



The University of Jordan

Genetics

Doctor 2011 | JUdoctor2011.wordpress.com

 /LajnehMedcom

أسئلة بينوات

mid-term :)

هشام

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DOCTOR 2011

Genetics Midterm Exam-2011

Note: some of the questions are answered, keep in mind that those answers are not absolutely correct, so it's better to verify and make sure about them..

the experiment of the drosophila legs (legs emerging from its head):

- 1-cell proliferation
- 2-cell differentiation
- 3-cell transformation
- 4-apoptosis

in the experiment of hershey & chase , they took advantage from one of the following "" this is not the way the question was ""

- 1- bacterial cells can get incontact to each others
- 2- differential radioactive labeling of both of DNA & Proteins
- 3- something about bacteriophages ...

after meiosis I, the primary spermatocytes have:

- 1- 23 chromosome & 46 chromatid
- 2- 46 chromosome & 92 chromatid
- 3- 23 chromosome &

one of these methods is used for the detection of DNA sequence and the bases appear in one lane:

- 1-radioactive labeled dideoxynucleotid
- 2- florescent labeled dideoxynucleotid
- 3- both 1 and 2 can be used
- 4- -radioactive labeled deoxynucleotid
- 5- florescent labeled deoxynucleotid

a diagram showing fragments of DNA ,, RFLP was used ,, which of the following might be a carrier state of the disease :

the answer is 2 bands one intermediate length and the other small length

A question about Repetitive DNA sequences, which one is true :

- A) LINE contain Alu sequence.
- B) VNTR are minisatellite
- c) STR are dispersed

A question showing a figure for a replication bubble , and asks to determine the lagging strand

The enzyme that is responsible for unwinding in prokaryotes :

- A) RNA polymerase
- b) general transcription factor

a figure that shows a termination sequence "hair pin " , and asking where we can find it :
occurs when the detachment happens between the DNA template & the RNA polymerase

About The proofreading mechanism , which one is correct :

- A) 3'-5' exonuclease
- B) 5'-3' endonuclease
- C) 3'-5' exonuclease

which of the following is NOT true about sigma subunit in the RNA polymerase :

- 1- not required for the basic catalytic activity of the enzyme
- 2- dissociate after a while of starting transcription
- 3- degraded immediately after dissociating
- 4- binds to specific sequences on the promoter

one of the following is true about telomerase:

- 1- inactivation of telomerase contributes for the extended lifespan of cancer cells.
- 2- telomerase extends the 5' end of the parental DNA template
- 3- it uses DNA template
- 4- it extends the daughter DNA strand to become longer than the parental DNA

how the microRNA function in inhibiting translation??

- 1- inhibit binding of RNA polymerase
- 2- degrade mRNA.
- 3- addition of stop codon

which of the following can't be detected directly by the PCR :

- 1- detection of bacterial or viral sequences(or infection)
- 2- cloning of specific DNA sequence
- 3- DNA sequencing
- 4- can detect the length

DNA microarray is used for the detection of all except :
the level of translation

a question about TF II H all are true except :

- 1- It is a helicase
- 2- It is a general transcription factor
- 3- It is specific to RNA polymerase II
- 4- It is a part of the transcription initiation complex

A question about lac operon regulation?!

A long question about how can u detect a mutation in an intron by using CG probe :

- A) comparative genomic hybridization
- B) western blotting
- C) northern blotting
- D) southern blotting

question about Chromatin Acetylation:

- 1_ Histone specific.
- 2_ enzymatic / non enzymatic reversible
- 3_ common in actively transcribed genes.
- 4_ protein-Coding region

how to make imperfect hybridization?

1- **lowering salt concentration**

2- Increasing the length

an image of tRNA having the anticodon sequence (3'- GAC -5') (the 3' -5' was concluded from the figure), what is the amino acid carried by it?

the answer >>Asp

-when an antibiotic was used to block translation in a prokaryote, the resulting protein was a dipeptide, what step was inhibited/blocked by this antibiotic?

1- initiation

2- translocation

a tRNA that is destined to bear the AA Cysteine was charged improperly by another AA which is Alanine. upon translation, what event of the following could take place?

the answer was:

the Cysteine anticodon in tRNA will base pair with corresponding codon in mRNA but it will incorporate Alanine instead of Cysteine

a characteristic that indicates prokaryotes:

the answer was>> the amino acid chain starts with N-formylmethionine

-In the presence of Heme, Protein (globin) synthesis is upregulated/stimulated by:

1- Dephosphorylation of eIF-2

2-increase the rate of hydrolysis of the GTP-elf2 complex

Constitutive expression of the lac operon in the absence of lactose could happen by:

Moving of the operator upstream to the promoter



one of the following is true about the depicted polyribosome structure:

the answer was that a ribosome closest to the 3' end of the mRNA bears a polypeptide chain that is longer from that in a one nearer to the 5' end ((which was not obvious in the one we encountered in the exam))

a transcription factor has a testosterone zinc finger domain, a domain that binds the hormone estrogen & an activation domain for progesteron, on of the following happens:

the answer was that testosterone zinc finger domain binds DNA then it was something related to estrogen binding...!

what does this abbreviation mean 11p15.5

answer>>on region 15.5 of the short arm of chromosome 11

in an experiment similar to that of griffth but with different naming and different observations, give what match the observations (he wants the transforming factor)

answer>>>

the transforming factor is a secretory protein that can transform live or killed non-pathogenic strains

why>>

protein...as when we used a protease the non-pathogenic strain did not transform.

secretory...as both live and killed pathogenic strains caused the transformation, if it were to be intracellular protein then the live pathogenic would not function

to make cDNA library and produce insulin from E.coli strains obtained, which of the following is not used in the process:

- a- RNA polymerase II
- b- antibiotic
- c...
- d...
- e...

in gel electrophoresis, which of the following is not true:

- a- migration in gel not only depend on size but on charge of DNA fragment as well
- b- a specific probe must be used to visualize the bands
- c- you can see the bands under the normal light
- e....

a question asking about the sequence of the mRNA for the given amino acid polypeptide chain:

>>>5'-AUG....UAG-3'

(only one of the options match those criteria of the mRNA)

about meiosis in males and females, which is true:

- a- commences in females at puberty, and at early embryonic life in males.
- b- primary oocyte take along time to completely finish MITOSIS.
- c- the products are always of equal size in both
- d- dictyotene is the fetal stage that is found in one of them but not the other.
- e....

a plot of PCR, asking the sequence of the template DNA strand.

method to answer>>>

-read the plot from down upward....this is the new strand from 5' to 3'

-in the question he didn't give a primer sequence...but if given put at the 5' end of the new

strand

- to find the template match with the new strand, but remember to reverse the direction so the template is 5' to 3'

ex) new strand 5'-GCTACGAA-3'....then template is 5'-TTCGTAGC-3'

all of the following indicate the presence of a protein coding region near (or proximal) to it, except:

a- termination signal.

b- enhancer

c- gene on the homology of the same specie. (sth like this)

d...

e...

which of the following is able to catalyze the formation of the peptide bond in the process of translation:

a- rRNA of the large ribosomal subunit

b- protein of the large ribosomal subunit

c- rRNA of the small ribosomal subunit

d- protein of the small ribosomal subunit

e...

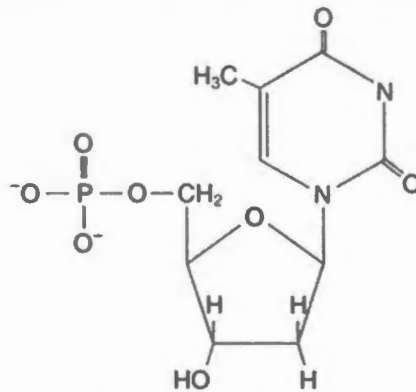
MOLECULAR BIOLOGY Midterm 2012

(1) The glycosidic bond that exists in nucleosides is between:

- (a) 3' carbon of sugar and N9 of adenine
 - (b) 1' carbon of sugar and N1 of guanine
 - (c) 5' carbon of sugar and N1 of cytosine
 - (d) 1' carbon of sugar and N9 of guanine
 - (e) 5' carbon of sugar and N1 of thymine
-

(2) The following molecule resembles:

- (a) Guanosine monophosphate
- (b) Cytidine monophosphate
- (c) Adenosine monophosphate
- (d) Deoxythymidin monophosphate
- (e) Deoxyadenosine monophosphate



(3) One of the following is part of the basal transcriptional complex:

- (a) Co-activator
 - (b) RNA polymerase
 - (c) Co-repressor
 - (d) Transactivator
 - (e) NONE of the above
-

(4) Which one of the following is less likely to have a DNA binding domain:

- (a) Myc
 - (b) Glucocorticoid receptor
 - (c) Thyroid hormone receptor
 - (d) TATA binding protein
 - (e) RNA polymerase
-

(5) Which of the following is **TRUE** regarding AZT:

- (a) Possesses a N₃ group on carbon 5'
 - (b) Not phosphorylated by the normal cellular kinases
 - (c) Has the same affinity towards viral reverse transcriptase and cellular polymerase
 - (d) It is a synthetic analog of a guanine nucleotide
 - (e) NONE of the above
-

(6) Which of the following is inhibited by the mushroom toxin α -amanitin:

- (a) RNA polymerase I
 - (b) RNA polymerase II
 - (c) RNA polymerase III
 - (d) DNA polymerase
 - (e) DNA Helicase
-

(7) The "proof reading" ability of DNA polymerase is due to:

- (a) 3' – 5' exonuclease activity
 - (b) 5' – 3' exonuclease activity
 - (c) 3' – 5' polymerase activity
 - (d) 5' – 3' polymerase activity
 - (e) NONE of the above
-

(8) Which of the following sequences can be cut by a restriction enzyme:

1	2	3	4	5
AATATTCGAGGATCAT	AAGGCTATTAGGCAAGCTTAGGCGAGCGAGTCGGACTCAAATCGAAG			

- (a) 1
 - (b) 2
 - (c) 3
 - (d) 4
 - (e) 5
-

(9) The polymerase enzyme responsible for mitochondrial DNA replication is:

- (a) δ DNA polymerase
 - (b) κ DNA polymerase
 - (c) γ DNA polymerase
 - (d) β DNA polymerase
 - (e) ζ DNA polymerase
-

(10) Which of the following is **TRUE** regarding the genetic code:

- (a) Not degenerate
 - (b) Absolutely universal
 - (c) Ambiguous
 - (d) Non-overlapping
 - (e) NONE of the above
-

(11) Which of the following is a signal transduction tumor suppressor:

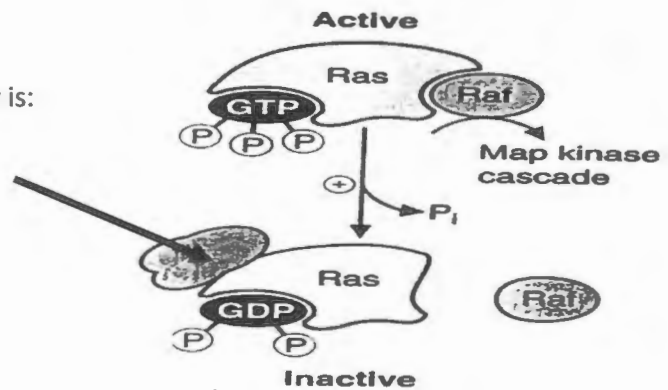
- (a) fos
 - (b) pRb
 - (c) p53
 - (d) E-cadherin
 - (e) NF-1
-

(12) Herceptin (trastuzumab) is a recent drug (monoclonal antibody) used to treat certain types of breast cancer that are influenced by which of the following characteristics:

- (a) BRCA 1 and BRCA 2 mutations
 - (b) HER2/NEU negative
 - (c) Overexpression of erb-b2
 - (d) Translocations
 - (e) NONE of the above
-

(13) The subunit indicated by the arrow is:

- (a) GTPase
- (b) GAP
- (c) p53
- (d) Rb
- (e) E2F



(14) A characteristic of cancer cells is:

- (a) Response to contact inhibition
- (b) Response to inhibitory growth signals
- (c) Resistant to senescence
- (d) Undergo apoptosis readily
- (e) Need stimulatory growth signals

(15) A sequence that is found at the 5' part of the 3' end of the promoter, just upstream to the transcription start point and is responsible for the binding of molecules required in regulating the level of transcription in prokaryotic cells is:

- (a) Operator
- (b) Repressor
- (c) Operon
- (d) Inducer
- (e) TATA box

(16) All of the following regarding gel electrophoresis are true EXCEPT:

- (a) Can be used to separate DNA but not RNA
- (b) Smaller molecules move faster than larger ones
- (c) Molecules move towards the positive electrode
- (d) The higher the density of the gel, the higher the resolution
- (e) Agarose and polyacrylamide gels can be used

(17) One of the following is a Bcl-2 family ion channel forming member responsible for the formation of pores in the mitochondrial membrane needed to release cytochrome c out to the nucleus:

- (a) Bcl-2
 - (b) Bcl-x
 - (c) Bok
 - (d) Bid
 - (e) NONE of the above
-

(18) The temperature needed by the thermostable DNA polymerase in PCR to synthesize a DNA strand is:

- (a) 94 °C
 - (b) 61 °C
 - (c) 55 °C
 - (d) 72 °C
 - (e) 14 °C
-

(19) Which of the following types of rearrangements is most responsible for hematological malignancies:

- (a) General recombination
 - (b) Retroviral reverse transcription
 - (c) Transposable elements
 - (d) Chromosomal translocations
 - (e) Chromosomal amplification
-

(20) Bulky adducts associated with DNA are repaired mainly by:

- (a) Nucleotide excision repair
 - (b) Base excision repair
 - (c) Mismatch repair
 - (d) Transcription coupled repair
 - (e) Translation coupled repair
-

(21) All of the following regarding introns is true **EXCEPT**:

- (a) Present only in eukaryotic cells
 - (b) Removed in the nucleus
 - (c) Spliced by snurps
 - (d) Intron shuffling results in the formation of proteins with similar functioning domains
 - (e) Possesses a 'AGGU' consensus sequence at its boundaries
-

(22) BRCA 1 and BRCA 2 repair genes are susceptible to mutations. Multiple forms of mutations have been associated with familial breast cancer in women. What is the best method to be used to diagnose these patients who have such a mutation:

- (a) Southern blotting
 - (b) DNA sequencing
 - (c) Northern blotting
 - (d) Allele-specific PCR
 - (e) Western blotting
-

(23) Which of the following regarding primers in eukaryotic cells is **TRUE**:

- (a) Made up of DNA subunits
 - (b) Consists of 200-300 nucleotides
 - (c) Formed by RNA primase
 - (d) Formed by the primase associated with DNA polymerase α
 - (e) Removed by DNase
-

(24) All of the following is true regarding mRNA synthesis in eukaryotes **EXCEPT**:

- (a) Takes place in the nucleus
 - (b) Synthesis takes place on one strand, the template strand
 - (c) Carried out by RNA polymerase II
 - (d) Splicing is the function of snRNPs
 - (e) Synthesis starts at the ATG codon
-

(25) One of the following is a post-translational modification:

- (a) 5' G-capping
 - (b) Polyadenylation
 - (c) Splicing
 - (d) Phosphorylation
 - (e) NONE of the above
-

(26) All of the following are true regarding polyadenylation **EXCEPT**:

- (a) Takes place in the nucleus
 - (b) 200-300 adenine residues are added to the 3' end of the mRNA
 - (c) Carried out by RNA polymerase II
 - (d) Is needed to stabilize the mRNA
 - (e) Adenine nucleotides are added after the polyadenylation sequence AAUAAA
-

(27) All of the following are true regarding p53 **EXCEPT**:

- (a) It is a transcription factor
 - (b) It stimulates the cell cycle by inducing expression of p21 CKI
 - (c) Its function is lost in most cancers
 - (d) It stimulates GADD45 transcription
 - (e) It can induce apoptosis by activating Bax if DNA repair is not successful
-

(28) Which of the following is not involved in regulation at the translational level:

- (a) Histone acetylase
 - (b) 5' mRNA loop
 - (c) 3' mRNA loop
 - (d) Eukaryotic initiation factors
 - (e) Ribosomes
-

(29) The genetic loci that are most recently used for DNA fingerprinting are:

- (a) VNTRs
 - (b) STRs
 - (c) snRNPs
 - (d) SNP
 - (e) RISC
-

(30) Which of the following is true regarding the energy requirement during translation:

- (a) 4 high energy bonds are needed (ATP and GTP are used)
 - (b) 1 ATP (cleaved to ADP) and 1 GTP (cleaved to GDP) are used
 - (c) 2 ATP are produced during the process
 - (d) No energy is needed
 - (e) NONE of the above
-

(31) Which of the following regarding aminoacyl tRNA synthetase is **TRUE**:

- (a) Attaches an amino acid to the 5' end of a tRNA
 - (b) The bond it forms between an amino acid and tRNA is a peptide bond
 - (c) Each aminoacyl tRNA synthetase is specific for a certain amino acid
 - (d) No energy is needed
 - (e) NONE of the above
-

(32) What is the type of mutation involved when the codon CGG undergoes mutation to become AGG:

- (a) Missense mutation
 - (b) Silent mutation
 - (c) Frame-shift mutation
 - (d) Nonsense mutation
 - (e) NONE of the above
-

	U	C	A	G	
U	Phe Phe Leu Leu	Ser Ser Ser Ser	Tyr Tyr STOP STOP	Cys Cys STOP Trp	U C A G
C	Leu Leu Leu Leu	Pro Pro Pro Pro	His His Gln Gln	Arg Arg Arg Arg	U C A G
A	Ile Ile Ile Met	Thr Thr Thr Thr	Asn Asn Lys Lys	Ser Ser Arg Arg	U C A G
G	Val Val Val Val	Ala Ala Ala Ala	Asp Asp Glu Glu	Gly Gly Gly Gly	U C A G

