization. In-vitro fertilization involves removing egg cells from a woman's ovaries and fertilizing them with sperm cells outside the body. To perform preimplantation testing, a small number of cells are taken from these embryos and tested for certain genetic changes. Only embryos without these changes are implanted in the uterus to initiate a pregnancy.
- Predictive and presymptomatic testing
- Predictive and presymptomatic types of testing are used to detect gene mutations associated with disorders that appear after birth, often later in life. These tests can be

helpful to people who have a family member with a genetic disorder, but who have no features of the disorder themselves at the time of testing. Predictive testing can identify mutations that increase a person's risk of developing disorders with a genetic basis, such as certain types of cancer. Presymptomatic testing can determine whether a person will develop a genetic disorder, such as [hereditary hemochromatosis](#) (an iron overload disorder), before any signs or symptoms appear. The results of predictive and presymptomatic testing can provide information about a person's risk of developing a specific disorder and help with making decisions about medical care.

- Forensic testing
- Forensic testing uses DNA sequences to identify an individual for legal purposes. Unlike the tests described above, forensic testing is not used to detect gene mutations associated with disease. This type of testing can identify crime or catastrophe victims, rule out or implicate a crime suspect, or establish biological relationships between people (for example, paternity).

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3. Discuss the benefits of genetic testing.

- Can predict impact of allele on functions.
- Testing for gene mutations

4. Identify the risks and limitations of genetic testing

- Genetic testing can provide only limited information about an inherited condition. The test often can't determine if a person will show symptoms of a disorder, how severe the symptoms will be, or whether the disorder will progress over time. Another major limitation is the lack of treatment strategies for many genetic disorders once they are diagnosed.

5. Discuss how genetic testing in a research setting differs from clinical genetic testing.

- The main difference between genetic testing in research and in clinical genetic testing is the purpose of the test and who receives the tests.
- Genetic testing: The goal of genetic testing in a research setting would be to identify unknown genes, learning how different genes work, and understanding the genetic conditions. The results in a research setting are not available to patients or healthcare providers.
- Clinical testing: This is done to find out information of the inherited disorder. Patients receive test results and can use them to help them make decisions.

Lecture Questions

Yellow pea color (Y) is dominant to green pea color (y). Round seed shape (R) is dominant to oval pea shape (r). Let's say the genes responsible for both of these phenotypes are on the same chromosome. Which of the following genotypes should result in a yellow pea plant with round seeds?

A. YyRr

B.

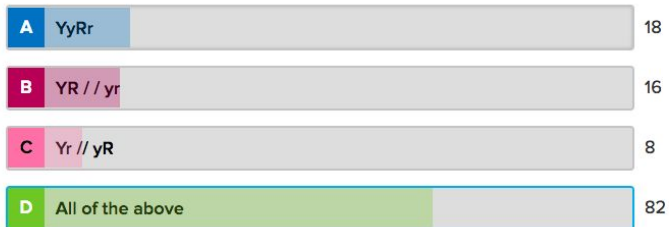
Y _____ R

C. y _____ r

Y _____ r

y _____ R
D. All of the above

D. All of the above




B and C = haplotypes

The arrangement of the alleles with respect to each other.

REVIEW CONCEPTS – GROUP ACTIVITY

What is the phenotype ratio of progeny in the following cross if the R and Y gene are very close to each other on the same chromosome?


Round, yellow



RrYy

X

Wrinkled, green



rryy

R _____ Y

r _____ y

ANS

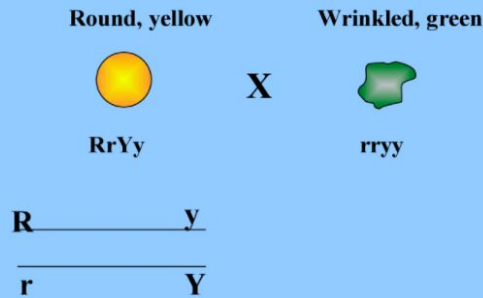
1:1

1 RY to 1ry (1 round yellow to 1 wrinkled green)

If there were crossing over we would expect a mix of alleles but since they are so close together we only get RY and ry

REVIEW CONCEPTS – GROUP ACTIVITY

What is the phenotype ratio of progeny in the following cross if the R and Y gene are **very close** to each other on the same chromosome?

