Nov 2014 H2 Bio Paper 2

N14	2Q1
(a)	 With reference to Fig 1.1, describe the curve between points A and B [2]. 1. As temperature increases from 35°C (pt A) to 50°C, the rate of reaction of enzyme increases gradually from 0 to 8a.u. 2. However, from 50°C to 65°C (pt B), there is a steeper increase in rate of reaction of enzyme from 8a.u. to 65 a.u. This increase is about 7 times that of the increase in rate from 35°C to 50°C.
(b)	 Explain why the reaction rate changes from: (i) A to B [3] 1. As temperature increase from 35°C (at A) to 65°C (at B), there is increase in kinetic energy of the enzyme and substrate molecule. 2. which increases <u>frequency of effective collisions</u>* between substrate and enzyme thus rate of formation of <u>enzyme-substrate complex</u>* increases; 3. increasing temperature also increases the <u>number of molecules having sufficient energy</u> to <u>overcome</u> the <u>activation energy</u>* barrier to form the products of reaction; (ii) C to D [3] 1. At <u>75°C</u> (pt C), the <u>optimum temperature</u>* of Taq polymerase, there is greatest number of molecular collisions and hence <u>max rate of reaction</u> 2. As temperature increase further to <u>85°C</u> (pt D), which is beyond optimum temperature, thermal agitation / vibration of enzyme molecule <u>breaks hydrogen</u>, ionic bonds and other weak interactions that stabilizes the 3D <u>conformation resulting in <u>denaturation</u>*</u> 3. The enzyme <u>active site</u>* no longer <u>complementary in shape</u>* and charge to the <u>substrate</u> and the <u>rate of reaction decreases steeply</u> from maximum rate of 95 a.u. to almost 0a.u.
(c)	 Suggest how the structural features of Taq polymerase make it thermostable [2] 1. <u>More Cysteine</u>* residue may be present in the Taq polymerase allowing the formation of <u>more disulphide linkages</u>* with another cysteine residue to maintain the 3D conformation 2. Disulphide linkages are <u>strong covalent bonds</u> difficult to be broken by high temperature 3. Hence the 3D conformation of active side maintained \
(a)	 Give the full name of the bond holding the two glucose molecules together in the way shown. [2] 1. α-1,4-glycosidic bond
(b)	 Describe how this bond may be broken to release two molecules of glucose. [2] 1. <u>Addition of water in <i>hydrolysis</i></u>* reaction 2. Enzymetric action of molecules

- Enzymatic action of <u>maltase</u>
 Acid hydrolysis by heating it with hydrochloric acid (either 2. or 3.)

- (c) (i) Describe the arrangement of phospholipids in cell membranes. [2]
 - 1. <u>2 layers</u> of phospholipid molecules in a <u>bilayer</u>
 - 2. With the <u>hydrocarbon tails</u> facing the <u>inside</u> of the membrane and the <u>phosphate heads</u> facing the <u>outside</u> next to <u>aqueous</u> medium
 - (ii) Explain how the structure of phospholipids is related to this arrangement in cell membranes. [3]
 - 1. <u>Phosphate heads are charged and hydrophilic and will form hydrogen with water</u>
 - 2. <u>Hydrocarbon tails</u> are <u>non-polar</u> and therefore <u>hydrophobic</u> and arranged away from aqueous medium
 - 3. And forms <u>hydrophobic interactions</u> with the hydrocarbon tails of other phospholipid molecules to form a hydrophobic core in the <u>bilayer</u> structure

	[Total : 9]
N14P2Q3	

(a) Describe how bacterial chromosomes differ from eukaryotic chromosomes in terms of structure and organization. [4]

Feature	Prokaryotic genome	Eukaryotic genome		
<u>Size</u>	10 ⁵ -10 ⁷ base pairs	10 ⁷ -10 ¹¹ base pairs		
Appearance	Generally a single, circular Multiple, linear molecules			
Molecule	Double Hel	lix DNA		
Association with	Yes - relatively less	Yes – large amounts		
proteins	e.g. histone-like proteins	e.g. histones, scaffold proteins		
Level of DNA packing/coiling	Relatively low : DNA double helix some looping around histone-like proteins	High: DNA double helix A Histones Linker DNA ("bead") Linker DNA ("bead")		
	Formation of looped domains	Nucleosome (10-nm fiber) - Euchromatin B		
	Supercoiling	30-nm fiber - Heterochromatin Protein scaffold Protein sc		
	 (A) Unfolded chromosome from <i>E. coli</i> has a diameter of 430µm. (B) DNA is folded into chromosomal looped domains by protein- 	(A Metaphase chromosome with proteins called histones.		
	DNA associations. Six domains	DNA molecules are		
	are shown, but actual number is	negatively- charged, histone		

