VNTR mean and application..

point mutation. long questionn: chromosomal non disjunction
types and causes..

and how mutation change alter function?
Define point mutation. 2 marks
Define epigenetics? 2 marks
Define reverse, forward and neutral mutation. 3 marks Write
any three mitocondrial mutation deasease. 3 marks
Frameshift mutation. 5 marks.
Write any five basic complication in pattern of inheritance. 5 marks:

What is chromosomal abberation?how it is classified? 10 marks
How mutations alters the gene function?10 mrks
Intronic position with respect to exon(2)
Epigenetic definition (2)
Point mutation and it's classifications (5)
Chromosomal aberration with type (5)
How chromosomal aberation change gene (10)
VNTR ? Application (5)
Fameshift mutations: 2marks
Define epigentics:2marks
3 disorders of Auntosomal dominant disorders: 3marks
Forward, reverse, and neutral mutations: 3marks
Point mutations and enlist 3 structural changes in chromosomes:5marks
5 complications in inheritance:5marks
Modification of epigenetica:10marks
Mutation alter genes expression:10marks
1:Write the function of STR?2 mrks
2:define epigenetics?2 marks
3:write the name of the three genetic disorders? 3 marks
4:How epigenetics occur?3 marks
5:what is the point mutation?enlist the structural mutations?5 mrks
6:what is VNTR?writes its five applications? 5 mrks
7:what is chromosomal abbreastion?how it is classified?10 mrks
8:how mutations alters the gene function?10 mrks
Q1: What is the function of Short Tandem Repeats Polymorphism? 2 marks
Q2: Define Epigenetics? 2 marks
Q3: What are the three main causes for epigenetics? 3 marks
Q4: Give three genetic disorders? 3 marks

Q5: What is chromosomal aberration? Write at least three variations caused due to change in chromosomal structure? 5 marks

Q6:Enlist 5 complications caused due to the inheritance pattern? 5 marks

Q7: What is point mutation? How mutation alter gene function? 10 marks

Q8: Explain Chromosomal non disjunction, its types and cause? 10 marks

(For MCQs prepare ppts very well. 30 out of 40 were exactly from there, other 10 were also from ppts but they were asked in a conceptual way). Non disjunctio of chromosomes?types?causes?10 marks Mutations ? Hoe can they alter the function of a gene ? 10 marks Single nucleotides polymorphism ?

5 complexities of inherited gene?

Mcqs were very confusing .. overall ..whole paper was confusing

1.Write the modification of epi genetics ? 10

2.How mutation alter gene function ? 10

SHORT QUESTION

3.Define epigenetics ? 2

4.Write the common mitochondrial diseases ? 2

5.What is VNTR ? write their 5 application. ? 3

6.What is point mutation and how change in chromosome structure? 3

7.Write rules of mutation ? 3 Epigenetic?

Cause of epigenetic?

Linker and causative SNP?

Point mutation?

Chromosomal aberration 10 marks?

Baqi bhool gya....

Mcq zyada tr mutation waly topic se thy

1.Write the modification of epi genetics ? 10

2.How mutation alter gene function ? 10

SHORT QUESTION

3.Define epigenetics ? 2

4.Write the common mitochondrial diseases ? 2

5.What is VNTR ? write their 5 application. ? 3

6.What is point mutation and how change in chromosome structure? 3

7.Write rules of mutation ? 3

Aik question or thaw o yaad nai h.... time 2 hrs

MCQ"S FROM PPTS.

1.epigenetic modification.10 marks

2- how mutation alter gene function.10 marks

3- define epigentics.2marks

4- enlist three common autosomal dominant disorders.3 marks

5- diffrentiate between farword, reverse and neutral mutations. 3 marks

6- what is point mutation. enlist three variation that occur due to change in chromosomal structure. 5 marks

7- five complications in inheritence pattern.5 marks

ESSENTIAL OF GENETICS 25 FEB 2017 4:30

SUBJECTIVE 40 MARKS

1.Write the modification of epi genetics ? 10

2.How mutation alter gene function ? 10

SHORT QUESTION

3.Define epigenetics ? 2

4.Write the common mitochondrial diseases ? 2

5.What is VNTR ? write their 5 application. ? 3

6.What is point mutation and how change in chromosome structure? 3

7.Write rules of mutation ? 3

Aik question or thaw o yaad nai h…. time 2 hrs

MCQ"S FROM PPTS.