(33) The process by which different mature mRNAs are produced after transcription is called:

- (a) RNA editing
 - (b) Capping
 - (c) RNA remodeling
 - (d) Polyadenylation
 - (e) Conjugation
-

(34) All of the following regarding telomeres is true **EXCEPT**:

- (a) Telomeres consist of a repeated sequence of TTAGGG
 - (b) Telomeres are shortened by each cycle of DNA replication
 - (c) Telomerase enzyme is a RNA dependant DNA polymerase
 - (d) A 3' overhang on one strand develops due to the incapability of the primase to lay down a primer at the end of the chromosome
 - (e) Telomerase enzyme lengthens the short strand resulting in no overhangs at all
-

(35) Spinal muscular atrophy (SMA) is an incurable autosomal recessive disease caused by a gene c defect in the SMN1 gene where a deletion mutation takes place. What is the best method to be used in order to detect this mutation:

- (a) Southern blotting
 - (b) Northern blotting
 - (c) Western blotting
 - (d) PCR and electrophoresis
 - (e) DNA microarrays
-

(36) A severe and serious disease known as familial adenomatous polyposis (FAP) is caused by a mutation in:

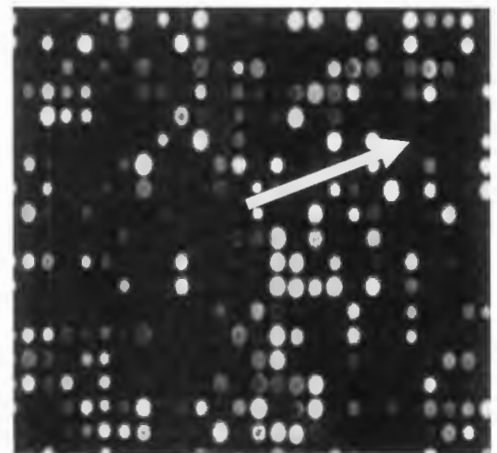
- (a) Mismatch repair genes
 - (b) β -catenin
 - (c) APC
 - (d) NF-1
 - (e) p53
-

(37) Which of the following is able to integrate a specific gene into a host cell:

- (a) Adenoviral vectors
 - (b) Retroviral vectors
 - (c) Liposomes
 - (d) Plasmids
 - (e) Cosmids
-

(38) A DNA microarray has been performed for 2 different cells. RNA obtained from those two cells was allowed to hybridize with the DNA chip. What does the dark spot (pointed arrow) indicate about the oncogene expression in cells A and B:

- (a) Oncogene is expressed more in cell A
- (b) Oncogene is expressed more in cell B
- (c) Equal amounts of oncogene expression in both cells
- (d) Neither cells express the oncogene
- (e) NONE of the above



(39) Which of the following regarding the promoter region is **TRUE**:

- (a) Located downstream the first transcription site
 - (b) Many genes share the same promoter
 - (c) A promoter can be activated by more than one inducing transcription factor
 - (d) TATA box sequences are the least conserved sequences
 - (e) NONE of the above
-

(40) Ras is considered a:

- (a) Growth factor proto-oncoprotein
 - (b) Growth factor receptor proto-oncoprotein
 - (c) Signal transduction proto-oncoprotein
 - (d) Transcription factor proto-oncoprotein
 - (e) NONE of the above
-

MARKSCHEIN

- | | |
|--------|-------|
| 1. D | 38. D |
| 2. D | 39.C |
| 3. B | 40.C |
| 4. E | |
| 5. E | |
| 6. B | |
| 7. A | |
| 8. C | |
| 9. C | |
| 10. D | |
| 11. E | |
| 12. C | |
| 13. B | |
| 14. C | |
| 15. A | |
| 16. A | |
| 17. C | |
| 18. D | |
| 19. D | |
| 20. A | |
| 21. D | |
| 22. B | |
| 23. D | |
| 24. E* | |
| 25. D | |
| 26. C | |
| 27. B | |
| 28. A | |
| 29. B | |
| 30. A | |
| 31. C | |
| 32. B | |
| 33. A | |
| 34. E | |
| 35. D | |
| 36. C | |
| 37. B | |

+ Genetics

Exam questions:-

Mid Exam Q. For the Past year...

Q. Which of the following is NOT true about DNA Replication?

- ☒ a) occurs during the M phase of the cell cycle
- ☐ b) makes a sister chromatids
- ☐ c) denatures DNA strands
- ☐ d) occurs semi-conservatively.
- ☐ e) code base pairing rules.

a → because we don't have DNA replication at Meiosis or mitosis.

Q. expression is:-

- a. the gene coding sequence to which RNA pol binds in order to initiate gene transcription
- b. the process by which the information contained in genes is used to produce proteins or functional RNA molecules

Q. Frame shift mutation:-

- a. single base substitution leads to coding for different a.a
- b. single base substitution leads to generation of stop codon

c. deletion + insertion of nucleotide NOT a multiple of 3 disturb the reading frame of a protein.

4. which of the following combination is not true?

- a. dominant -ve / multimeric prots subunits
- b. loss function / familial hypercholesterolemia
- c. gain mutation / charcot marie tooth
- d. trin expansion / fragile x syndrom
- ☒ e. wild allele mutation / large deletion

5. which of the following is not true?

- a. AR both parents of an affected person are heterozygous for the mutant gene
- ☒ b. each sibling of an affected person has absence of being affected.

6. genes that excrete effect on multiple effect of physiology & anatomy:-

- ☒ a. pleiotrophy
- b. germ line mosaicism
- c. delayed age onset
- d. reduced penetrance
- e. variable expression

7. Turner syndrome all is true except?

- ☒ a. premature cataract is common (No cataract)
- ☒ b. cells contain 45 x
- c. offspring have 50% of inherited disease
- d. ↑ risk of hypertension
- e. patient below average height

14] which is not true about mendelian inheritance :-

a. involvement of more than 1 generation in both autosomal + sex linked dominance by males + females are affected with equal frequency in both dom. and rec. autosomal inheritance

c. ↑ incidence of consanguinity in aut. R + d. x-linked usually males are affected

☒ male to male transmission can occur

15] Figure 1.1

in each age there is affected person → so dominant

in Male + Female so not x-linked

if there is skip in 1 generation, with M + F → autosomal R.

16] repeated P

17] mutation is single base substitution lead to coding for diff. a.a.
- Missense mutation

Rest in our micro lecture

على ما ذكره في المحاضرة السابقة

في خطنا سنرى شرحا

5

شغل عقلنا

في المحاضرة

Subject _____

Quiz Genetics

① According to Autosomal Recessive all. ✓ except:-
(Two deletions in the 2 ~~parents~~ parents in

Cystic Fibrosis (means 2 parents are carriers)

ASAS
AA AS
The proportion to have affected son is 50%

② Single-gene disorder in Familial hypercholesterolemia
due to :-

(Receptors) LDL

③ All following are Molecular techniques
except :-

(quinacrine G-banding)

④ Why the monosome is ~~lethal~~ ^{is} lethal?
(vital organs need two functional
monosomes to develop)

⑤ Ionizing radiation and chemicals in DNA
lesions :-

(strand breaks)

⑥ Imprinting, what is true :-

(expression occurs only from either
father or mother (one parent))

Subject _____

⑦ According to X-chromosome all ✓ except:-
(Lyon hypothesis ----- aneuploidy)

⑧ In _____, two or ~~more~~ three alleles in
the same locus, and it consists (1.4) of
population :-
(polymorphism)

⑨ Mixing of chromosomes in cross over
Recombination

⑩ Function of telomerase in dividing cell is
protect chromosomes ends only

Subject

Genetics

Chapter 3

(13) Wing combination :-

Marble disc / Autosomal Dominant

(14) Wing combination :-

Fragile X mainly / Frame shift

(15) According to ^{unlike} Amino acids which Galax :-
(it does not used to for chromosomal abnormalities)

(16) In Amino acids which of the following can make :-

(Karyotyping)

(17) Imbalance in chromosome segregation is :-
(Trisomy)

(18) How do you explain that every ~~it~~ have 50% to
and 50% to be a female :-

(BCO2 - egg from the mother has X)

(19) Fragile - syndrome which :-

(Affected male has > 200 repeated sequence)

(20) You have X chromosome, and chromosome 6

Why trisomy for 6 is lethal which it is not
lethal for X :-

(because X needs only one X for its activity, while other
2 Xs are inactivated)

(21) $\frac{1}{2} \times \frac{1}{2} = \frac{1}{4}$ or 25% probability

because X is excited in each genotype and
whether X, or Y have a probability of
being at about 50%.