	about 50.	are positively-charged .
	(C) Supercolling cause further	bistones by electrostatic
	fills an area of about 1 um	interactions
	niis an area or about 1 µm.	Most of DNA is wound
		around octamors of 8
		histono protoine to
		form nucleosomes the 10
		nm fibre Remainder of DNA
		called linker, joins adjacent
		nucleosomes
		nucleosonico.
		(B) The 10-nm fibre coils around
		itself to form a 30 nm
		chromatin fiber (or solenoid).
		(C) The 30-nm fibre forms loops
		called looped domains (a
		300-nm fibre) when
		associated with scaffold
		proteins.
		(D) Supercoiling present. The
		loops further coil and fold to
		produce characteristic
		metaphase
		chromosome.
Location	Nucleoid region, not membrane-	Nucleus (surrounded by nuclear
	bound	envelope)
Extrachromosomal	Present – plasmids (much smaller	No plasmids (however
DNA	rings of DNA compared with bacteria	mitochondria and chloroplast
	chromosome)	have their own DNA)
Number of genes	4,500	25,000
Non-coding regions	Not common – typically less than	Common – about 98%
(between and within	15%	
genes)	None present	Many propert
I. Introns	(rore: only in some genes)	many present
ii promotore	(rare, only in some genes)	
iii enhancere/	Rarely present	Present
silencers		I IESCIIL
iv repeated	Few	Many
sequences		many
v operons	Many	Few known ones (e.g. in
		nematodes)
Origin of replication	One	Many
(per chromosome)		

*Genome refers to a complete set of genetic material in a particular cellular component.

- (b) Describe what occurs from the end of stage 3 up to stage 4, as shown in Fig. 3.1. [4]
 (i)
 - 1. After contacting a recipient cell, the sex pilus retracts, pulling the two cells together.
 - 2. the <u>F⁺ cell</u> then forms a temporary cytoplasmic *mating bridge** with the <u>F⁻ cell</u> and transfers its <u>F plasmid</u> DNA to it.
 - 3. <u>one strand of the double-stranded F plasmid is nicked by a nuclease</u>.
 - 4. The free <u>3' end</u> of the nick is <u>extended by DNA polymerase</u> for the synthesis of a <u>new</u> <u>complementary strand</u> using the intact strand as the template
 - 5. The <u>newly synthesized strand displaces the nicked strand which is transferred</u> <u>concurrently</u>, via the 5' end, across the mating bridge into the recipient cell.
 - 6. Upon completion of a unit length of the plasmid DNA (after 1 round), <u>another nick</u> <u>occurs to release the original strand</u> and end the replication of the newly synthesized strand;

(ii) Explain the role of F plasmid. [3]

- 1. F factor on F plasmid, <u>codes for proteins</u> necessary for the <u>formation of sex pili</u> and subsequent <u>cytoplasmic mating bridge</u>,
- 2. <u>allowing for conjugation</u> to occur between bacteria.
- 3. This allows for <u>bacterial genes</u> to be <u>transferred between bacteria</u> and hence <u>increases</u> <u>genetic variation</u> between bacteria.

[Total : 11]

N14P2Q4

(a)

- Suggest the reason for the peak in the rate of DNA synthesis shown in Fig. 4.1. [2] 1. Gene coding for ribosomal RNA was amplified;
 - **2.** To allow for an increase/high rate of ribosomal RNA synthesis during egg cell growth;
- (b) Describe and explain the pattern of transcription visible on the part of the DNA coding for ribosomal RNA, labelled X in Fig. 4.2. [2]
 - 1. <u>Multiple ribosomal RNA</u> transcripts are produced concurrently from transcription of a <u>single gene</u>,
 - and <u>length</u> of the ribosomal <u>RNAs increase progressively from left to right of</u> region X;
 - **3.** This is because the direction of transcription takes place in a single direction/ribosomal RNA is produced in 5' to 3' direction;
- c) Explain why such large amounts of ribosomal RNA are required in frog egg cells. [2]
 - 1. Large amounts of ribosomal RNA are required for synthesis of <u>large numbers of</u> <u>ribosomes</u> in the frog egg cells;
 - This is required for <u>high rate of protein synthesis</u> following fertilisation of the egg cells;