Difference b/w chiasmata & cross over?

Meiosis I nd II?

Prophase I?

Stages of prophase I?

Which cell division occurs in somatic cells?

Oogenisis and spermatogenisis? and

mcqs was from ppt slides totally

define karyotype idiotype,,linkage inheritance , rule organizes my international committe , 3 genetic disorders , single gene and multifactorial disorders , vntr aflp ,,inheretance ,dominant allele , frequency of dominant alllele, ,

5 basics which cause difficulty in inheretance pattern and all the mcqs were from starting topics

301 Paper 4-03-16

*Diff b/w forward and reverse genetics

*Reason of drawing pedigree

*what is frequency range of autosomal dominant disorder?

*Diff b/w RAPD and AFLP

*Location of intronic sequence with respect to exons

*Rule of mutation nomenclature

*3 mitochondrial diseases

*3 autosomal dominant disorder

*Types of genetic disorders

LONG OF 5 MARKS

*Five complications in inheritance pattern
*what is VNTR? Write five applications of VNTR.
50% objective from first 20 and 50% from 100 to 125

1 Comment

301 Paper 3-03-16
*Difference between linked and causative SNP
*Diff b/w single gene and multifactorial gene
*Diff b/w forward and reverse genetics
*Function of STRP
*Reason of drawing pedigree
*Diff b/w centromere and telomere
*what is frequency range of autosomal dominant disorder?
*Diff b/w RAPD and AFLP
*Diff b/w meiosis I and II
*Location of intronic sequence with respect to exons
*Rule of mutation nomenclature
*3 mitochondrial diseases
*3 autosomal dominant disorder
*Types of genetic disorders
LONG OF 5 MARKS
*Five complications in inheritance pattern *what is VNTR? Write five applications of VNTR. types of poymorphisims.,VNTR function ,locus,mitochondrial disorder,autosomal dominant recesseive disorder Some d!ffr ques of b!03o1
Def!ne nd expla!n m!tot!c appratus?
Why h!gh gene dens!ty !n parokaryouts?

ANS of BIO301 Paper

types of poymorphisims.

Types of Polymorphic DNA Sequences

- RFLP: Restriction Fragment Length Polymorphism
- VNTR: Variable Number Tandem Repeats (8 to > 50 base pairs)

Types of Polymorphic DNA Sequences

- STR: Short Tandem Repeats (1– 8 base pairs).
- SNP: Single Nucleotide Polymorphisms
-

AFLP: Amplified Fragment Length Polymorphism,

,locus

locus (plural loci), in genetics, is the specific location or position of agene, DNA sequence, on a chromosome. Each chromosome carries many genes; humans' estimated 'haploid' protein coding genes are 20,000-25,000, on the 23 different chromosomes. A variant of the similar DNA sequence located at a given locus is called an allele. The ordered list of loci known for a particular genome is called a gene map. Gene mapping is the process of determining the locus for a particular biological trait. ,mitochondrial disorder

Common mitochondrial diseases

- Myoclonic epilepsy
- Mitochondrial recessive ataxia
- Leber hereditary optic neuropathy
- Sensory ataxia neuropathy

,autosomal dominant disorder

Autosomal Dominant diseases

- Marfan syndrome
- Huntington's disease
- Retinoblastoma
- Waardenburg syndrome
- Myotonic dystrophy
- Polycystic kidney disease
- Achondroplasia
- Polydactyly
- Heredity hearing loss

recesseive disorder

Common recessive disorders

- Bloom syndrome
- Carpenter syndrome
- Cystic fibrosis
- Thalassemia
- Many forms of mental retardation
- Gaucher's disease
- Glycogen storage diseases
- Rotor syndrome
- Many of eye diseases

Classification of genetic disorders

- Multifactorial disorders
- Single gene disorders
- Chromosomal disorders
- Mitochondrial disorders

Somatic mutations (cancers)

Karyotype

A karyotype is the number and appearance of chromosomes in the nucleus of a eukaryotic cell. The term is also used for the complete set of chromosomes in a species, or an individual organism

idiotype

He defined idiotype as the set of epitopes on the V region of an antibody molecule, where epitope means an antigenic determinant. He also defined the "paratope" to be that part of an antibody variable region that binds to an antigen.