(22) Don't forget tyrosine kinase enzyme

Subject _____

which of the following is not a storage dis. :-

(Galactosemia)

when I change the third nucleotide in the codon :-

(usually we have the same amino acid in reading)

According to Mendelian inheritance which follows :-

In Autosomal Dominant persons, his child 25% will be affected

Consanguinity ~~not~~ seen in :-

(Autosomal recessive)

You have a pedigree and you have a patient with no family history, what do you expect :-
(autosomal recessive)

Which combination :-

(large deletion / mild allele)

In which the following I ~~still~~ have normal karyotyping

(In mothers have child with Down syndrome)

UPD 15, 16, 17, 18, 19, 20, 21, 22

According also to Mendelian which follows

(In X-linked Autosomal recessive there is ~~not~~ rule for male transfer)

According to Down syndrome which ~~is~~ :-
(all of the above)

Down syndrome result from :-

(Maternal disjunction)

which of the following do not escape X inactivation

~~not~~ (G6PD gene)

According to mitochondrial inheritance which follows :-

(its protein heavily heavily from only mitochondrial genes)

Genetics

Which of the following is not correct about the XIST gene

- ☐ It is expressed only on the inactive X chromosome.
- ☐ It produces an RNA product (which coats the inactivated X chromosome) but no protein product
- ☐ It is expressed during embryonic development.
- ☐ It is expressed at twice the level in females as in males
- ☐ All of the above are true

Mutation in fibroblast growth factor receptor 3 (FGFR3)

- ☐ Retinoblastoma
- ☐ Achondroplasia
- ☐ Neurofibromatosis type 1
- ☐ Huntington disease
- ☐ Marfan syndrome

Abnormal binding of gene product to GAPDH (enzyme involved in glycolysis)

- ☐ Retinoblastoma
- ☐ Achondroplasia
- ☐ Neurofibromatosis type 1
- ☐ Huntington disease
- ☐ Marfan syndrome

phosphorylation of gene product by cyclin-dependent kinases (CDK); binding of gene product to transcription factors such as E2F

- ☐ Retinoblastoma
- ☐ Achondroplasia
- ☐ Neurofibromatosis type 1
- ☐ Huntington disease
- ☐ Marfan syndrome

Mutations in fibrillin gene

- ☐ Retinoblastoma
- ☐ Achondroplasia
- ☐ Neurofibromatosis type 1
- ☐ Huntington disease
- ☒ Marfan syndrome

Which of the following could produce an XY female?

- ☐ Deletion of the Sry gene
- ☐ Point mutation in the Sry gene
- ☐ Translocation of the Sry gene to the X chromosome during meiosis in the father
- ☐ None of the above
- ☒ All of the above

Which of the following is not a characteristic of cystic fibrosis?

- ☐ Chloride channel defect
- ☐ Hyperabsorption of intracellular sodium
- ☐ Elevated sweat chloride
- ☒ Fibrous ovarian cysts
- ☐ Pancreatic insufficiency

Each of the following chromosome abnormalities involves a 20 megabase region of the long arm of chromosome 5 (5q). Which abnormality is most likely to cause severe disease?

- ☒ Deletion of the region
- ☐ Duplication of the region
- ☐ A balanced translocation involving the region (i.e., in the translocation carrier)
- ☐ Pericentric inversion
- ☐ Paracentric inversion

Which of the following diseases is a good example of locus heterogeneity?

- ☐ Prader-Willi syndrome
- ☐ Myotonic dystrophy
- ☒ Osteogenesis imperfecta

- ☐ Duchenne muscular dystrophy
- ☐ Hemophilia A

Why are some autosomal dominant disorders (e.g., Marfan syndrome) seen more commonly in the offspring of older fathers?

- ☐ Replication errors accumulate as sperm-producing stem cells continue to divide
- ☐ Rate of nondisjunction increases in older males
- ☐ Recombination rates increase in older males
- ☐ All spermatocytes are produced during male embryonic development, so older males produce older sperm cells
- ☐ None of the above

A woman with phenotypically normal parents has two brothers with Duchenne muscular dystrophy. She experiences mild muscle weakness in her legs. Which of the following mechanisms is most likely to be directly involved?

- ☐ Germline mosaicism
- ☐ Skewed X inactivation
- ☐ Mutation near the pseudoautosomal region of the Y chromosome
- ☐ New mutation in this woman
- ☐ Nondisjunction of her mother's X chromosomes

Consider a fetus affected with one of the following conditions. For which condition is spontaneous loss during pregnancy most likely?

- ☐ Down syndrome
- ☐ Neurofibromatosis type 1
- ☐ Retinoblastoma
- ☐ Huntington disease
- ☐ Trisomy 18

A woman with phenotypically normal parents has two brothers with Duchenne muscular dystrophy. She experiences mild muscle weakness in her legs. Which of the following mechanisms is most likely to be directly involved?

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- ☐ Mutation near the pseudoautosomal region of the Y chromosome
- ☐ New mutation in this woman
- ☐ Nondisjunction of her mother's X chromosomes

X

Which of the following would not tend to raise mutation rates for single-gene disorders in humans?

- ☐ CG dinucleotides within the gene
- ☐ elevated paternal age
- ☒ caffeine exposure
- ☐ large genes
- ☐ all of the above are known to raise mutation rates in humans

Which of the following diseases results from faulty DNA repair?

- ☐ neurofibromatosis type 1
- ☐ retinoblastoma
- ☒ xeroderma pigmentosum
- ☐ imprinting on the X chromosome
- ☐ incomplete X inactivation

Which of the following does not pertain to cystic fibrosis (CF)?

- ☒ consanguinity is commonly seen in parents with CF offspring
- ☐ variable expression
- ☐ more than one mutation at the CF locus can cause the disease
- ☐ potential candidate for gene therapy
- ☐ pleiotropy

Which of the following is not true of Huntington disease?

- ☐ triplet repeat expansion is observed
- ☐ age of onset is delayed
- ☐ choreic movements are seen
- ☐ recurrence risk for offspring of an affected parent is 50%
- ☒ offspring of affected mothers have earlier age of onset than those of affected fathers

A likely explanation for the fact that some 45,X conceptions (Turner syndrome) survive to term is

- ☒ mosaicism in the fetus
- ☐ delayed age of onset of the disorder
- ☐ germline mosaicism in one of the parents

☐ imprinting on the X chromosome

☐ incomplete X inactivation

Which of the following is not a diagnostic feature of neurofibromatosis type 1?

☐ neurofibromas

☐ positive family history

☐ benign hamartomas of the iris

☐ melanomas

☐ cafe-au-lait spots

Which of the following is not true of the dystrophin protein?

☐ binds actin

☐ binds to sarcoglycan/dystroglycan complex

☐ can be assayed to diagnose Duchenne muscular dystrophy

☐ is absent in patients having Becker muscular dystrophy

☐ composes only a very tiny fraction of the mass of a muscle cell

Which individuals would you not expect to see a Barr body?

☐ 45,X female

☐ XY female

☐ a and b

☐ b and c

Which of the following is not often seen in the individual with Klinefelter syndrome?

☐ webbed neck

☐ increased stature

☐ diminished IQ (on average)

☐ sterility

☐ gynecomastia

What is the most likely explanation for the expression of hemophilia A in a female who is a heterozygote for an X-linked mutation in the factor VIII gene?

☐ nonsense mutation causing truncated protein

☐ her father is affected, and her mother is a heterozygous carrier

☐ a high proportion of the X chromosomes carrying the mutation are active in this

female

- ☐ one of her X chromosomes carries the SRY gene
- ☐ X inactivation does not affect the entire chromosome

Which of the following is least likely to increase the recurrence risk of trisomy 13 in a family?

- ☐ advanced maternal age
- ☐ mother carries a 13/14 Robertsonian translocation
- ☐ father carries a 13/14 Robertsonian translocation
- ☐ germline mosaicism in the mother in which some cells are 47,XX,+13
- ☒ advanced paternal age

Which of the following mutation types is expected to produce the mildest form of disease?

- ☐ Mobile element insertion
- ☐ frameshift deletion
- ☐ nonsense mutation
- ☒ missense mutation
- ☐ frameshift insertion

Individuals who inherit two active copies of the insulin-like growth factor 2 gene (Igf2)—one from their mother and one from their father—are at increased risk for developing the Wiedemann-Beckwith syndrome (large size at birth, large tongue, increased incidence of kidney tumors). This situation is an example of:

- ☒ autosomal recessive inheritance
- ☐ X-linked dominant inheritance
- ☐ imprinting
- ☐ mitochondrial inheritance
- ☐ relaxation of imprinting

Which of the following features would you be least likely to observe in neurofibromatosis type 1.

- ☐ cafe-au-lait spots
- ☐ Lisch nodules
- ☒ malignant tumors
- ☐ neurofibromas
- ☐ previous family history of the disorder

Concerning retinoblastoma, which of the following is not predicted by the two-hit model?