d)	S	uggest poss enes. [2]	sible roles for the	non-transcribe	<mark>d DN</mark> /	A that is f	ound b	etween	the ribosomal
	J	1. the no 2. which <u>trans</u>	the non-transcribed DNA contain <u>promoter</u> *; which binds <u>RNA polymerase</u> * and <u>general transcription factors</u> * to form the <u>transcription initiation complex</u> * for transcription to take place;						
	U	3. the no 4. which <u>facto</u>	the non-transcribed DNA may contain <u>enhancer</u> *; which binds <u>activators</u> * to promote the assembly of <u>general transcription</u> <u>factors</u> * and <u>RNA polymerase</u> * at the <u>promoter</u> * of the ribosomal gene to form						
	O	5. the nc 6. which <u>facto</u> the tr	the <u>transcription initiation complex</u> *; the non-transcribed DNA may contain <u>silencer</u> *; which binds <u>repressors</u> * to prevent the assembly of <u>general transcription</u> <u>factors</u> * and <u>RNA polymerase</u> * at the <u>promoter</u> * of the ribosomal gene to form the transcription initiation complex*:						
e)	E	xplain how	operons allow for	rapid response	by ba	acteria to	enviro	nmental	change. [2]
	 <u>multiple genes</u> coding for products involved in the same biochemical pathway a <u>under the control of a single <i>promoter</i></u>*; these genes are therefore <u>turned "on" or turned "off together</u>, allowing the bacteria to respond rapidly to changes in the environment; 						allowing the		
	N14P2	25							
5(a)	 Draw a genetic diagram to explain both crosses. [5] Use the following symbols to represent the different alleles involved: R -red flowers r -white flowers A -axial flowers a -terminal flowers Parental phenotype Red, axial flowers X White, terminal flowers Parental genotype RRAA X rraa Gametes (RA) 						S		
	F1 phe F1 phe F1 self	enotype enotype fing		RrAa All red, axial flowers RrAa x RrAa				[1] [1]	
		Gametes	RA	Ra	(rA	\mathbf{i}	ra	a	
		RA	RRAA Red , axial	RRAa Red, axial	Re	RrAA ed, axial	R Red	rAa , axial	
		Ra	RRAa Red , axial	RRaa Red, terminal	Re	RrAa ed, axial	R Red, t	raa erminal	
		rA	RrAA Red, axial	RrAa Red, axial	ial vhite, a		rı white	Aa axial	
		ra	RrAa Red, axial	Rraa Red, terminal	wh	rrAa ite, axial	rı White,	raa terminal	
	Offsori	na aenotype	RA	R aa		rrA		rraa]
	Offspring phenotype		e Red, axial flowers	Red, terminal flor	White, W wers axial flowers te		White, termina	nite, minal flowers	
	Phenotypic ratio		9.5	3.1		2.7			
	≈	ot offenring a				3		1	
	[1] relate genotype to phenotype								

[1] correct phenotypic ratio of 9:3:3:1

- (b) Explain how different characteristics can be inherited independently in dihybrid inheritance. [2]
 - 1. 9: 3: 3: 1 offspring/F₂ phenotypic ratio indicates the <u>alleles of each character are found on</u> <u>different chromosomes</u> and inherited independently of one another;
 - 2. The <u>alleles of different genes</u> <u>assort independently</u>* of each other because homologous pairs of chromosome <u>align randomly on either side of the metaphase plate</u>/ because <u>alignment of each homologous pair is independent</u> of other homologous pairs/ because two alleles for flower colour <u>segregate independently</u> of the two alleles for flower position.
- (c) Explain why heterozygous plants for this gene, Tt, have the same phenotype as homozygous dominant plants, TT. [3]
 - 1. Allele T codes for a <u>functional</u> enzyme/is the <u>functional</u> copy of the gene involved in synthesis of gibberellic acid.
 - 2. Allele t codes for the <u>non-functional</u> enzyme/ is the <u>non-functional</u> copy of the gene.
 - 3. In a heterozygote, allele T masks the presence of allele t.
 - Presence of 1 T allele (in <u>homozygous dominant</u> or <u>heterozygous</u> individuals) causes T allele to be expressed, producing functional enzyme that causes plants to be grow tall. (Allele T is dominant over t)

[Total : 10]