Single gene can give codominance, incomplete dominance, overdominance

Genetic maps have been used successfully to find the single gene responsible inherited disorders, like cystic fibrosis and muscular dystrophy.

Multifactorial diseases

- Asthma
- Cancers
- Ciliopathies
- Cleft palate
- Diabetes Heart diseases
- hypertension inflammatory
- bowel disease
- intellectual disability
- mood disorder
- Obesity
- infertility

Frequency of genetic disorders

Frequency of autosomal dominant disorders

Frequency of autosomal dominant disorders varies between 3 – 9.5 per 1000 individuals

Frequency of autosomal recessive disorders

Frequency of autosomal recessive disorders is between 2-2.5 per 1000 individuals.

Frequency of x-linked disorders

Frequency of X-linked disorders is between 0.5 – 2 per 1000 individuals.

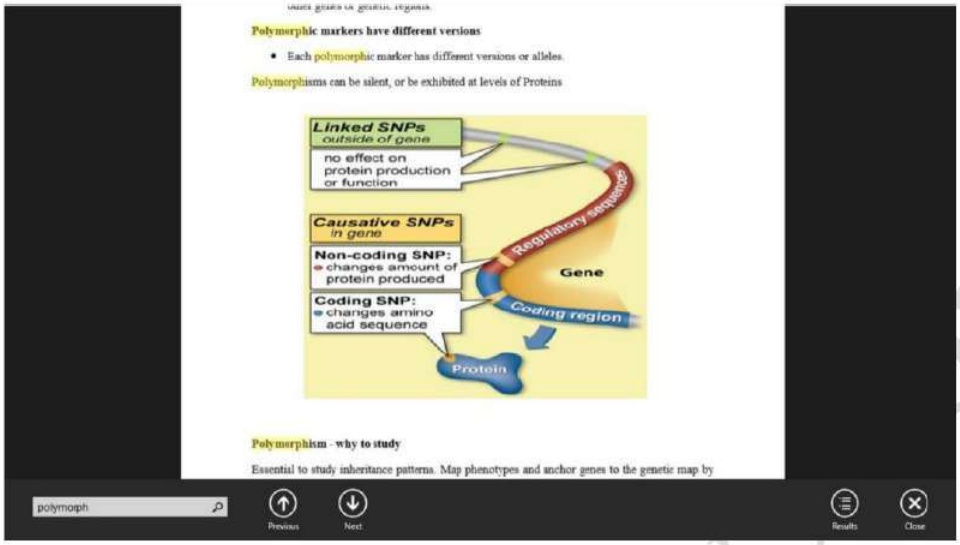
Frequency of chromosomal disorders

Frequency of chromosomal disorders is between 6-9 per 1000 individuals.

Frequency of multifactorial disorders

Frequency of multifactorial disorders is between 20 – 50 per 1000 individuals.

Most common multifactorial disorders are cancers, diabetes, heart diseases.



What is RFLP: RFLP is a difference in homologous DNA sequences that can be detected as fragments of different lengths after digestion of the DNA samples.

Most RFLP markers are heterozygous and highly locus-specific.
RFLP probe is a labeled DNA sequence that hybridizes with one or more fragments of the digested DNA sample after they were separated by gel electrophoresis.

VNTR

- VNTR are short nucleotide sequences organized as a tandem repeats on genomes.
- Found on many chromosomes
- Show variations in length between individuals.

Size of VNTR

The size of repeat is about 7 to few tens of base pairs long.

Each variant acts as inherited allele

Each variant acts as an inherited allele, allowing them to be used for personal or parental identification

Importance of VNTR

VNTRs are an important source of RFLP genetic markers used in linkage analysis (mapping) of genomes. When removed from surrounding DNA by the PCR or RFLP methods, and their size determined by gel electrophoresis or Southern blotting. As a result, they produce a pattern of bands unique to each individual.

