Virtual University of Pakistan

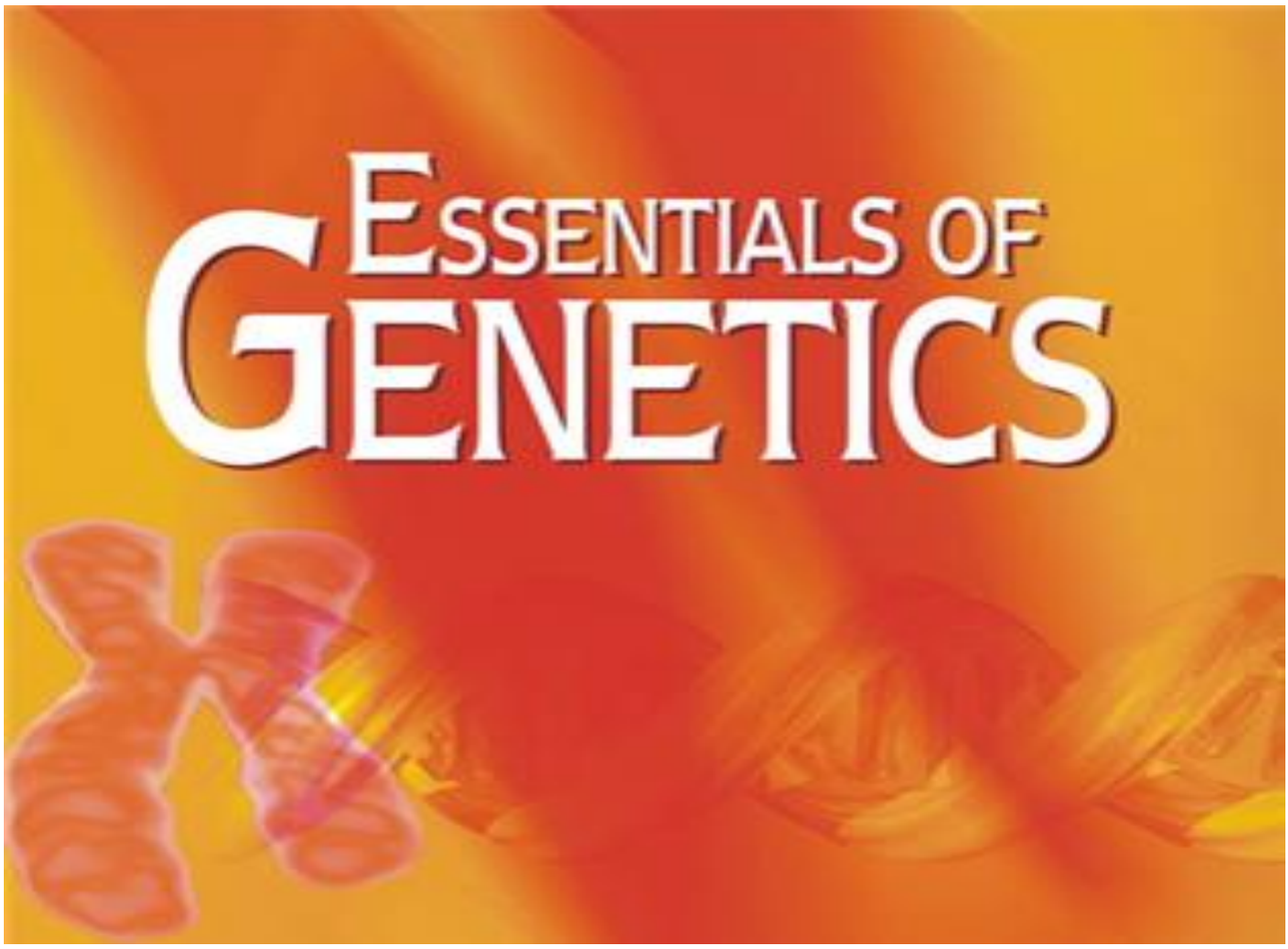
Federal Government University

Past -quiz /grand quiz/ mid term / final term /
Solved paper

World-Class Education at Your Doorstep

MSC-ZOOLOGY

BIO 301-Essentials of Genetics



PAST PAPERS

MUHAMMAD IMRAN

MUHAMMAD IMRAN	BIO301-Essentials of Genetics
PAST MCQs	ALL IN ONE FILE

QUIZ NO 1	QUIZ NO 2	GRAND QUIZ	MID TERM	FINAL TERM

MCQ

Mutiple Choice Questions

Virtual University

1. THE TYPE OF CELL DIVISION IMPORTANT FOR GROWTH IS _____**MITOSIS**
2. THE REGION WHERE TWO SISTER CHROMATIDS OF A CHROMOSOME JOINED OR “HELD TOGETHER” DURING MITOTIC METAPHASE IS CALLED_____**CENTROMERE**
3. .IN HUMANS…. OF SOMATIC CELLS. _____, **~ 220 TYPES**
4. WHICH THE FOLLOWING STATEMENTS IS TRUE ABOUT HOMOLOGOUS CHROMOSOMES IN HUMAN..._____**MALE CELLS HAVE 22 PAIRS OF HOMOLOGOUS AND ONE PAIR OF NON-HOMOLOGOUS CHROMOSOMES.**
5. HOW MANY NUMBERS OF GENES ARE PRESENT IN YEAST?.._____**5,800**
6. NUCLEAR DNA REPLICATE IN THE _____**S PHASE.**
7. G0 PHASE IS_____ **RESTING PHASE**.
8. DURING THE SUMMER, THE ARCTIC FOX PRODUCES ENZYMES THAT CAUSE ITS FUR TO BECOME REDDISH BROWN. THESE ENZYMES DO NOT FUNCTION DURING THE WINTER, CAUSING THE FOX'S FUR TO BECOME WHITE AND BLEND IN WITH THE SNOW.THE CHANGE IN THE FUR COLOR SHOWS THAT.....**THE EXPRESSION OF CERTAIN GENES IS AFFECTED BY TEMPERATURE.**
9. GROUP OF ADJACENT NUCLEOTIDES ARE JOINED BY__**A PHOSPHODIESTER BOND.**
10. KINETOCHORES ARE FORMED AT_____**CENTROMERES**.
11. WHICH TYPE OF GENETICS DEALS WITH MOVEMENT OF GENES AND GENETIC TRAITS FROM PARENTS TO OFFSPRING?._____.**TRANSMISSION GENETICS.**
12. CHROMOSOME NUCHROMOSOMEMBER CAN BE COUNTED EASILY ONLY DURING.. _____**MITOTIC METAPHASE**.
13. WHICH OF THE FOLLOWING IS SHOW THE PLEITROPIC IN HUMAN?..**ALL OF THE GIVEN.**
14. MEIOSIS RESULT IN..**.4 HAPLOID DAUGHTER CELLS.**
15. IN LIFE FORMS WITH THE ZZ-ZW CHROMOSOMAL SEX DETERMINATION,THE MALES HAVE.....**HOMOGAMETIC**
16. GIANT CHROMOSOME FIRST DISCOVERED BY …IN 1881 IN DIPTERAN SALIVARY GLANDS AND THUS ALSO KNOWN AS SALIVARY GLAND. _____ **BALBIANI**
17. A CELL WITH 10 CHROMOSOMES UNDERGOES MITOSIS. HOW MANY DAUGHTER CELLS ARE CREATED? ___
18. EACH DAUGHTER CELL HAS ___ CHROMOSOMES..**.2, 10**
19. LAMBRUSH CHROMOSOMES ARE FOUND IN._____..**ALL OF THE MENTIONED**
20. WHICH OF THE FOLLOWING ENZYMES REMOVE HELIX AND PRODUCES A NICK TO UNCOILED DNA…. **GYRASE AND TOPIOSOMERASE.**
21. DARKER BANDS ARE GENERALLY FOUND NEAR THE CENTROMERE OR ON TH ENDS TELOMERES OF THE CHROMOSOME.THE POSITION OF THE DARK STAINING ARE ….OR…. _____**HETROCHROMATIC REGION , HETROCHROMATIN.**
22. VARIATION ARE ENTIRELY GENETICALLY CONTROLLED …..**DISCONTINOUS**.
23. IN HUMANS , A ZYGOTE IS A---- CELL HAVING----- CHROMOSOMES. ____**DIPLOID 46**.
24. WHEN ---- ARE REMOVED DUE TO CHROMOSOME BREAKAGE , THE DAMAGED CHROMOSOME ENDS CAN READILY FUSE OR UNITE WITH BROKEN ENDS OF OTHER CHROMOSOME**TELOMERE**.

25. ----- PROVIDE TERMINAL STABILITY TO THE CHROMOSOME AND ENSURE ITS SURVIVAL…._____.**TELOMERE**
26. DURING METAPHASE MITOSIS CHROMOSOME…_____ **LINE UP AT THE EQUATOR**