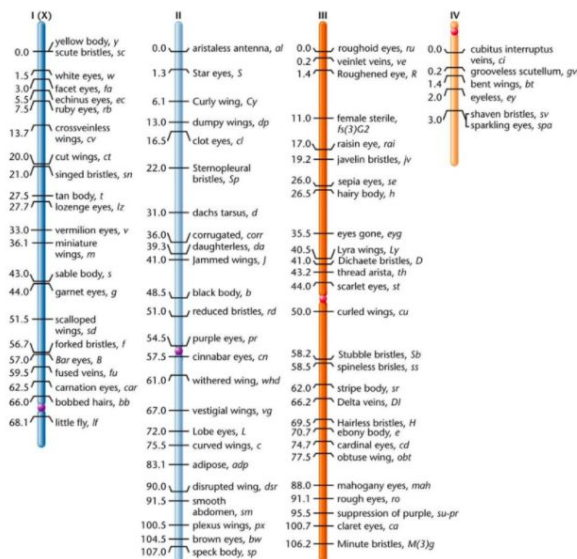


The haplotype can have a huge impact on the phenotypic ratio.

Ans 1:1 Ry to rY (1 Round green to 1 wrinkled yellow)

Whole point is if you have a new gene and don't know where it belongs in the genome, do crosses to link it to something else. This linkage with something known will allow you to know where it is.

Linked vs Unlinked Genes - Crosses



Map units called centimorgan and are the possibilities of crossing over in this region.

- When the 2 genes very close together less likely of crossing over. The closer together, the more likely. Final

Last 2 classes are the recombinants

Chromatids on the extreme end DO NOT have a recombination but the ones in the middle do have a recombination

Have to consider 3 situations

1. No linkage, independent assortment

Parental generation → AABB x aabb

Test Cross → AaBb x aabb

From AaBb get (AB, Ab, aB, ab) and from aabb get (ab)

Gametes : $\frac{1}{4}$ ABab

$\frac{1}{4}$ Aabb

$\frac{1}{4}$ aaBb

$\frac{1}{4}$ aabb

If we get 1:1:1:1 ratio → probably NOT linked

What is the benefit of doing a test cross?

- The phenotypic ratio reflects the types of gametes made in their proportion as well. No way to measure gametes directly.
- Parentals have a phenotype that looks like the parents crossed.
- Recombinants have a phenotype not seen in the parent generation
 - 50% recombinant = genes not on the same chromosome

2. Complete linkage

Parental line Ab/Ab x aB/aB

AaBb x aabb

AB/ab x ab/ab

1AB ab

1ab

Result 1: 1 of AB /ab to ab / ab

- BOTH are parental types, have 0% recombinant types.

3. Recombination

AB/AB x ab/ab

Ab/ab x ab/ab

Assume crossing over →

AB

Ab ab

aB
ab

Results : 10% recombinants (10cM / map units)

AB/ab 45%

Ab/ab 5%

aB/ab 5%

ab/ab 45%

Biggest ratio = the parental types most likely

Closer the two genes are together the less likely there is crossing over

The further apart two genes are the more likely crossing over

CHECK PARENTAL LINE TO KNOW WHICH ONES ARE SUPPOSED TO BE RECOMBINANTS

Practice Question

Brown seed pods (**B**) in a plant species is dominant to green (**b**), and elongated pods (**E**) is dominant over squished (**e**).

- (a) A fully heterozygous plant has the dominant alleles linked in trans (i.e., dominant alleles **not** on the same homologue) at a map distance of **20 cM**. What will be the genotypes of gametes produced by this plant, and in what frequencies (or percentages)?
- (b) If this plant is **self-pollinated**, what progeny phenotypes will you expect to see, and in what frequencies? Use a Punnett square to illustrate your answer.