<mark>N14P2Q6</mark>

- (a) Name the structures labelled A, B, C, and D on Fig. 6.1. [4]
 - A Intergranal lamella
 - **B** Chloroplast envelope (R: inner membrane or outer membrane)
 - C Stroma
 - D Granum
- b(i) Suggest two advantages of these large protein complexes being held in the membranes of structures A and D in Fig. 6.1. [2]
 - Maintains the sequential arrangement of the <u>photosystems</u>* and <u>electron carriers of</u> <u>electron transport chain</u>* for the <u>flow of electrons</u>;
 - 2. <u>Maintains proton gradient for ATP synthesis</u> since the <u>hydrophobic core</u>* of the membrane is impermeable to protons and this is essential for chemiosmosis OR

The <u>enclosed thylakoid space</u> allows the build up of protons here by proton pumps found in the electron transport chain, enabling the <u>establishment of a proton gradient</u> across the thylakoid membrane for chemiosmosis.

- 3. Allows <u>ATP synthase</u> to be embedded in the <u>correct orientation</u>, where the <u>active site faces</u> <u>the stroma</u> / for <u>protons to diffuse</u> from <u>thylakoid space to stroma</u>.
- 4. Through <u>compartmentalization</u>, it <u>improves the efficiency</u> of the different reactions by bringing the large molecules involved in photophosphorylation <u>closer</u> together
- The thylakoid membrane is highly folded, provides a <u>large surface area to embed</u> Any of the elaboration: <u>many photosynthetic pigments / chlorophyll molecules</u> in the photosystems for <u>light</u> <u>absorption*</u> <u>many</u> electron carriers in the <u>electron transport chain</u> and <u>ATP synthase</u> for <u>ATP</u> production.
- (ii) Outline the role of ATP synthase that is held in these membranes. [2]
 - 1. <u>Diffusion of protons</u> from <u>thylakoid space into the stroma</u>, via ATP synthase
 - 2. Leads to formation of <u>ATP from ADP and inorganic phosphate</u> via <u>chemiosmosis</u>*.

- (c) Describe the role of NADP in linking the light dependent reactions to the Calvin cycle. [2]
 - 1. NADP is the *final electron acceptor** at the end of the <u>non-cyclic light dependent reaction</u>, which is then <u>reduced to NADPH</u>.
 - 2. <u>Reduced NADP/NADPH</u> is used to drive the <u>reduction of glycerate phosphate (GP) to</u> <u>glyceraldehyde-3-phosphate (G3P)</u>, with the use of ATP in the Calvin cycle.

[Total : 10]

N14P2Q7

- (a) Describe events occurring at A and B in Fig. 7.1. [4]
- Stage A
 - 1. Haemagglutinin* binds to sialic acid receptors* on host cell membrane
 - 2. Virus then enters host cell by <u>endocytosis</u>* where host cell membrane <u>invaginates and</u> <u>pinches off</u>, placing virus in an endocytic <u>vesicle</u>.

Stage B

- 3. Vesicle then <u>fuses with a lysosome</u>, and the resulting <u>drop in pH</u> stimulates <u>fusion of viral</u> <u>envelope with vesicle membrane</u>
- 4. Releasing nucleocapsid and eventually viral RNA into cytosol of host cell
- (b) Explain why it is necessary for the viral RNA to enter the host cell nucleus. [3]
 - 1. For <u>transcription</u>* where the <u>viral genome **RNA**</u>* is used as a <u>template</u>* to synthesise the <u>mRNA</u>* strands catalyses by the viral <u>**RNA** dependent RNA polymerase</u>*
 - 2. Which can be transported into cytosol as a *template** for *translation* of viral proteins
 - 3. which in turn acts as a *template** for the replication of new viral RNA genome
 - 4. Using the host cell's ribonucleotides (and ATP), which is abundant in nucleus, for its synthesis

(c) Suggest in outline how new strains of influenza virus may arise. [3]

- 1 New strains of viruses are formed as a result of <u>mutation</u> of the genome due to the <u>change</u> in the ribonucleotide sequence known as <u>antigenic drift*</u>
- 2 As a result of the <u>lack of proof reading</u> ability of <u>RNA-dependent RNA polymerase</u> in influenza / the <u>fast/high rate of replication</u> of the virus
- 3 gives rise to changes in the conformational of the glycoproteins
- 4 New viral strains are also formed when <u>two or more strains</u> of influenza viruses <u>infect a</u> <u>common/same host cell</u> where
- 5 <u>Reassortment of the different RNA segments</u> occur resulting in <u>recombination of genetic</u> <u>material</u> in a <u>virion*</u>, giving rise to <u>antigenic shift*.</u>