27. HOW MANY NUCLEAR DIVISIONS DOES A CELL GO THROUGH FOR MITOSIS? HOW MANY TIMES DOES NUCLEUS DIVIDE_____....**1**
28. CELL SPLITS INTO TWO…_____**CYTOKINESIS**
29. IN LIFE FORMS WITH THE ZZ ZW CHROMOSOMAL SEX DETERMINATION , THE FEMALES ARE…._____ **HETROGAMETIC**.
30. WHO IS CONSIDERED THE FATHER OF GENETICS?...___**GREGOR JOHN MENDEL**.
31. WHICH OF THE FOLLOWING STATEMENT IS NOT CORRECT ABOUT INCOMPLETE DOMINANCE…._____.**ONE OF THE ALLELE IS DOMINANT.**
32. DURING PROPHASE , THE NUCLEAR ENVELOPE…_____.**DISAPPEAR**
33. COAT COLOUR IN LABRADOR RETRIEVER IS CONTROLLED BY EPISTASIS. B SPECIFIES BLACK PIGMENT, B IS FOR BROWN PIGMENT (RECESSIVE) AND E LOCUS DETERMINES THE EXPRESSION OF B LOCUS. WHICH ONE OF THE FOLLOWING IS WRONG? _____**A) BB EE GIVES YELLOW**
34. WHICH OF THE FOLLOWING IS SHOW POLYGENIC PATTERN OF INHERITANCE… _____**ALL OF THE GIVEN**
35. WHICH ONE IS FIRST OBSERVED BY FLEMMING 1882? _____**LAMPBRUSH CHROMOSOME**
36. WHEN DNA POLYMERASE IS IN CONTACT WITH GUANINE IN THE PARENTAL STRAND, WHAT DOES IT ADD TO THE GROWING DAUGHTER STRAND …._____**CYTOCIN**.
37. IDENTIFY THE PURINE BASE OF NUCLEIC ACID…_____ **ADENINE**
38. IN AN EXPERIMENT YOU ATTACH AN ICE PACK ON THE CHEST OF THE HIMALAYON RABBIT FOR SEVERAL DAYS.WHAT WILL YOU OBSERVE…._____**THE COAT COLOR OF THE AREA WILL BE DARKER.**
39. WHICH OF THE FOLLOWING STATEMENT IS NOT CORRECT ABOUT GENOME CHARACTERISTICS OF PROKARYOTES_____….**PROKARYOTES HAVE LOWER GENE DENSITY AS COMPARED TO EUKARYOTES.**
40. THE GIANT CHROMOSOMES FOUND IN … AND THEY MAY BE AS LONG AS 300 µ AND UP TO 10 µ IN DIAMETER…_____**DIPTERAL**
41. WHICH OF THE FOLLOWING STATEMENT IS NOT CORRECT ABOUT MEIOSIS 1…._ _____**NONE OF THE MENTIONED ABOVE.**
42. THE DIAMETER OF CONDENSED SCAFFOLD ASSOCIATED CHROMATIN IS…_____- **30NM**
43. WHICH OF THE FOLLOWING STATEMENT IS NOT CORRECT ABOUT CONTINOUS VARIATION?... _____**CONTINUOUS VARIATION HAS DISTINCT CATEGORIES.**
44. A HEREDITY UNIT ON CHROMOSOMES IS CALLED …_____ **GENE**.
45. WHICH OF THE FOLLOWING DOES NOT REVERT TO THE EUCHROMATIN STAGE_____…_____**CONSTITUTIVE HETROCHROMATIN.**
46. CHROMATIN FIBERS UNDERGO FOLDING, COILING AND SUPERCOILING DURING --- SO THAT THE CHROMOSOMES BECOME PROGRESSIVELY THICKER AND SMALLER…. _____**PROPHASE**
47. WHEN CHROMOSOMES STAINED THEY SHOW A DARK-STAINED REGION THAT IS THE…. _____**CENTROMERE**
48. SEX DETERMINATION IN INSECTS IS BASED ON._**XX XO CHROMOSOMAL MODEL**
49. SPINDLES MICROTUBULES ATTACH TO KINETOCHORES SPINDLE MICROTUBULES ATTACH TO THE KINETOCHORES OF CHROMOSOMES AND MOVE THE CHROMOSOMES TO THE…_____ **METAPHASE PLATE**.
50. THE SISTER CHROMATIDS SEPARATE DURING ANAPHASE STAGE OF…._____ **MEIOSIS 2 AND MITOSIS.**
51. IN THE PARENTAL GENERATION , WHEN MENDEL CROSSED A TRUE BREEDING PEA STRAIN THAT PRODUCED SMOOTH SEEDS__ **ALL F1 PROGENY SEEDS WESE SMOOTH.**
52. ….. IS AN EXAMPLE OF INHERITED VARIATION…_____**FINGER PRINT**.
53. MITOTIC APPARATUS HELPS IN …._____ **CHROMOSOMAL MOVEMENT.**
54. HOW MANY MOST COMMON CHROMOSOMAL BANDING ARE PRESENT__--_____-… **4**
55. IN DNA DOUBLE HELIX , THE TWO DNA CHAINSARE HELD TOGETHER BY_____- _____…. **HYDROGEN BONDS.**