Heterozygote genotype = $\frac{B \quad e}{b \quad E}$

Recombinant gametes = **B E** and **b e**, 20% total = 10% each

Parental type gametes = **B e** and **b E**, 80% total = 40% each

		gametes and frequencies			
		0.4 Be	0.4 bE	0.1 BE	0.1 be
parental	0.4 Be	Be/Be 0.16	bE/Be 0.16	BE/Be 0.04	be/Be 0.04
	0.4 bE	Be/bE 0.16	bE/bE 0.16	BE/bE 0.04	be/bE 0.04
non-parental	0.1 BE	Be/BE 0.04	bE/BE 0.04	BE/BE 0.01	be/BE 0.01
	0.1 be	Be/be 0.04	bE/be 0.04	BE/be 0.01	be/be 0.01
Progeny phenotypes:		BE 0.51	Be 0.24	bE 0.24	be 0.01

EXAMPLE

Example: If we cross **pr vg / + +** x **pr vg / pr vg**, and the distance between these two genes is 11 cM, what is the probability of obtaining an individual that is **+ vg**?

- 0.30%
- 2.23%
- 5.5%
- 20.7%

Example: If we cross $pr\ vg / ++ \times pr\ vg / pr\ vg$, and the distance between these two genes is 11 cM, what is the probability of obtaining an individual that is $+ vg$?

- A** 0.30% 6
- B** 2.23% 17
- ✓ **Correct C** 5.5% 83
- D** 20.7% 13

EXAMPLE

Example: If we cross $pr\ vg / ++ \times pr\ vg / pr\ vg$, and the distance between these two genes is 11 cM, what is the probability of obtaining an individual that is $+ vg$?

a. 0.30%
 b. 2.23%
 c. 5.5%
 d. 20.7%

Practice Problem

Spock (1st officer of the Enterprise) is from planet Vulcan. His mother is from Earth. A Vulcan has pointed ears (P), no adrenal gland (no synthesis of adrenaline – A), and a heart located on the right (R). All Vulcan alleles are dominant to Earthling alleles. The genes are also all linked on an autosomal chromosome

$\begin{array}{c} P/p \qquad \qquad \qquad A/a \qquad \qquad \qquad R/r \\ \leftarrow 15\text{ U.C.} \rightarrow \qquad \qquad \qquad \leftarrow 20\text{ U.C.} \rightarrow \end{array}$

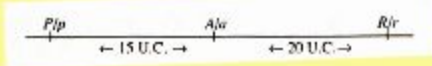
If Spock marries an Earth woman which proportion of their children:

d) Will have Vulcan ears, and the heart and adrenal gland of an Earthling?

A. 6.0%
 B. 8.5%
 C. 12.0%
 D. 17.0%

Practice Problem

Spock (1st officer of the Enterprise) is from planet Vulcan. His mother is from Earth. A Vulcan has pointed ears (P), no adrenal gland (no synthesis of adrenaline – A), and a heart located on the right (R). All Vulcan alleles are dominant to Earthling alleles. The genes are also all linked on an autosomal chromosome



If Spock marries an Earth woman which proportion of their children:

- c) Will have Vulcan ears and heart, but with an Earthling adrenal gland?

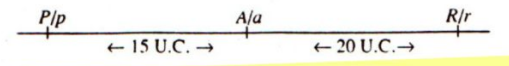


- A. 1.5%
B. 3.0%
C. 6.0%
D. 12.0%

Practice Problem

Spock (1st officer of the Enterprise) is from planet Vulcan. His mother is from Earth. A Vulcan has pointed ears (P), no adrenal gland (no synthesis of adrenaline – A), and a heart located on the right (R). All Vulcan alleles are dominant to Earthling alleles. The genes are also all linked on an autosomal chromosome

Previous slide



If Spock marries an Earth woman which proportion of their children:

- a) Will have the Vulcan characteristics?

- A. 34%
B. 35%
C. 68%
D. 70%

MODULE 9:

Which of the strands of DNA could act as a primer for the DNA sequence shown below?