[Total : 10]

N14P2Q8

- (a) Suggest why different species of picture-wing fly show different banding. [3] 6L
 - 1. (<u>Phenotypic differences/different wing patterns</u>) <u>distinguishes species</u> in picture-wing flies;
 - 2. Such differences are due to <u>differences in DNA sequences/mutations</u> in <u>several gene</u> <u>loci</u>;
 - 3. These changes in DNA structure are <u>reflected</u> by the <u>different staining of DNA</u> / <u>different banding patterns</u> in the chromosomes;

- (b) Suggest why different species of picture-wing fly evolved on different islands. [4] 8L
 - 1. The islands were <u>geographically isolated</u> with the <u>sea</u> separating the <u>islands/allopatric</u> <u>speciation</u> (with mention of type of barrier);
 - 2. This meant that the **<u>gene flow</u>** between populations of flies on the islands were <u>**disrupted**</u>*;
 - 3. With the <u>different environment</u> with their <u>different selection pressures*</u>, e.g. predation, <u>habitats, available food sources</u> (name one example);
 - 4. <u>Allele frequencies</u> change because of <u>natural selection</u> and <u>genetic drift</u>*;
 - 5. The subpopulations <u>evolve independently</u> and <u>accumulate different mutations</u> and allele frequency changes, that over <u>time</u> led to <u>reproductive isolation</u> and formation of different species;

(c) Explain the differences between classification and phylogeny. [3]

Fig.8.3 was shown to direct you towards reasons related to the branching network of a phylogram. Any 3

- 1. Classification groups organisms based on <u>overall/morphological similarities</u> and does <u>not take into account the evolutionary history</u> of organisms while phylogeny traces the evolutionary history of organisms based on <u>ancestor-descendent relationships</u>;
- Classification involves grouping organisms into a <u>kingdom, phylum, class, order, family, genus and species</u> using a <u>hierarchical classification*</u> system but in phylogeny, organisms are arranged based on their <u>evolutionary relationship with each other</u> with each organism assigned a position on a branching tree relative to other organisms;
- 3. In classification, the organism is presented as a <u>binomial nomenclature/name</u> but in phylogeny, the organism is placed on a branch on a <u>phylogenetic tree</u>;
- 4. Classification <u>does not allow inference of speciation events</u> as it is just a naming system while phylogeny indicates <u>speciation events as the node</u> of the phylogenetic tree;
- 5. Classification does not allow inference of common ancestors while in phylogeny, descendants of a common ancestor are represented in the same branch;
- 6. You <u>cannot infer of how closely related 2 species</u> are especially since they are grouped together in the <u>same hierarchy</u>, e.g. "species" in classification. In phylogeny, how recently a branch point occurs indicates how closely related 2 species are;

[Total : 10]

N14P2Q9 (OUT OF SYLLABUS)

N14P2Q10

10 (a) Describe the structure and role of tRNA

[8]

Structure:

- 1. Transfer RNA (tRNAs) are single-stranded RNA
- 2. It <u>folds</u> back upon itself and held in shape by <u>hydrogen bonding between</u> <u>complementary base pairs</u> at certain regions to form a 3D L-shaped structure
- 3. <u>3 bases</u> form an <u>anticodon</u>*
- 4. <u>3' end with CCA stem</u> is <u>attachment</u> site for a specific <u>amino acid</u> that corresponds to anticodon.

Function:

- 5. <u>Aminoacyl-tRNA synthetase</u>* catalyse formation of bond between <u>specific amino acid</u> and <u>tRNA</u> with <u>specific anti-codon</u>.
- <u>Different tRNA</u> will have different <u>specific conformation and charges</u>* which are <u>complementary</u>* to active site of <u>corresponding aminoacyl-tRNA synthetase</u>.
- 7. tRNA carries amino acid to the ribosome
- Each tRNA has a <u>specific</u> <u>anticodon</u>* that binds to a <u>specific mRNA</u> <u>codon</u> via <u>complementary base-pairing</u>*.

- 9. Where <u>adenine</u> base pairs with <u>uracil</u> and <u>cytosine</u> base pairs with guanine, by forming <u>hydrogen bonds</u>*.
- 10. the <u>sequence of bases on mRNA</u> is translated into a <u>specific sequence</u> <u>of amino acids</u> in forming polypeptide chain.