56. XO SYSTEM OF SEX IS PRESENT IN …_____ **GRASSHOPPER**.
57. ----- PUSH AGAINST EACH OTHER,THAT STRETCH THE CELL INTO AN OVAL SHAPE……_____ **NON KINETOCHORE SPINDLE FIBERS.**
58. THE DIAGRAM WHICH SHOWS THE ARRANGEMENT OF METAPHASIC CHROMOSOMES ACCORDING TO THEIR POSITION IS CALLED…_____- **IOEGRAM**.
59. GENETICS IS THE STUDY OF …_____**ALL OF THE MENTIONED.**
60. WHICH OF THE FOLLOWING HAVE CHROMOSOME NUMBER FROM 8 TO 16 AND THEIR MOST COMMON NUMBER IS…_____.**ANGIOSPERM**
61. WHICH OF THE FOLLOWING IS NOT AN EXAMPLE OF GENETIC CAUSES OF VARIATION…_____ **NUTRIENT**.
62. BANDING PATTERN OF CHROMOSOME USEDVARIETY OF TECHNIQUES AND STAIN THE GC RICH DNA…_____-. **R BANDING**.
63. FOR HIGH RESOLUTION CHROMOSOMES ANALYSIS ,WHICH STAGE OF THE CELL CYCLE ARE ARRESTED BEFORE STAINING…_____- **METAPHASE**.
64. THE DIAMETER OF DNA DOUBLE HELIX IS …_____-.**2NM**
65. WHICH OF THE FOLLOWING STATEMENT IS NOT CORRECT ABOUT THE HEREDITARY VARIATION CONCEPTS FROM MENDELS WORK…_____. **THERE WAS NOT A CLEAR INDICATION OF HEREDITY AND VARIATIONS IN HIS WORK.**
66. IN 14% OF THE NUCLEOTIDES FROM DNA NUCLEOTIDE CONTAINS THE BASE T WHAT % WILL CONTAIN THE BASE G…_____.**36%**
67. THE DIAMETER OF PACKED NUCLEOSOME…_____**…11NM**
68. HISTONES HAVE TWO BASIC AMINO ACIDS_____ **ARGININE AND LYSINE**
69. WHAT IS THE OTHER NAME OF A POLYTENE CHROMOSOME?....._____ **SALIVARY GLANDS CHROMOSOME**.
70. MEIOSIS IN MALES IS CALLED…_____.. **SPERMATOGENSIS**
71. GENETICS OR RESEARCH CANSTUDY THE GENOTYPE BASED ON THE PHENOTYPE IN CLASSIC GENETICS THROUGH…_____.**GENE KNOCKOUT**
72. CHROMOSOMES WERE DESCRIBED BY STRAUSBERGER IN 1875. CHROMOSOME WAS FIRST USED BY_____ **WALDEYER IN 1888**.
73. VATRIATION IS A DIFFERENCE THAT CAN BE SEEN IN INDIVIDUAL OF THE SAME SPECIE…_____-_____.. **IT CAN BE DUE TO GENETIC MAKEUP, ENVIRONMENT OR BY COMBINATION OF BOTH**.
74. WHICH OF THE FOLLOWING STATEMENT ABOUT EUKARYOTIC CHROMOSOME IS WRONG_____**….EUKARYOTIC CHROMOSOME IS FOUND IN CYTOPLASM**.
75. --- ARE PROTEINS USED TO PACKAGE DNA…_____.**HISTONES**.
76. WHICH OF THE FOLLOWING HAS HIGHEST NUMBER OF CHROMOSOM__**HORSETAIL**
77. HOW MANY NUM OF BANDS CAN BE RECOGNIZED ON GIANT CHROMOSOME OF THE DROSOPHILA…_____ **5000**
78. CHROMOSOME IDENTIFY BY…_____ **ALL OF THE GIVEN.**
79. IS AN EXAMPLE OF ACQUIRED VARIATION …_____. **ATHLETIC SKILLS.**
80. WHICH OF THE FOLLOWING STATEMENT ABOUT PROKARYOTIC CHROMOSOME IS WRONG…____. **PROKARYOTIC CHROMOSOME MADE OF DNA AND HISTONES**
81. CARGAPHS RULES FOR DNA BASE PAIRING INDICATES THAT …_____..**A+T =G+C**
82. THE LONGEST STAGE IN THE CELL CYCLE IS …_____**INTERPHASE**
83. CHROMOSOME HAVING CENTROMERE AT ONE END OF ARMS _____ **TELOCENTRIC**
84. HAVING THE SAME LENGTH, SHAPE, LOCATION AND GENES…_____..**AUTOSOMES**.
85. SKIN COLOR PIGMENTATION IN HUMANS IS CONTROLLED BY AT LEAST …_____..**THREE GENES**.
86. HOW MANY HOURS DOES THE G1 PHASE OF INTERPHAE TAKE TO COMPLETE A CYCLE_____….**6 TO 12**
87. HISTORICALLY FIRSTLY DEVELOPED SUB DISCIPLINE OF GENETICS…_____.**TRANSMISSION GENETICS**
88. DICOTS IN GENERAL , HAVE A – OF CHROMOSOME THAN MONOCOTS…_____..**HIGHER LONGER.**
89. WHICH ONE CHARACTERISTICS EXPRESSES ITSELF OVER THE OTHER KNOWN AS…_____. **DOMINANT**.