- ☐ sporadic tumors will be unifocal
- ☐ inherited tumors will be multifocal
- ☐ penetrance is incomplete
- ☐ two copies of the gene must be disabled for tumor expression to occur
- ☐ imprinting will be observed

Which of the following will yield the highest recurrence risk for Down syndrome in a family?

- ☐ 45-year-old mother
- ☐ 65-year-old father
- ☐ 25-year-old mother with a previous child with trisomy 21
- ☐ 20-year-old mother who carries a 21/14 Robertsonian translocation and has had no previous children with Down syndrome
- ☐ 20-year-old father who carries a 21/14 Robertsonian translocation and has had no previous children with Down syndrome

Which of the following chromosome disorders is the extra chromosome contributed in nearly all cases by the mother and the father?

- ☐ 47,XXY
- ☐ trisomy 21
- ☐ trisomy 13
- ☐ trisomy 18
- ☐ 47,YYY

A crossover event that places the SRY gene on the X chromosome can cause

- ☐ an XX male
- ☐ a male who has features similar to those of Klinefelter syndrome
- ☐ an XY female
- ☐ all of the above
- ☐ none of the above

Which of the following is not often seen in the child with Down syndrome?

- ☐ congenital heart defect
- ☐ hypotonia

- ☐ mental retardation
- ☐ respiratory infections
- ☐ simian crease

What is the best explanation for the muscle weakness seen in 8-10% of females who are heterozygotes for the Duchenne muscular dystrophy gene?

- ☐ variable expression of the disease gene
- ☐ X-linked recessive inheritance
- ☐ a high proportion of the X chromosomes carrying the mutation are active in this female
- ☐ relaxation of imprinting
- ☐ X-inactivation does not affect the entire chromosome

It has been suggested that schizophrenia is more severe and has an earlier age of onset in the more recent generations of schizophrenia families. If so this would be an example of:

- ☐ the two-hit model
- ☐ imprinting
- ☐ delayed age of onset
- ☐ anticipation
- ☐ increased numbers of mutagens in more recent generations

Which of the following is true of chromosome abnormalities?

- ☐ They are the leading known cause of spontaneous pregnancy loss
- ☐ in general, monosomies have more severe consequences than trisomies
- ☐ most of the aneuploidies seen among live births increase with maternal age
- ☐ none of the above
- ☐ all of the above

Sickle-cell disease is the result of a single nucleotide substitution that produces a single amino acid substitution. This is best described as a

- ☐ frameshift mutation
- ☐ nonsense mutation
- ☐ splice-site mutation
- ☐ missense mutation
- ☐ none of the above

Which of the following is not true of imprinting?

- ☐ It is often associated with methylation.
- ☐ Diseases involving imprinting genes can result from either over-expression or under-expression of the gene product.
- ☐ It can be specific to only certain tissues.
- ☐ Imprinting results in differential expression of a gene, depending on the gender of the individual who expresses the disease.
- ☐ Imprinting can affect multiple genes in a single chromosome region.

What is a likely explanation for the high prevalence of cystic fibrosis in European populations?

- ☐ Most of the disease genes are hidden in heterozygotes.
- ☐ The locus has a high mutation rate.
- ☐ Many different mutations can cause cystic fibrosis.
- ☐ Inbreeding is common among Europeans.
- ☐ Heterozygotes may have an advantage because of increased resistance to typhoid fever.

In one form of familial isolated growth hormone deficiency, deletion of an exon of a gene on chromosome 17 alters the protein product. In heterozygotes, this protein product then interacts with the normal protein product encoded by the normal copy of the gene on chromosome 17, disabling the normal growth hormone molecule. This is best described as an example of

- ☐ dominant negative effect
- ☐ haploinsufficiency
- ☐ gain of function mutation
- ☐ loss of function mutation
- ☐ none of the above

Which of the following characterize adult polycystic kidney disease?

- ☐ A. locus heterogeneity
- ☐ B. gain of function mutations
- ☐ C. 2-hit model of causation
- ☐ D. a and b
- ☐ E. a and c

The mutation that causes Huntington disease is best described as

- a CAG repeat expansion that occurs in the 3' untranslated region of the gene
- a CAG repeat expansion that produces a protein that tends to form aggregates in the nucleus of the cell
- a CAG repeat expansion that leads to haploinsufficiency and thus neuronal death
- a CAG repeat expansion that causes earlier onset of the disease in males than in females
- all of the above

Which of the following is not a characteristic of hemochromatosis?

- high prevalence in European populations
- locus heterogeneity
- pleiotropy
- different penetrance in males and females
- early diagnosis can lead to effective treatment and prevention

A phenotypically normal woman has had two children with trisomy 13. Which of the following scenarios would be least likely to result in this recurrence?

- she is a germ-line mosaic for trisomy 13
- she carries a Robertsonian translocation of chromosome 13 to chromosome 14
- she carries a Robertsonian translocation of chromosome 21 to chromosome 14
- she has had both children in her late 40s
- she carries a Robertsonian translocation of chromosome 13 to chromosome 15

Which of the following is not a feature of Down syndrome?

- hypotonia (poor muscle tone)
- reduced IQ
- association with elevated maternal age
- increased risk of leukemia
- cleft lip/cleft palate

Uniparental disomy in a live birth is caused by which of the following mechanisms?

- trisomic conception followed by subsequent loss of the chromosome that was contributed in single copy by one parent.
- triploid conception followed by subsequent loss of the extra set of chromosomes
- fusion of an egg cell and a polar body, resulting in a conception with no paternal genetic contribution

- monosomic conception followed by the gain of a chromosome
- all of the above

~~X~~ inactivation

- involves activation of the XIST gene on the X chromosome that becomes inactive.
- does not affect the entire inactivated X chromosome
- is associated with methylation of the inactive X chromosome.
- can lead to disease symptoms for an X-linked recessive disorder in a heterozygous female if the proportion of inactivated paternal and maternal chromosomes deviated strongly from 50/50.
- all of the above

Which of the following is not true of the trinucleotide repeat expansions that cause the neurodegenerative disorders Huntington disease and the spinocerebellar ataxias?

- A. they typically involve large expansions to several hundred or several thousand repeat units.
- B. They consist of CAG repeats located within exons
- C. They are considered to be gain of function mutations
- D. Expansion is more likely to occur when the repeat is passed through the father than through the mother
- E. In at least some cases, the mutation causes a buildup of protein aggregates in the cell nucleus.

Segmental neurofibromatosis, in which disease features are seen in only part of the body, would best be described as

- A. inherited mutation
- B. new mutation
- C. germline mosaicism
- D. somatic mosaicism
- E. none of the above

Which of the following features is not true of achondroplasia?

- a. nearly all disease-causing mutations occur at a single methylated CG dinucleotide within the gene
- b. surgical repair is possible for this disorder
- c. it is caused by mutations in a fibroblast growth factor receptor gene

- d. most cases are the result of a new mutation
- e. homozygotes and heterozygotes are equally severely affected

Which of the following is true of the Utah population?

- a. elevated rates of inbreeding are seen
- b. elevated prevalence of PKU, cystic fibrosis, and hemochromatosis
- c. relatively high rates of genetic drift
- d. genetically similar to northern European populations
- e. all of the above

In which of the following chromosome abnormalities is mosaicism most likely to be seen?

- a. Klinefelter syndrome
- b. Down syndrome
- c. XXX syndrome
- d. Turner syndrome
- e. Translocation Down Syndrome

Which of the following best explains variable expression in mitochondrial diseases?

- a. locus heterogeneity
- b. heterozygosity
- c. heteroplasmy
- d. modifier loci
- e. imprinting

Which of the following would be most likely to produce an imbalance in the amount of essential genetic material in the carrier?

- a. Robertsonian translocation
- b. Paracentric inversion
- c. Pericentric inversion
- d. Reciprocal translocation
- e. Isochromosome

Which of the following statements regarding chromosome abnormalities is true?

- ☐ a. Structural abnormalities increase with paternal age, while abnormalities of chromosome number increase with maternal age
- ☐ b. All chromosome abnormalities increase with maternal age
- ☐ c. For all abnormalities of chromosome number, the extra or missing chromosomes are most often the result of meiotic error in the mother
- ☐ d. all of the above
- ☐ e. none of the above

Which of the following conditions is most compatible with survival to term?

- ☐ a. trisomy 13
- ☐ b. trisomy 18
- ☐ c. monosomy X (45,X Turner syndrome)
- ☐ d. monosomy 21
- ☒ e. trisomy 21

Genetics Exam Questions

Question #1 of 18

It has been observed that cystic fibrosis (autosomal recessive) has a high prevalence in some populations because heterozygotes are resistant to the effects of chloride-secreting diarrhea. This is best described as an example of

- ☐ Mutation
- ☐ Gene flow
- ☐ Genetic drift
- ☒ Natural selection
- ☐ Linkage disequilibrium

Natural

Which of the following is not true of genetic diagnosis using fetal cells in the maternal circulation?