VNTR function

Applications of VNTR

- Microbiology
- DNA fingerprinting
- Genetic diversity
- Forensics
- Mapping of genomes
- Breeding patterns of wild or domesticated animals

Mutation nomenclature

All variants should be described at the most basic level, i.e. DNA level.

- Descriptions should always be in relation to a reference sequence, either a genomic or a coding DNA sequence.
- Although theoretically a genomic reference sequence seems best, in practice a coding DNA reference sequence is preferred.
- Good for promoters, alternative splicing etc.
- When the entire genomic sequence is not known, a cDNA reference sequence should be used.
- To avoid confusion in the description of a variant it should be preceded by a letter indicating the type of reference sequence used.
- Several different reference sequences can be used.
- "c." for a coding DNA sequence (like c.76A>T)
- "g." for a genomic sequence (like g.476A>T)
- "m." for a mitochondrial sequence (like m.8993T>C,)
- "r." for an RNA sequence (like r.76a>u)
- "p." for a protein sequence (like p.Lys76Asn)

Describing genes / proteins, only official HGNC (HUGO gene nomenclature committee) gene symbols should be used (www.genenames.org)

DNA reference sequence used should preferably be from the RefSeq database, listing both database accession and version number (like NM_004006.2)

Type of Chromosomes based on Centromere positioN Chromosomes may differ in the position of the centromere.

Centromere is near the middle, metacentric

Centromere is toward one end, acrocentric or sub metacentric

Centromere is very near to end, telocentric pedigree

A complete, detailed family history is called a pedigree. Pedigrees are used to determine the pattern of inheritance of a genetic disease within a family.

Generations of a pedigree as roman numerals
Generations in a pedigree diagram are numbered, by using Roman numerals, starting with the parental generation, at the top of the diagram as generation I.

Members of each generation are numbers from left to right

For convenience, the members of each generation are numbered across the line, from left to right, using normal numerals

Difference B/W Centromere and Telomere?

A centromere is a cellular structure involved in meiosis that creates spindle fibers and allows chromosomes to separate in cells. Telomeres are single-stranded DNA tails that form at the end of replicated DNA strands because they cannot be replicated. They are nonsense DNA, so nothing is lost in not replicating them. They protect exons--expressed DNA segments, and work in the cell aging process.

How mutation alter gene function? 10 marks
What are point mutations and described three chromosomal structure changes? 5 marks..?STRP function?
Define point mutation. 2 marks
Define epigenetics? 2 marks
Define reverse, forward and neutral mutation. 3 marksFrameshift mutation?

Forward, reverse, and neutral mutations?3 genetics disorder

Long questions
Complications to inheritance patterns
Mutation alter gene function
Point mutation and variation structural changes
Non disjunction chromosomes types and causes
Short
Define epigenetics and causes linked
and causative SNP
Frequency of autosomal dominant disorder frameshift
Mcqs mostly mutation k topic me sy thy

describe complications to inheritance pattern?
How Mutation alter gene function?
what is Point mutation and how variation in structural changes accur? Non disjunction ? Types of chromosomes ?and causes?
#Short:
Define epigenetics and causes? what linked and causative SNP? what is Frequency of autosomal dominant disorder? define frameshift?

Splicing pathway... CRISTR STANDS FOR??? termination in short... Rna processing .. rna has short life span so how we can analyzed through what process fr elongation short

301 genetics,,,,,, 11.00 am enlist 3 genetics disoders.point mutatin,chromosoml

aberration and cllsfcation., how mutation chnge gene function.

definition of forwrd ,reversd and neutral muation,complications of inheritance pattern,intronic location,epigenetic define, difference b/w single gene disorders and multifactorial disorders. 2m what is the location of intronic sequences wd respect to exons? 2m define epigenetics. 2m diffrentiate b/w RAPD and AFLP. 3m differentiate b/w forward, reverse and neutral mutation. 3m wt r basic 5 complications in inheritance pattern. 5m
wt point mutation? enlist at least 3 variations that occur due to change in the structure of chromosome? 5m
write a note on epigenetic modification. 10m how
mutations alter gene function ? 10m

ByBio (301) today paper
How mutation alter gene function? 10 marks
What are point mutations and described three chromosomal structure changes? 5 marks..?STRP function?
Bio 301. 3 march, 2017
1: Define point mutation. 2 marks

2: Define epigenetics? 2 marks

3: Define reverse, forward and neutral mutation. 3 marksFrameshift mutation?