90. CENTROMERE BREAKS BY THE REMOVAL OF A PROTEIN …_____. **SECURIN**

91. WHICH SUB DISCIPLINE OF THE GENETICS IS CONSIDERED THE HEREDITY OF THE TRAITS IN GROUPS, BUT THE TRAITS OF CONCERNS ARE DETERMINED BY MANY GENES… _____. **QUANTITATIVE GENETICS.**

92. GENERALLY KARYOTYPES REPRESENTED BY ARRANGING IN THE --- ORDER OF THE SIZE KEEPING THEIR CENTROMERE IN A.…_____ **DESCENDING ,STRAIGHT LINE.**

93. WHICH OF THE FOLLOWING ARE USEFULL INDICATORS TO CHECK THE WATER QUALITY_____…**PHYTOPLANKTON**

94. THERE ARE ABOUT --- LOOPS IN THE CHROMOSOME OF E.COLI .THESE ARE HIGHLY TWISTED OR SUPERCOILED STRUCTURE WITH ABOUT ---- NUCLEOTIDE PAIRS…_____..**50 FOUR.**

95. EPISTASIS IS INTERACTION BETWEEN –GENES…_____.**2**

96. STAGE OF PROPHASE IN WHICH NUCLEAR MEMBRANE DISSOLVES…..._____ **STAGE 5 OR 1 STAGE**

97. WHAT TYPE OF SEGREGATION OCCURS WHEN ALLELES AT THE TWO GENE LOCI SEGREGATE TOGETHER AND ARE TRANSMITTED AS A UNIT…_____..**INDEPENDENT SEGREGATION**.

98. WHICH TYPE OF INHERITANCE APPEARS IN AB BLOOD GROUP PHENOTYPE_____ ….**CO DOMINANCE**

99. ACCORDING TO…… GENETIC MATERIAL PARENTS MIXES DURING FERTILIZATION…. _____**BLENDING HYPOTHESIS**.

100. PROPHASE… _____**CHROMOSOME CONDENSE AND BECOME VISIBLE**

101. Primary oocyte produces polar bodies....._____ **2**

102. Which two alleles are called co-dominant one another and are expressed when present?_____**Allele A and Allele i**

103. During recessive epistasis. which color of pigeon is produced by recessive red gene with or without spread?_____**Any color**

104. The DNA between nucleosome is called as linker DNA. Its length is _____**20-60 bp**

105. Mendel determined the segregation of alleles at single gene locus based on _____**Monohybrid cross**

106. Darker bands are generally found near the centromere or on the ends (telomeres) of the chromosome. The position of the dark-staining are or _____ _____**Heterochromatic region, heterochromatin**

107. During metaphase mitosis chromosome _____**Line up at the equator**

108. During stage telophase I chromosomes are..... _____**Haploid**

109. The law of independent assortment with dihybrid cross gives genotypic ratio in F2 as: _____**9:3:3:1**

110. In …. centromere splits into two parts while in….. it does not split. _____**Mitosis, Meiosis I**

111. A male bee sex is _____**Haploid**

112. Shepherd's purse has two shape heart and narrow. If you cross a true breeding heart to a true breeding narrow, F1 will give _____**Narrow**

113. The diameter of condensed scaffold associated chromatin is_____**11 nm**

114. Which of the following is not the part of Interphase in the cell cycle?_____ **M phase**

115. Which of the following is true about phosphodiester linkage?____ **5'-phosphate group of one nucleotide unit is joined to the 3'-hydroxyl group of the next nucleotide**

116. What is the composition of nucleotide**?_____ a base + a sugar + phosphate**

117. Which one of the following statement is correct about human genes?____ **30% genes in humans are di-allelic and they exist in two forms**

118. The diameter of chromatin is_____**11 nm**

119. The diameter of DNA nucleosome on the chromosome scaffold is_____**2 nm**

120. Alleles separate during _____and reunite during _____**Gametogenesis, Fertilization**

121. ____states that the two alleles of a gene found on each of a pair of chromosomes separate independently of one another into sex cells._____ **Mendel's law of segregation**