- ☒ PCR is used to amplify DNA from the fetal cell
- ☐ The procedure involves no risk to the fetus
- ☐ The procedure involves virtually no risk to the mother
- ☒ The procedure is more accurate than amniocentesis
- ☐ FISH analysis can be used to detect aneuploidy in the fetal cells

Using linkage analysis, you have mapped a disease gene to a 1 megabase (Mb) region of a specific chromosome. Which of the following approaches would be least useful in identifying and cloning the gene?

- ☐ Single strand conformation polymorphisms (SSCP) analysis
- ☐ Testing for cross-species conservation
- ☐ DNA sequencing
- ☐ Isolation of CG islands
- ☐ Radioactive in situ hybridization

Which of the following is a major difference in the mechanisms for generating diversity in T cell receptors and B cell immunoglobulins (antibodies)?

☐ Somatic hypermutation is seen in immunoglobulin genes but not in T cell receptor genes

☐ There are multiple variable region genes for immunoglobulins but not for T cell receptors

☐ Somatic recombination is seen only in immunoglobulin genes

☐ Junctional diversity is generated only in T cell receptor genes

☐ Junctional diversity is generated only in immunoglobulin genes

Why is there so much MHC (major histocompatibility locus) allelic diversity among individuals in populations?

☐ This region is especially susceptible to the effects of genetic drift

☐ The MHC region has a high level of somatic hypermutation

☐ Diversity enables the population to combat a larger number of different pathogens

☐ There are hundreds of different MHC loci, and they are shuffled by somatic recombination

☐ MHC diversity ensures that most tissue grafts will be rejected

A likely explanation for autoimmune disease is

☐ Repeated infection by the same microbe eventually stimulates an autoimmune response

☐ Depletion of T cell activity as a result of interleukin deficiency

☐ Depletion of B cell activity as a result of interleukin deficiency

☐ Lack of a memory B cell response

☐ Mimicry of self proteins by a foreign pathogen

A Northern Blot is used to

- ☐ Test polymorphisms in linkage analysis
- ☐ Analyze protein variation
- ☐ Analyze DNA sequence
- ☐ Analyze gene expression in different tissues
- ☐ Analyze a Southern blot that has mistakenly been run upside down

Which of the following is true of autosomal dominant breast cancer?

- ☐ It is characterized by locus heterogeneity
- ☐ It accounts for nearly half of all breast cancer cases in the United States
- ☐ It can be detected by hybridization with a single oligonucleotide probe
- ☐ Penetrance is close to 100%, with nearly all gene carriers developing breast cancer by age 80
- ☐ Autosomal dominant breast cancer affects females but not males

Which of the following is not true of type 2 diabetes (non-insulin dependent diabetes)?

- ☐ This form of diabetes is more highly heritable than type 1 diabetes
- ☐ Susceptibility to type 2 diabetes is partially due to the MHC and insulin genes
- ☐ Diet and exercise can decrease susceptibility to this form of diabetes
- ☐ Milder severity onset of diabetes in the young) is considered a highly heritable subset of this form
- ☐ This type of diabetes typically has a later age of onset than type 1 diabetes

Huntington disease is thought to be caused by a gain-of-function mutation. Which type of gene therapy would be potentially effective in treating this mutation?

- ☐ Replacement therapy with retroviral vectors
- ☐ Replacement therapy with adenovirus vectors
- ☐ Replacement therapy with adeno-associated vectors
- ☐ Antisense therapy
- ☐ All of the above

A neural tube defect (anencephaly or spina bifida) could be detected by

- ☐ Amniocentesis
- ☐ Chorionic villus sampling
- ☐ Ultrasound

X* X*

☐ A and B

☐ A and C

Which of the following is not an indication for prenatal diagnosis by amniocentesis?

☐ Previous child with a serious chromosome abnormality

☐ Both parents are heterozygous carriers of a mutation that causes Tay-Sachs disease

☒ Both parents have bipolar affective disorder (manic depression)

☐ Maternal age > 35

☐ Previous child or children with a neural tube defect

Which of the following would not be involved in a quantitative trait locus analysis?

☐ Linkage analysis

☐ Breeding of experimental animals

☐ Search for human homologs with a non-human probe

☒ Estimation of heritability

☐ Positional cloning of a disease-causing gene

Which of the following genetic abnormalities is not associated with Alzheimer disease?

☐ Mutations in the beta-amyloid precursor protein (BAPP) gene

☐ Presenilin mutations

☐ Trisomy 21

☐ Apolipoprotein E e4 allele

☒ LDL receptor mutations

Age-related macular degeneration

☐ BRCA1 and BRCA2 (breast cancer) gene products

☐ HOX (homeobox) gene family

☐ Leptin mutations

☒ Stargardt's disease mutations

☐ Glucokinase mutations

Interact with the Rad51 DNA repair system

☒ BRCA1 and BRCA2 (breast cancer) gene products

☐ HOX (homeobox) gene family

Leptin mutations

Stargardt's disease mutations

Glucokinase mutations

Transcription factors involved in limb development

BRCA1 and BRCA2 (breast cancer) gene products

HOX (homeobox) gene family

Leptin mutations

Stargardt's disease mutations

Glucokinase mutations

Maturity onset diabetes of the young (MODY)

BRCA1 and BRCA2 (breast cancer) gene products

HOX (homeobox) gene family

Leptin mutations

Stargardt's disease mutations

Glucokinase mutations

In one gene mapping technique, denatured DNA from metaphase chromosomes is hybridized with a radioactively labeled probe. This DNA is then exposed to film to reveal the approximate chromosomal location of the DNA in the probe. Which technique does this best describe?

Southern blotting

In situ hybridization

Somatic cell hybridization

Fluorescence in situ hybridization

Single strand conformation polymorphism (SSCP) analysis

Consider the table of LOD scores shown below for an autosomal dominant disease. The first line gives recombination frequencies, and each subsequent line represents the LOD score pattern found for a specific family. What phrase best describes this pattern?

q:	0.0	0.1	0.2	0.3	0.4
	2.2	1.4	1.0	0.8	0.2
	1.5	1.2	1.2	0.6	0.1
	2.0	1.0	0.8	0.3	-0.1
	1.2	0.8	0.2	0.1	0.1

- ☐ tight link
- ☐ Allelic heterogeneity
- ☐ Loose linkage
- ☐ Linkage disequilibrium
- ☐ Heterogeneity

Suppose you are attempting to find a disease-causing gene, and you have identified a number of families in which the disease is transmitted. If you have no knowledge of the gene product and no reasonable candidate locus, which of the following would be the first technique you would be most likely to use?

- ☒ Linkage analysis
- ☐ DNA sequencing
- ☐ Single strand conformation polymorphism (SSCP) analysis
- ☐ Denaturing gradient gel electrophoresis (DGGE)
- ☐ Fluorescence in situ hybridization (FISH)

Now suppose that you do have a reasonable candidate locus for the disease you are studying. Which of the following would be least likely to contribute useful information?

- ☐ DNA sequencing
- ☐ Single strand conformation polymorphism (SSCP) analysis
- ☐ Northern blotting
- ☐ Denaturing gradient gel electrophoresis (DGGE)
- ☒ Chromosome karyotype

Some autosomal recessive diseases have a high prevalence in large populations, even though they are often fatal (e.g., sickle cell disease in Africans, cystic fibrosis in Europeans). Which of the following is the most likely explanation for this phenomenon?

- ☐ Inbreeding
- ☐ High mutation rates in specific populations
- ☒ Survival advantage in heterozygous carriers
- ☐ Survival advantage in individuals who are normal homozygotes
- ☐ None of the above mechanisms explain the pattern

Which of the following would you not expect to see in a typical multifactorial disorder?

- ☒ Positive correlation between prevalence of the disorder in the population and sibling recurrence risk



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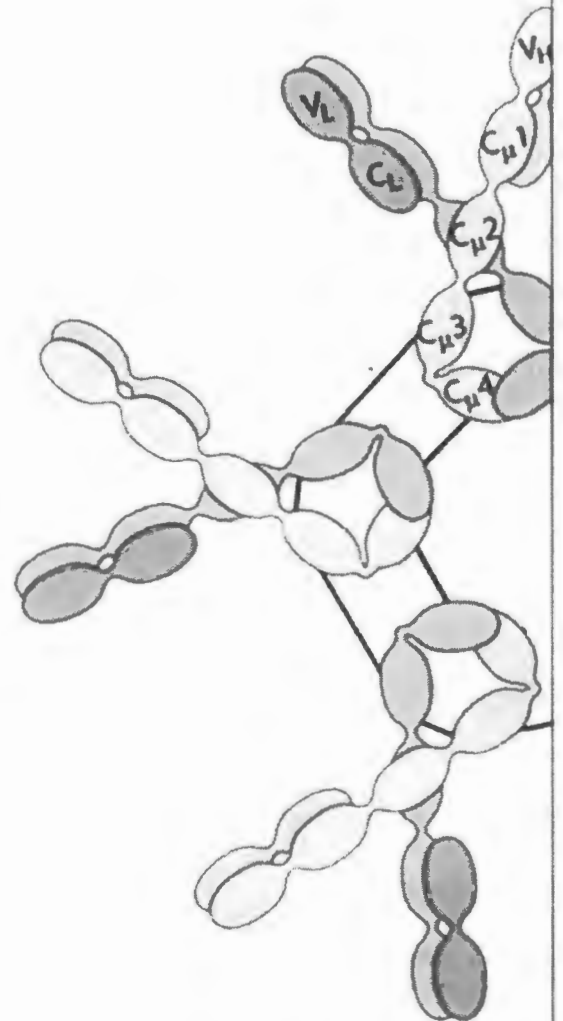
أسئلة سنوات

mid-term :)