Forward, reverse, and neutral mutations?3 genetics disorder? Almost all q past paper se thee?

bio 301 final term 11AM

12 aug

36 MCQs

37.....difference b/w single gene disorders and multifactorial disorders. 2m

38.... what is the location of intronic sequences wd respect to exons? 2m 39....define epigenetics. 2m

40.... diffrentiate b/w RAPD and AFLP. 3m

41...differentiate b/w forward, reverse and neutral mutation. 3m

42....wt r basic 5 complications in inheritance pattern. 5m

43.... wt point mutation? enlist at least 3 variations that occur due to change in the structure of chromosome? 5m

44... write a note on epigenetic modification. 10m

45..... how mutations alter gene function ? 10m

- My today paper bio 301 at 11am

  Long questions

  1. Complications to inheritance patterns
  2. Mutation alter gene function
  3. Point mutation and variation structural changes
  4. Non disjunction chromosomes types and causes

  Short

  Define epigenetics and causes

  3.linked and causative SNP

  4. Frequency of autosomal dominant disorder

  5.frameshift

  Mcqs mostly mutation
- topic me sy thy....
- Quiz Bio 301

  Q: 1

  Mutation which type a mutant phenotype back into the wild type.

  Ans: Reverse mutation

  Q: 2

  A genetic disease or disorder appears as a result of mutations in an individual's _____.

  Ans: DNA

  Q: 3

  Hunter's disease is an example of_____.

Ans: X-linked recessive disorder

Q: 4

A condition in which some people with an appropriate genotype fail to express the phenotype is known as the _____.

Ans: incomplete penetrance

Q: 5

Which one of the following is not influenced by the silent mutation?

Ans: Phenotype

Q: 6

Organism that exhibit suppressor mutation is/ are_____

Ans: Double mutant

Q: 7

Mutation that produce the change in the amino acid in protein product (histidine in for arginine)

Ans: Missense mutation

Q: 8

Base analog have the similar structure with_____

Ans: standard bases of the DNA

Q: 9

Mutation which convert a DNA sequence into the stop codon.

Ans: Non sense mutation

Q: 10

The term Ashkenazi refers to the specific people of following population. Ans: Jew population

- Bio 301paper
- Bio 301 ka zaid past ..ma se tha...vNTR or us ke application?2)epigenetics modification?genetics disorder?SNTR function,? mutation alter gene function? Farwad reverse and natural mutation? freamshift mutation define?point mution or sath ma change chromosome structure pouch tha,?epigenetics define?
- subbject: bio 301

Timing: 11am, 12/08/2017

#final term #paper.

#long

1) describe complications to inheritance pattern?
2) . How Mutation alter gene function?
3) what is Point mutation and how variation in structural changes accur?
4) Non disjunction ? Types of chromosomes ?and causes?

#Short:

1) Define epigenetics and causes?
2) what linked and causative SNP?
3) what is Frequency of autosomal dominant disorder?
4) define frameshift?

- 301 Genetics ...timming 11.00 (final term)Enlist 3 genetic disorders.Point mutation.structural muation of chromosomes.Chromosomal abetration and classfy them.How mutation change the gene function.5 basic complications of inheritance pattern.Define :Forward.reversed and neutral muation.

301 genetics,,,,,, 11.00 am enlist 3 genetics disoders.point mutatin,chromosoml aberration and cllsfcation.,how mutation clnge gene function. definition of forwrd ,reversd and neutral muation,complications of inheritance pattern,intronic location,epigenetic define,?

Bio301 final term paper 12-8-17 at 8:00 am

What is Choromosomal non-disjunction types, causes. 10
Mutations alter gene function 10
VNTR and its applications 5
Point mutation 5
Causes of epigenetics 3
Write the name of 3 autosomal resesive disorders 3
Dif. b/w multifactorial and singal gene mutation 2
What is framshift mutation 2

Bio 301 paper.....