122. The haploid cells of eukaryotes are usually known as _____**Gametes**

123. Which one of the following is show polygenic pattern of inheritance?_____ **All of the given**

124. How many primary molecular mechanisms of epigenetics are identified?_____ **Two**

125. E.coli has about _____genes are organized into its one circular chromosome____ _____**3000 to 4000**

126. Pepper colour is controlled by _____Two genes
127. ABO blood system is controlled by_ _____A tri-allelic gene
128. Which one of the following statement is not correct about Co-dominance?_____
_____Both alleles are expressed in a homozygous individuals
129. Which one is not regulate gene expression? _____Nucleotide modification
130. Darker bands are generally found near the centromere or on the ends (telomeres) of the chromosome. The position of the dark-staining are _____or
_____Heterochromatic region, heterochromatin
131. In case of two gene interaction, the gene which is masking the expression of another is called
_____Recessive
132. Which of the following is not a common mode of histone modification in eukaryotes?___
_____Sulphonation
133. Mitotic apparatus helps in....._____ Chromosome movement
134. The nucleosome is composed of a core of _____histone proteins and DNA
135. _____bp wrapped around them._____ Eight, 147
136. Name the phenomenon which can break the occurrence of linkage?_____ Crossing over
137. _trait is a stable and heritable phenotype resulting from changes in chromosome without alterations in the DNA sequence_____ Epigenetic
138. Which one of the fur color of rabbit is controlled by maximum numbers of genotypes?_____ Chinchilla
139. Which one of the following statement is not correct about genome characteristics of prokaryotes?____ Prokaryotes have less genome size as compared to eukaryotes
140. Inherited variations result from the_____ Activity of genes
141. Which of the following is not the property of Garden Pea plant that was used by Gregor Mendel for his experiment?_____ Long generation time
142. Which comb shape is appeared when the chicken is homozygous recessive in both the R and
143. P locus?_____ Walnut Combs
144. Which sub discipline of the genetics is considered the heredity of traits in groups of individuals, but the traits of concern are determined by many genes simultaneously?_____ Quantitative genetics
145. Which of the following organism has highest numbers of chromosome?_____ Horsetail
146. In physical map, units of distance are expressed in_____ Base pairs or Mbs
147. When one characteristic expresses itself over the other known as_____ Dominant
148. The trait that does not show through in the first generation is called_____Recessive
149. In____sister chromatids separate and move along the kinetochore microtubules toward opposite ends of the cell._____ Anaphase
150. Building blocks of nucleic acids are _____ Nucleotides
151. In which of the cats the pattern observed in coat colour is due to environmental effect?____
_____ Siamese
152. When one phenotypic trait is determined by multiple alleles then it is called as __Pleiotropic
153. How many numbers of gene are present in Drosophila? _____13700
154. Which is a hereditary disorder characterized by lung congestion, infection and malabsorption of nutrients by the pancreas? _____Huntington disease
155. Which one following histone protein has high molecular weight, greater number of amino acid and high percentage of lysine and arginine?
_____H1
156. Which of the following blood group is considered a universal donor? _____O
157. Which statement is correct about genetic and physical mapping?
_____The some genes are close together on the genetic map and also are close together on the physical map of the chromosome
158. Polygenic traits are also affected by the persons environment, so they are called
_____Multifactorial
159. Which is a histone deacetylase inhibitor? _____ vorinostat
160. Generally, karyotypes represent by arranging in the… order of size keeping their centromere in a….. _____Descending, straight line
161. The spindle apparatus is formed during the phase of mitosis. _____Prophase
162. During, _ the effects of prophase and pro-metaphase are reversed _____Telophase

163. Meiosis results in, _____----_____4 diploid daughter call
164. The allelic series refers to _____Multiple alleles
165. The crossing-over occurs at _____Chiasmata
166. Which one of following statement is not correct about continuous variation?
_____-Continuous variation controlled by a lot of genes
167. have the same length, shape. centromere location. and genes ____Paired chromosomes
168. Mutational changes are….. and epigenetic changes can be….. __Irreversible, Reversible
169. Position dependent method of diseased gene identification follows _____Reverse genetics
170. Which statement is not correct about epigenetic regulation in identical twins?
_____The epigenetic tags can have such an effect on the twins that one can develop a disease and the other is also
171. Which one of the following genotype expresses the Green color of pepper? _____rr/yy
172. Which one of following statement is correct? _____-_Father with X-linked disorder will transmit the mutant allele to all daughters but to no sons
173. Band that indicates constitutive heterochromatin _____C band
174. What is the process of transfer of human blood known as? _____Transfusion
175. All offspring of same variety is called as _____True breed
176. In DNA double helix. the two DNA chains are held together by
_____Hydrogen bonds between the pair of bases
177. if 14% of the nucleotides from a DNA molecule contain the base T. what percent will contain the base G? _____14%
178. According to. . genetic material of the parents mixes during fertilization._____Blending hypothesis
179. Epistasis is the interaction between….. genes _____2
180. On what basis four major blood groups is classified as positive and negative blood type?
_____Rh factor
181. Which one of following statements is correct about to the Mendel Law of Dominance?_____In a cross of parents that are pure for contrasting traits only one form of the trait will appear in the next generation
182. Which one is building blocks of chromosomes? _____Nucleosome
183. Genetists or researchers can study the genotype based on the phenotype in classical/forward genetics through _____DNA sequencing
184. According to particulate hypothesis, parents pass on distinct heritable units. Those distinct heritable units are now called as _____Genes
185. About 90% of the time taken for meiosis is required for _____Prophase 1
186. How much length of the DNA sequence within kinetochores regions? _____120 bp
187. What is the unit of linkage map? _____Centi-morgan
188. Due to… during meiosis |, the two sister chromatids of each chromosome are no longer genetically identical _____Crossing-over
189. DNA replication is a type of. _____ semi-Conservative
190. **70%** genes in human are _____Meno-allelic
191. Which is the cause of epigenetics? _____All of the mentioned
192. Technique that gives an rough idea of gene location _____Genetic mapping
193. A female bee sex is _____Zw
194. Fragile X chromosome affects the gene. _____FMR1
195. Stage of prophase at which nuclear membrane dissolves... _____Stage 2
196. How many numbers of alleles control the fur color of rabbit? _____Four
197. The ABO blood group in humans is determined by _____Multiple alleles
198. Which map is primarily based on the locations of landmarks along a DNA molecule?
_____Physical mapping
199. Genetic make-up of an organism is called _____Genotype
200. The type of cell division important for tissue regeneration is _____Mitosis
201. …occurs when one mutation has the opposite effect when in the presence of another mutation
_____Sign epistasis
202. Which of the following has longer chromosomes. _____ plants
203. Which one of the following is not correct about the influence of environmental factors on epigenetics? _____Social interactions and physical activity not affect the epigenome