Handwritten notes: 6ATTCA: 5' and CCTC: 3'

5' - CCCTGGGCTCTGTAAATGTTTCTAAGTG - 3'
3' - GGGACCCGAGACATTTACAAAGATTTCAC - 5'

A 3' - ACTGTTAGA - 5' (7)
B 3' - AAATTTGGC - 5' (10)
C 3' - ATGCTTTGA - 5' (5)
D 5' - GGGACCCGA - 3' (28)
E 5' - CCCTGGGCT - 3' (85)

Answer: E. Top strand:

Reversible primer

Which enzyme(s) will produce a DNA fragment that contains the entire vgp gene and has "sticky ends"?

The diagram shows a section of human DNA that contains a gene for VIDEO GAME PROFICIENCY (vgp gene), shown in red. Shaded areas mark the restriction sites of four restriction enzymes. EcoRI, HaeIII, BamHI, and HindIII. Arrows indicate where each enzyme cuts the two DNA strands:



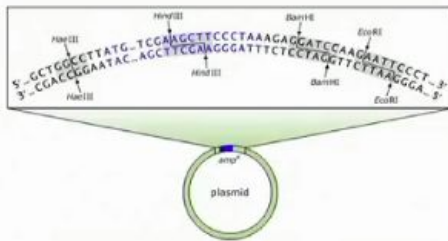
Which enzyme(s) will produce a DNA fragment that contains the entire vgp gene and has "sticky ends"?

- | | |
|--------------|--------------|
| I. BamHI | A. Only I |
| II. EcoRI | B. I and IV |
| III. HindIII | C. II |
| IV. HaeIII | D. I and III |
| | E. Only III |

ANS D

Which one restriction enzyme satisfies all three of the requirements listed above?

To clone the vgp gene into the plasmid, a restriction enzyme must do all of the following: (1) cut the human DNA on both sides of the vgp gene, (2) cut the plasmid without cutting inside the ampicillin gene (amp^R), (3) produce sticky ends when it cuts both the human DNA and the plasmid so that the human DNA fragments can combine with the plasmid.



Which one restriction enzyme satisfies all three of the requirements listed above?

- A. BamHI
- B. EcoRI
- C. HindIII
- D. HaeIII

ans A

Some of the bacterial cells fail to grow on medium + antibiotics. Why?

- A. The gene for antibiotic resistance is not cut by restriction enzymes in dead bacterial cells.
- B. Your DNA of interest only replicates a few times in dead bacterial cells.
- C. Those cells die because the DNA of interest (with the antibiotic resistance gene) never got in there.
- D. The antibiotic gene is not in dead bacterial cells but the plasmid is there.

ANS C

Below is a list of techniques and substances involved in the steps to make recombinant DNA. Choose the answer that best describes the correct order of using these techniques/substances in cloning.

Below is a list of techniques and substances involved in the steps to make recombinant DNA. Choose the answer that best describes the correct order of using these techniques/substances in cloning:

- I. PCR (Polymerase Chain Reaction)
- II. Introduce DNA into bacterial cells
- III. Medium + Antibiotics
- IV. Restriction Enzymes
- V. DNA Ligase

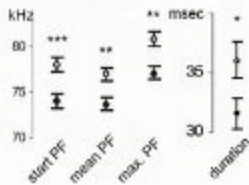
- A. I, II, III, IV, V
- B. II, III, IV, V, I
- C. I, IV, V, II, III
- D. I, II, IV, V, III

- A. I, II, III, IV, V
- B. II, III, IV, V, I

ANS C

Researchers did sound tests on normal and FOXP2 transgenic mice. Which is the most reasonable conclusion drawn from the figures?

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- Normal Mice
 - FOXP2 Transgenic Mice
- PF: Peak Frequency

- A. The sound duration time of transgenic mice is less than the mean PF of normal mice.
- B. Transgenic mice have lower mean PF than normal mice.
- C. The max PF of transgenic mice is similar to the mean PF of normal mice.
- D. All of the above.

- A. The sound duration time of transgenic mice is less than the mean PF of normal mice.
- B. Transgenic mice have lower mean PF than normal mice.
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ANS D