Slides

Sheet

Handout



د. ف. ج. ح.
٢٠١٣ / ٥ / ١

Immunology Mid exam 2011

1. side effects of cyclophosphamide → hyperglycemia

2. About selection, All of the following are true except.

- A) negative selection happens in the cortico-medullary junction of the thymus.....
- B) the cells that leave the medulla are only 5% of the original thymocytes.....
- C) negative selection leads to killing of mostly DP and SP by thymic epithelial cells *** (by macrophages)
- D) the selection depends on the degree of receptor occupancy, duration and affinity

3. about RSS which is wrong

- a- haptamer and nonamer are aligned back to back
- b- they are separated by 12 or 23 spacer. *** (AND not OR !!!)
- c- rearrangement occurs for segments on the same chromosome
- d- the same type of segments cannot join each other
- e- imperfect & random loss or addition of nucleotides at and around the coding junction

4. all of the following are different between IgG1 and IgG3 except:

- a- serum concentration
- b- half life
- c- complement activation strength.
- d- number of disulfide bonds
- e- Placental passage ***

5- a question about allotype and idotype (wrong statement required)

- a- allotype occurs for all heavy chain types
- b- allotype not idotype is responsible for generation of immunogenicity of Ig
- c- allotype is a genetic marker present within all genes
- d- ...
- e- ...

6- wrong statement about somatic hypermutation ?

- 1- Hypermutation is T cell dependent
- 2- Mutations focussed on ICRs)
- 3- Cells with accumulated mutations are selected
- 4- Stimulated memory B cells & plasma cells
- 5- result from "affinity maturation" *** (It's the reason not the result!!!)

7- T and B cell differ in all of the following signal transduction steps except:

- a- generation of DAG from phospholipase C- gamma ***

which of the following is wrong about signal transduction pathway in T cell (or immunological synapse...not sure)

a- RAS is activated by $MA\bar{C}$ ***

b- IP3 is associated with CD28 and Fyn

c-...

8- the wrong statement about the classical & alternative pathways of the complement system ?

A) both pathways have the same c3 convertase ..***

9- T cell affect all of the following B cell processes except

a- clonal deletion ***

TCR differ from BCR as it

a- don't have an effector function***

b- unable to bind insoluble antigen

c- have variable and constant region

d- Unable to signal transduction.

10- about serological reaction which is not true:

a- indirect ELISA is used to detect the Ag

b- indirect IF detect Ab in serum

c- flow cytometry separate according to size, granulation, and surface marker

d-....

e- agglutination is the least sensitive ***

11- the wrong statment about Treg :

a- Development and maintenance are dependent upon B7

b- Suppress the activity of effector Th and Tc cells

c- Inhibition depends on cell:cell contact

d- Develop in the bone marrow from DP\SP cells *** " Develop in the THYMUS!!! "

e- Antigen specific and MHC restricted.

12- in radial immunodiffusion, which is not true:

1- Ab is diluted and put in a gel

2- require 48-72 hours to complete

3- concentration of Ag is directly proportional to the ring diameter square at end point

4- used to measure serum proteins

5- to faster the reaction electrophoresis is applied in a process known as rocket technique.

13- all true about Ig except:

a- papain treatment result in two Fab and one Fc

b- pepsin result in one F(ab)₂ fragment and one Fc ***

c- reduction result in 2 heavy chain and 2 light chain

d-...

e-...

14- binding of peptide to MHC molecule is all but (... i forget the statement):

a- saturable

- b- with low affinity
- c- slow in rate and very slow out rate
- d- noncovalent
- e- discriminate between self and non-self ***

15- all the following molecules are related to antigen processing and loading except:

- 1-TAP
- 2- invariable chain
- 3- HLA-DM
- 4- Calneurin *** (Its CALNEXIN!!!)

16- which of the following IL molecule is pro-inflammatory and activate B cell

- 1- IL-4
- 2- IL-5
- 3- IL-6 ***
- 4- IL-10
- 5- IL-1

17- all of the following are different between MHC I & MHC II except:

- a- structure
- b- cell distribution
- c- binding groove
- d- heterodimeric structure *** (Both are heterodimers)
- e- Antigen specificity

18- all of the following CD can be found in natural killer cell except:

- 1- CD8
- 2- CD5 *** (CD8 is found on the Natural killer cells!!! Review the CDs table its important))
- 3- CD16
- 4- CD56
- 5- CD45

19- which of the following is an absolute requirement of immunogenicity, and can't be manipulated

- 1- foreignness ***
- 2- molecular weight
- 3- complexity
- 4- physical nature
- 5- antigenic determinant

20-All the following are from the second line of defense except :

- 1- macrophages
- 2- neutrophils
- 3- antimicrobial proteins
- 4- anti-inflammatory responses
- 5- acidity of the stomach ***

21-regarding Interferon gamma, the wrong statement is:

- 1- it's an Immunosuppressant *** (It's an immunomodulator)

- 2- Induced by viral infection
- 3- produced using gene cloning technique.
- 4- cause flu-like symptoms
- 5- have anti-tumor effect

22- Of the following which is wrong regarding Ig Gene rearrangement?

- 1- the structure for variable (VL) gene segment is similar.
- 2- Heavy chain gene rearrangement involves three recombinational steps
- 3- Light chain gene rearrangement involves 2 recombinational steps
- 4-?
- 5-?

23- all the following sites are possible sites for hematopoiesis in adults except:

- 1- femur ***
- 2- skull
- 3- vertebrae
- 4- bones of the pelvis
- 5- ribs & sternum

24- all methods uses electrophoresis & gel diffusion except :

- 1- western blotting
- 2- immunonephelometry ***
- 3- immunoelectrophoresis

25- on if the following is NOT true about complement:

- A) homologous restriction factor prevents the binding of C5b to cell surface

26-One of the following is the only "current" use for Thalidomide:

- A) treatment of leprosy ***
- B) Multiple Myeloma

27- all the following is true except :

- 1- T- cells : periarteriolar sheaths in spleen & paracortex in lymph nodes
- 2- B-cells : primary & secondary follicles & in the cortex of lymph nodes
- 3- macrophages : in lymph node medulla & in marginal zones of spleen
- 4- direction of maturation of cells in the bone marrow is from center to periphery, while in the thymus from peripheral to center *** (Both from periphery to center).

28-all the following is true except :

- 1- IgG is the only antibody that is produced during the fetal life *** (Its IgM)

29- clonal deletion of B cells occur at the level of :

- 1- early pro-B
- 2- late pro-B
- 3- large pre-B
- 4- small pre-B
- 5- immature - B cell ***

30- All of the following methods are detecting primary Ag-Ab reactions except:

- 1- enzyme immunoassay
- 2- radioimmunoassay
- 3- immunonephelometry
- 4- immunofluorescence
- 5- immunofixation ***

31-an old drug that is widely used in organ transplantation & autoimmune diseases but with many toxicities :

- 1- thalidomide
- 2- cyclosporine ***
- 3- azathioprine
- 4- cyclophosphamide

32-sirolimus eluting coronary stents :

- 1- bind to immunophilin like 506 binding protein 12
- 2- they inhibit cell proliferation locally
- 3- they are more effective than aspirin in platelet aggregation
- 4- less renal toxicity **

By Lejan 2009\2010

الحمد لله الذي تتم بنعمته الصالحات
اللهم انك عفو كريم تحب العفو فاعفو عنا

IMMUNOLOGY MIDTERM 2012

(1) Which of the following immunoglobulins is homocytophilic and exhibits the lowest concentration in serum:

- (a) IgG
 - (b) IgM
 - (c) IgD
 - (d) IgA
 - (e) IgE
-

(2) All of the following is true regarding immunoglobulins EXCEPT:

- (a) Domains have intramolecular disulfide bonds
 - (b) Domains are constant in light chains, but vary in heavy chain
 - (c) Both light and heavy chain are glycosylated
 - (d) Hypervariability takes place in the variable region
 - (e) Kappa chains are more frequent than lambda chains
-

(3) The most constant region in an immunoglobulin is:

- (a) Fab
 - (b) Fc
 - (c) Hinge Region
 - (d) Heavy Chain
 - (e) Light Chain
-

(4) TCR and BCR differ in all of the following EXCEPT:

- (a) Type of the antigen
 - (b) Molecules needed for activation
 - (c) Ability to cross-link the antigen receptor
 - (d) Ability to bind MHC molecules
 - (e) Ability to trigger effector functions
-

(5) The best test to diagnose monoclonal gammopathy is:

- (a) Flow cytometry
 - (b) Electroimmunoassay
 - (c) Immunoelectrophoresis
 - (d) Immunofixation
 - (e) Western Blotting
-

(6) Mature B cells have all of the following **EXCEPT**:

- (a) FCγR
 - (b) CR1 and CR2
 - (c) IL-7R
 - (d) MHC class II
 - (e) IgM
-

(7) Macrophages secrete all of the following **EXCEPT**:

- (a) IL-1
 - (b) IL-3
 - (c) IL-6
 - (d) IL-8
 - (e) TNF
-

(8) All of the following cytokines are needed in hematopoiesis **EXCEPT**:

- (a) IL-3
 - (b) Stem Cell Factor
 - (c) IL-1
 - (d) EPO
 - (e) Colony Stimulating Factor
-

(9) Activated B cells increase expression of all the following molecules during the cell cycle **EXCEPT**:

- (a) CD80/CD86
 - (b) IL-2
 - (c) IL-4
 - (d) CD28
 - (e) Antiapoptotic proteins
-

(10) All of the following molecules regarding T cells are involved in the immune synapse **EXCEPT**:

- (a) LFA-1
 - (b) CD3
 - (c) ICAM-1
 - (d) CD2
 - (e) CD28
-

(11) All of the following tests are used to detect secondary immune complexes **EXCEPT**:

- (a) Immunonephelometry
 - (b) Radial immunodiffusion
 - (c) Immunofixation
 - (d) Immunoelectrophoresis
-

(12) All of the following cells have immunoglobulins or complement receptors **EXCEPT**:

- (a) Monocytes
 - (b) B cells
 - (c) T cells
 - (d) Macrophages
 - (e) NK cells
-

(13) NK cells kill infected cells using all of the following molecules **EXCEPT**:

- (a) NKG2D
 - (b) CD16
 - (c) CR3 and CR4
 - (d) MHC class II
 - (e) KIR
-

(14) Antigen-naïve T cells differ from memory T cells by which of the following markers:

- (a) CD28
 - (b) CD3
 - (c) CD45
 - (d) CD2
-

(15) All of the following tests are used to detect primary immune complexes **EXCEPT**:

- (a) Flocculation
 - (b) Immunonephelometry
 - (c) Radioimmunoassay
 - (d) Enzymeimmunoassay
 - (e) Immunofluorescence
-

(16) All of the following regarding the development and differentiation of T cells is true **EXCEPT**:

- (a) IFN- γ blocks differentiation into Th2
 - (b) IL-2 is involved in both Th1 and Th2 development
 - (c) IL-10 stimulates development of Th1
 - (d) IL-4 blocks development of Th1
 - (e) IL-12 stimulates differentiation into Th1
-

(17) IL-17 secreted by Th17 cells suppress the differentiation of which of the following cells:

- (a) Th1
 - (b) B cells
 - (c) NK cells
 - (d) Th2
 - (e) Treg
-

(18) All of the following regarding the systemic effects of activated macrophages are true **EXCEPT**:

- (a) Activation of complement and opsonization
 - (b) Increases temperature of the body
 - (c) Decreased specific immune response
 - (d) Stimulation of phagocytosis
 - (e) Decreased viral and pathogen replication
-

(19) The molecule that is stabilized by properdin is:

- (a) C3b
 - (b) C5b67
 - (c) C3bBb
 - (d) C4b
 - (e) MBL
-

(20) A molecule important in the signal transduction pathway of T cells and its absence can lead to immunodeficiency is:

- (a) LAT
 - (b) ZAP-70
 - (c) Lck
 - (d) Ras
 - (e) IP3
-

(21) All of the following molecules are involved in antigen processing **EXCEPT**:

- (a) TAP
 - (b) CLIP
 - (c) Invariant chain
 - (d) HLA-DM
 - (e) Calneurin
-

(22) All of the following are true regarding intraepithelial lymphocytes **EXCEPT**:

- (a) Majority are T lymphocytes of the CD8+ $\gamma\delta$ type
 - (b) They recognize Ag directly (no need for MHC)
 - (c) They develop from double positive cells in the thymus
 - (d) Responsible for oral tolerance
-

(23) Conventional T cells are characterized by all of the following **EXCEPT**:

- (a) They use the $\alpha\beta$ TCR
 - (b) They express CD4+ (helper) and CD8+ (cytotoxic)
 - (c) Majority of T cells
 - (d) Restricted by MHC class I and II
 - (e) Broadly specific
-

(24) All of the following regarding complement system activation is true **EXCEPT**:

- (a) C5b binds to cells and activates C6 and C7
 - (b) Only IgG and IgM activate the complement system
 - (c) C1, C2 and C4 may not be used
 - (d) C5b67 binds to C8 and C9 forming MAC
-

(25) Light chain rearrangement will result in B cell maturing into what stage:

- (a) Early Pro-B cell
 - (b) Mature B cell
 - (c) Late Pre-B cell
 - (d) Immature B cell
 - (e) Late Pro-B cell
-

(26) All of the following molecules have an anaphylatoxin or chemoattractant activity **EXCEPT**:

- (a) C3b
 - (b) C5a
 - (c) C3a
 - (d) C5b67
 - (e) C4a
-

(27) All of the following are oxygen in-dependant methods of killing by effector cells **EXCEPT**:

- (a) Lysozymes
 - (b) Defensins
 - (c) Hypochlorite
 - (d) Hydrolytic enzymes
-

(28) Regarding the location of T cells, all of the following are true **EXCEPT**:

- (a) PALS in spleen
 - (b) Cortex of lymph nodes
 - (c) Lymphoid organs
 - (d) Medulla of thymus
 - (e) Serum
-

(29) Ideotype is best described as:

- (a) Variation in the F_{ab} fragment
 - (b) Antigenicity of the Fab fragment
 - (c) Variation in the CDR
 - (d) Internal image of the antigen
-

(30) Regarding IL-3, all of the following is true **EXCEPT**:

- (a) Produced by T cells
 - (b) Binds to receptors on progenitor cells
 - (c) Induces proliferation
 - (d) Maintains stem cells and progenitor cells
 - (e) Induces differentiation
-

(31) All of the following are possible after immunoglobulin replacement therapy **EXCEPT**:

- (a) Activation of the complement pathway
 - (b) Promotion of phagocytosis
 - (c) Antigen recognition
 - (d) Neutralization of toxins and viruses
 - (e) Killing of target cells by NK cells (ADCC)
-

(32) All regarding HLA is true **EXCEPT**:

- (a) Polygenecity
 - (b) Polymorphism
 - (c) Antigen specificity
 - (d) Co-dominance
 - (e) Linkage disequilibrium
-

(33) The most significant reason for TCR diversity is:

- (a) Recombination of genes that involves addition and loss of random nucleotides
 - (b) Combinatorial Diversity
 - (c) Large number of genes
 - (d) Somatic hypermutation
-

(34) All of the following are true regarding superantigens **EXCEPT**:

- (a) Does not interact with MHC
 - (b) Escapes antigen processing and presentation
 - (c) Stimulates a large group of lymphocytes at once
 - (d) Binding to the variable β chain
-

(35) Which of the following regarding T cell selection is true:

- (a) Cells that do not recognize peptides by MHC and those that recognize peptides with high avidity have the same fate
 - (b) Double negative cells undergo positive selection
 - (c) T cells can wait indefinitely for a positive selection signal
 - (d) Negative selection occurs for double negative cells and positive selection occurs for double positive cells
-

(36) All of the following matches are correct **EXCEPT**:

- (a) TAP : MHC class I
 - (b) CLIP: MHC class II
 - (c) Proteasome : antigen degradation
 - (d) Tapasin: $\alpha:\beta_2$ complex attach to TAP dimer
 - (e) Calreticulin : MHC class II
-

(37) All of the following are functions of the Fab fragment **EXCEPT**:

- (a) Detect an antigen
 - (b) Precipitate an antigen
 - (c) Induce inflammation
 - (d) Block the active site of toxins and other pathogen associated molecules
 - (e) Block the interaction between the host and pathogens
-

(38) Phosphorylated ITAM activates ZAP-70 in B cell transduction pathway (question wants the **wrong** answer)

(39) B cells originate from the bone marrow with IgD expression (question wants the **wrong** answer)

(40) No cross over occurs in MHC genes (question wants the **wrong** answer)

Answers

1. E
2. C
3. B*
4. C
5. D
6. C*
7. B
8. C
9. D
10. C
11. A
12. C
13. D
14. C
15. A
16. C
17. D
18. C
19. C
20. B
21. E
22. C
23. E
24. B
25. D
26. A
27. C
28. B
29. B
30. E
31. C
32. C
33. A
34. D*
35. A
36. E
37. C

Check the ones marked *