204. Name the organism whose first genetic map was made? _____Drosophila
205. Which one of the statement is not correct about the position dependent method of disease causing gene mapping in human? _____Starting at the phenotype. determining which protein was involved and then getting to the gene through the protein
206. Which is the correct sequence of Bottom up mapping technique? _____Make random clones" Sequence each clone" Overlap sequence read Overlap contigs for complete sequence
207. The numbers of genotypes can be calculated on the basis of, _____Numbers of alleles
208. When a cell divides. what do we call the 2 new cells? _____Daughter cells
209. Which statement is not correct about recessive epistasis in pigeons? _____Patter is epistatic to spread color
210. Which is correct about the comb shape in chickens? _____Four different comb shapes is controlled by two genes found on two different chromosomes
211. Chromosomal theory of inheritance states that behavior of chromosomes is according to _____Mendel's law of segregation and independent assortment
212. A nucleosome consists of _____DNA and histones
213. The DNA in a bacterial (prokaryotic) chromosome is best described as _____A single circular double-helical molecule
214. In humans, a zygote is a cell having chromosomes _____Diploid: 46
215. Which trait will produce in F1 generation of the crossing a brown pepper (RRyy) with a yellow pepper (VY)? _____Brown
216. Which one of following is an example of diploid cells? _____All of the mentioned
217. The splitting of sister chromatids occur at _____Anaphase
218. Which are major factors that affect the epigenome? _____Exposure to toxins and diet
219. There are about… loops in the chromosome of E. coli. These loops are highly twisted or supercoiled structure with about….million nucleotide pairs. _____50. four
220. During anaphase of mitosis. push against each other. that stretches the cell into an oval shape. _____Non-kinetochore spindle fibers
221. Differential expression of the genetic material depending on its parentage of inheritance gives. _____ expressivity
222. Nuclear DNA replicates in the phase. _____s phase
223. Which one of following statement is correct? _____Carrier female mates normal male, 50% chance that each daughter will be carrier; 50% chance that each son will have the disorder
224. Which of following blood groups are consists of two different genotypes as homozygous and heterozygous? _____A&B
225. The molecular weight of the histone protein H4 is_____11236
226. occurs when two deleterious genes are beneficial when together ___Reciprocal epistasis
227. The sugar molecule in a nucleotide is_____Pentose
228. Which one of following is show the pleiotropic effects in humans? _____All of the given
229. Which of the following is not correct when gene is switched OFF and transcription is not possible? _____Acetylated histones
230. Chromosome not visible in …due to ….but can be seen during cell division _____Active nucleus, high water content
231. Histone modifications are cause of _____Epigenetics
232. More than …..specific trinucleotide repeats is seen in individuals with fragile X. _____200
233. Fruit colour of summer squash plant is an example of _____Dominant epistasis
234. How many pairs of homologous chromosomes are present in human cell? _____22 pair
235. Which one restores the paired condition for both chromosomes and genes? ___Fertilization
236. Which of followings have single genotype _____Blood group Band O
237. Meiosis, unlike mitosis involves _____1 nuclear division
238. The mitotic apparatus/spindles microtubules comprise of _____ _____centrosomes, Polar microtubules and astral microtubules
239. ….variation describes the situation in which there are many intermediates between the extremes. _____continuous
240. The molecular weight of the histone protein H1 is _____ 22500

241. Genetic material in cells is organized into chromosomes. Prokaryotes generally have chromosome. Eukaryotes have chromosomes in their nuclei _____One circular, Multiple and linear

242. Which epistasis between beneficial mutations generates improvements in function? _____Positive epistasis

243. Exchange of DNA segments between chromosomes is known as _____Crossing over

244. Which one of the following is not correct about organism with their chromosome numbers? _____Angiosperms have 78 chromosomes

245. The frequency of crossing over between any two linked genes is _____Proportional to the distance between them

246. A linkage map _____shows the actual ordering and spacing of genes on a chromosome

247. GO phase is. _____Post-mitotic phase

248. The cytokinesis in animal cells is characterized by _____Cleavage furrow

249. When two mutations together have a smaller effect than expected from their effects when alone. itis called _____Negative epistasis

250. Groups of human chromosomes are _____7

251. In metaphase |, line up at the metaphase plate, with one chromosome facing each pole. _____Tetrads

252. Which one of following statement is correct about homologous chromosomes in human cell? _____Male cells have 22 pairs of homologous and one pair of non-homologous chromosomes

253. Which of the following sequences shows the correct life cycle for humans? _____haploid egg + haploid sperm --> diploid zygote --> mitosis -> diploid adult --> meiosis -> haploid gametes

254. Variation is a differences that can be seen in individuals af the same species. Which one af following statement is correct about variation? _____-It can be due to influence of genetic makeup, environment factors or by combination of both

255. Modern concept about the law of dominance is. _____-The gene for a particular inherited character resides at a specific locus on homologous chromosome

256. In blue-eyed Mary plant a white substance is first converted to….. Substance by ist gene and that to a …substance by 2nd gene to give a blue flower. _____-Colorless: Magenta

257. Which condition is called as polygenic inheritance? _____When two or more genes influence the expression of one trait

258. In the ABO system, blood group O is characterized by the: _____absence of both antigen A and antigen B

259. What is the name of the region where double-stranded single circular DNA is found in the prokaryotic call? _____Nucleoid

260. Which of the following is widest DNA?_____A-DNA

261. Causes genes to not segregate independently. Causes genes to be more likely to be inherited together. Represents an exception to Mendel's Law of Independent assortment. _Gene linkage

262. It is estimated that about genes are imprinted and they can be found on several different chromosomes. _____40

263. Tetrads align at center..... _____Metaphase |

264. Meiosis in males is called….. and produces sperms _____-___Spermatogenesis

265. Each identical side of a chromosome is the _____Chromatid

266. How many types of enzymes are involved in histone acetylation? _____Two

267. A point mutation in the completely eliminates the ability of the centromere to function during chromosome segregation. _____CDE-III sub-domain of CEN DNA.