(b) Describe how gene mutations may affect the protein coded for by a gene.

[7]

- 1. A gene mutation is a change in the nucleotide /base sequence in DNA*;
- 2. <u>Substitution</u> mutation where a nucleotide/base is replaced by a different nucleotide
- Consequence of mutation on RNA: Results in a change in sequence of the mRNA, and a <u>change in *codon*</u>*
- 4. Resulting in a <u>different sequence of amino acid/primary structure of protein;</u>
- 5. leads to <u>different</u> folding of polypeptide to <u>tertiary structure / 3D</u> <u>conformation.</u>

OR

<u>same</u> folding of the polypeptide to <u>tertiary structure / 3D conformation</u> occurs as the new amino acid coded for is <u>chemically similar</u>;

 Insertion or addition – occurs when one or several nucleotides are inserted in a sequence OR

<u>**Deletion**</u> – occurs when one or several nucleotides are removed from a sequence of base(s)

- 7. Consequence of mutation on RNA: Results in a frameshift mutation
- 8. Resulting in a <u>different sequence of amino acid/primary structure of protein;</u>
- 9. leads to <u>different</u> folding of polypeptide to <u>tertiary structure / 3D</u> <u>conformation.</u>

OR

truncated polypeptide hence non functional

- Inversion a segment of nucleotide sequences separates from the allele and rejoins at the original position but it is inverted;
- 11. Consequence of mutation on RNA: resulting in <u>different *codons*</u>* on the mRNA
- 12. Resulting in a <u>different sequence of amino acid/primary structure of protein;</u>
- 13. leads to <u>different</u> folding of polypeptide to <u>tertiary structure / 3D</u> <u>conformation.</u> OR

truncated polypeptide hence non functional

(c) For a named genetic disease, describe the causal mutation and outline its effect on the phenotype of an organism. [6]

Sickle cell anaemia

- 1) One genetic disease caused by mutation is *sickle cell anaemia**
- 2) <u>sickle cell anaemia</u> is a result of a point substitution mutation / a single base <u>substitution</u>,
- <u>thymine is replaced by adenine</u> in the gene coding for <u>the β globin</u> <u>chain;</u>

- 4) A change in a single base in the <u>6th triplet codon</u> resulting in <u>Valine</u> <u>replacing Glutamate</u> amino acid;
- 5) Valine being non-polar compared with charged glutamate results in a change in properties of the polypeptide chain;
- Causing changes in the <u>1°, 2°, and 3° structure</u> resulting in <u>change in</u> <u>3D conformation</u> and normal haemoglobin becomes sickle cell haemoglobin
- 7) When oxygen <u>levels are low</u> in the blood, the Hb S molecule undergoes an unusual conformation change that allows the Hb S to polymerise / <u>crystallise to form rigid fibres</u>.
- **8)** This causes red blood cells to change from a circular biconcave shape to <u>a sickle shape;</u>
- **9)** Sickle shaped RBC may obstruct blood vessels and interfere with blood circulation organ failure/pain;
- 10) Sickled RBC more fragile and makes them susceptible to lysis and active destruction by spleen results in <u>reduction in red blood cell</u> <u>numbers</u> causing anaemia/shortness of breath/tiredness;
- 11) It is a recessive condition requiring both alleles of the β globin chains to be mutated in order for the symptoms to appear;

OR

Cystic fibrosis

- 1. One other genetic disease caused by mutation is cystic fibrosis*
- 2. Which is caused by of <u>deletion mutation</u> involving the deletion of 3 nucleotides on chromosome 7, resulting in <u>loss of phenylalanine</u> in the polypeptide.
- This cause changes in the <u>1°, 2°, and 3° structure</u> resulting in <u>change in</u> <u>3D conformation of the cystic fibrosis transmembrane conductance</u> <u>regulator (CFTR)</u>
- 4. It is an autosomal recessive inheritance
- 5. This mutation results in either missing or defective CFTR
- 6. CFTR controls <u>movement of Cl⁻ into or out of cells</u> and influences Na⁺ transport indirectly.
- 7. When Cl⁻ is not transported out of epithelial cells, Na⁺ is also retained in the cell causing a high ion concentration, lowering water potential in cell
- 8. water cannot diffuse out of cells via osmosis, so water is retained in cells causing mucus in the lumen to be thick making it conducive for bacteria growth
- 9. thus causing severe breathing difficulty

[Total: 20]