268. Which of the following organisms have 16 chromosomes? _____Onion and Mold

269. New phenotypic variations produced when T.H. Morgan test cross performed between F idihybrid (Wild type. gray with normal wings) with Double mutant (black with vestigial wings) because of _____Crossing over and linkage that break the genes physical connection

270. Which one of following statement about color blindness is correct? _____-Color blind mother will transmit the mutant allele to all sons but ta no daughters

271. Which of following statement is correct about the epigenetic modification? _____When histones are acetylated, chromatin is open and genes are potentially active
272. Genes will be switched ON when _____Histones are acetylated
273. The genome length of prokaryotic chromosome is smallest in _____RNA viruses
274. Exchange of DNA Segments between. ___chromosomes is known crossing over
275. Control of expression of gene by The expression of other Gene _____Epistasis
276. Expression of Phenotype of particular allele by number of individuals ___Expressivity
277. Centromere inverted in on of the following Chromesomal inverron _____Pericentric inversions
278. Human made Chromosomes is normally written as _____46.XY
279. Chrome somal aberrations are classified into _____2 types
280. Formation of Plenipotent cells from totipotent cells is called ___Cellular differentiation
281. Expression of "Dhene type of particular ailele by number_____ Expressivity
282. Addition of a complete set chromosome is named as _____Euploidy
283. Two genes close together, frequency of crossing over would be_____ Low
284. Large arm of chromosomes is ? _____arm
285. Colchicine to promoter _____Polyploidy
286. :Polygenic inheritance is controlled by _____Multiple genes
287. Fur color in Labrador is controlled by _____2 genes
288. cancer, infertility are result of _____Translocation
289. Which is following is not chromosomal aberration _____Translation
290. which is result of frame shift_____ Deletion
291. Which is not true recessive trait _____Females are more affected
292. : Molecular biology, forensic , genes mapping are application of _____VNTR
293. About 25000 SNA in human are _____Non-Synapse
294. Determination of pattern inheritance _____Pedigree
295. Types of single nucleoide polymorphism that change the sequence of amino acid_____Non-coding
296. Expression of phenotype _____Incomplete inheritance
297. Small case for alphabets used for _____RNA level
298. Frequency of autosomal recessive _____2-2.5
299. Medelian disorder is _____Single gene
300. Addition mutation is addition of _____Nucliotide
301. Which is one of chromosomal duplication _____Retro – transposition
302. Turner syndrome is an example of _____Monosom
303. Mutation which type a mutant phenotype back into the wild type _____Reverse mutation
304. A genetic disease or disorder appears as a result of mutations in an ___individual' DNA
305. Hunter's disease is an example of_ _____X-linked recessive disorder
306. A condition in which some people with an appropriate genotype fail to express the phenotype is known as the_____ incomplete penetrance
307. Which one of the following is not influenced by the silent mutation? _____Phenotype
308. Mutation that produce the change in the amino acid in protein product (histidine in for arginine_____Missense mutation
309. Base analog have the similar structure with _____standard bases of the DNA
310. Mutation which convert a DNA sequence into the stop codon. ____Non sense mutation
311. The term Ashkenazi refers to the specific people of following population _____ Jew population
312. ABO BLOOD group system is an example of _____multiple alleles
313. **Gene maping** give adea of gene location.
314. Which of following have single genotypes _____Blood group AB and O
315. Fruit clor of summer squish plant is an example _____Dominent epistatis
316. Pepper color is controlled _____2 Gene
317. Position depended method of disease identification following _____Reverse genetics
318. Genetics can be studied by _____Two ways
319. Example of Mendilian genetics _____None
320. Condition in which an intermediate phenotype is developed from cross of parental generation._____incomplete dominance

321. Condition in which one gene prevent the expression of other _____gene epitasis
322. chromosome is made up of_____ DNA
323. structure for chromosome stability _____telomers
324. Broken ends of chromosomes are _____sticky
325. Centromere is unable to work due to mutation in _____CDE III
326. XO system of sex is present in _____grasshopper
327. prokaryotic cell divides by _____binary fission
328. Chromosomes become fit in the central line _____metaphase
329. Haploid cells are produced by _____mitosis
330. Tetrads align at center_____ metaphase
331. Genes are present on chromosomes first time said by _____greogr mendal
332. Cross for determination of genotype of an individual _____test cross
333. In ABO blood group system _____Alleles A and B show co-dominance
334. If two genes are more close then frequency of cross over will be _____high
335. In nature variation is_____ Wide Spread.
336. Intermediate are produced in _____Incomplete Dominance.
337. Alleles at one gene hid/ prevent expression at second locus_____ Epitasis.
338. Lung congestion occurs in _____Cystic fibrosis.
339. Cell spend 90% of its time in _____Interphase.
340. Chromosomes are stabilized by_____ Telomere.
341. Nucleosomes folds and form _____Loops.
342. Which band is not found in Plants _____G-band
343. Most common genetic markers _____Micro satellite SNPs
344. The possible arrangement of the these genes in which eye color is placed _____between the other two.
345. Cross b/w two for finding genome _____Test Cross
346. Mechanisms of genetics _____2
347. Condition in which an organism is with 47 chromosomes with XXX _____triple X syndromes
348. Histone Deacetylase inhibitor is _____Vorionstat
349. When organism / or individual with allele for disease is unable to express the trait_____Incomplete Peiet
350. What is not chromosomal aberration _____Translation.
351. Cancer, infertility leukemia are diseases of _____Translocation.
352. Chromosome 13 trisomy_____ Patau's syndrome
353. Disease due to monosomy_____ Turner's syndrome
354. Cochicine Promotes _____Polyploidy.
355. Mutation in each cell _____6
356. Mutation that pass onto next generation _____Green Line.
357. Organism with suppressor mutation is a _____Double mutant.
358. **Point mutation** effects single loci on DNA.
359. Effect DNA sequence but not protein: _____Silent mutation.
360. Mutations in with small letters are used for nucleotide _____RNA Level
361. indicates_____ affected residue.
362. SNP are found on _____coding & non coding region.
363. 25000 SNPS_____ None synonymous.
364. **Pedigree** helps to determine mode of inheritance.
365. Frequency of autosomal recessive disorder is _____2-2.5
366. In X-linked disorder all are true except _____Female are more affected than male.
367. When gene and environment both contributes _____Multifactorial.
368. About 25000 SNA in human are _____Non Synapse.
369. Expression of Phenotype_____ Incomplete Inheritance
370. Mendilian disorder is_____ single gone.
371. Addition mutation is addition of _____Nucleoside
372. Types of single nucleoside polymorphism that change the sequence of amino acid _____Non-Coding.
373. Which mutation convert amino acid into stop codon _____non sense mutation.
374. Change in chromosome number is _____aneuploidy.

375. Type of trisomy which extra chromosomes arms derived from different chromosomes_____ **Tertiary Truism**.
376. Which blood group do not carry any antigen_____ **O Blood Group**.
377. Cytokines is occurs in_____ **Ten Phase**.
378. The Physical location of an allele on a gene is _____**Locus**
379. Sex determination chromosomal pattern in fishes is_____ **Z W system**
380. The larger arm of the chromosome is named as_____ **P-arm**
381. Medallion inheritance is_____ **Polygenic**.
382. Genetic marker for a single locus is _____**AFLP**
383. Discontinous variation is_____ **Permanent**.
384. Which one has larger chromosome _____**Plants**
385. No. of chromosome in Tobacco Plant _____**48**
386. How many types of chromosomes banding _____**9**
387. Histone Protein are basic bamino acid _____**argentine & lysine**
388. In Bacteria DNA is unwind by _____**DNA Gyrase**
389. DNA is_____ **left handed helix**
390. Replication in lagging strand is _____**discontinuous**
391. Di-allelic gene generate genotype _____**3**
392. DNA duplication caused by _____**Ratio Transposition**.
393. Which on is sexual disease _____**Alzheimer's disease.**
394. Phenotype depend on _____**both**.

QUIZ NO 2

1. ….is & type of mutation that has effect on DNA sequence but no effect on the protein. _____ _____**Silent mutation**
2. Mutations that cause loss of functions are usually _____**Recessive**
3. How many numbers of alleles can generate 10 genotypes? _____**Genes with 4 alleles**
4. …10-15 % of chromosomal deletions occur due to _____**Balanced translocations**
5. Which two alleles are called co-dominant one another and are expressed when present? _____**Allele A and Allele B**
6. Suppose that the distance between two genes on a linkage map is 7 units. The cross-over frequency between the two genes is _____**O.7%**
7. Organisms that exhibit suppressor mutations is/are _____**Double mutant**
8. Formation of pluripotent cells from totipotent cells is called _____**Cellular differentiation**
9. If purines (A/G) is replaced by purines and pyrimidines (T/C) is replaced by pyrimidines is called _____**Transition**
10. Which blood group does not carry any antigen? _____ **o**
11. Base analogs have similar structures with _____**Standard bases of DNA**
12. In case of summer squash, which statement is not correct? _____**Dominant W locus inhibit the color**
13. ..... Changes the wild type phenotype into mutant type phenotype. ____**Forward mutation**
14. The tendency of two or more than two genes to stay together during inheritance is called _____**Linkage**
15. Suppressor mutations are useful for identifying new ..... Sites which affect a biological process of interest _____**Genetic**
16. ....the mutation that changes a mutant phenotype back into the wild type. __**Reverse mutation**
17. Which genotype is not of the golden cast color in Labrador retriever? _____**bbee**
18. Most common phenotype in natural populations of the organism is called a _____**Wild type phenotype**
19. Which statement is correct about DNA methylation? _____-**DNA methylation is the addition of a methyl group ta the DNA base cytosine (C)**

MUHAMMAD IMRAN
PAST SOLVED PAPERS

1. Function Of Pedigree?

A pedigree may be used to establish the probability of a child having a particular disorder or condition. It may be used to discover where the genes in question are located (x, y, or autosome chromosome), and to determine whether a trait is **dominant** or recessive.

2. Define Epigenetics?

Epigenetics is the study of heritable phenotype changes that do not involve alterations in the DNA sequence

3. RAPD And ALFP Difference?

RAPD is Random Amplification of Polymorphic DNA'. It is a type of PCR, but the segments of DNA that are amplified are random.

AFLP is a PCR-based tool used **in genetics** research, DNA fingerprinting, and **in** the practice of **genetic** engineering. Developed **in** the early 1990s by Keygene, **AFLP** uses restriction enzymes to digest genomic DNA, followed by ligation of adaptors to the sticky ends of the restriction fragments.

4. Point Mutation?

A **point mutation** or substitution is a genetic **mutation** where a single nucleotide base is changed, inserted or deleted from a sequence of DNA or RNA. **Point mutations** have a variety of effects on the downstream protein product—consequences that are moderately predictable based upon the specifics of the **mutation**.

5. Complications Of Inheritance?

Complications to inheritance patterns: Multiple genes - epistasis, polygenic traits Genes & the environment - sex-influenced traits, incomplete penetrance.
Complications : A common recessive inheritnace can give dominant pattern, Autosomal dominant inheritance with non-penetrance, Autosomal dominant inheritance with variable expression.

6. Mutation Effects In Functioning Of Genes?

Some mutations don't have any noticeable effect on the phenotype of an organism. This can happen in many situations: perhaps the mutation occurs in a stretch of DNA with no function, or perhaps the mutation occurs in a **protein**-coding region, but ends up not affecting the amino acid sequence of the **protein**.

7. Causes Of Epigenetics?

Stress Hormone **Causes Epigenetic** Changes. Researchers found that chronic exposure to a stress hormone **causes** modifications to DNA in the brains of mice, prompting changes in gene expression

8. Genetic Mapping In Humans?

???????

9. Forward, Reverse, And Neutral Mutations: 3marks

Forward mutation: Mutation that alters the wild type phenotype into mutant phenotype.
Reverse mutation: Mutation that changes a mutant phenotype back into the wild type.
Neutral mutation: Mutation that alters the amino acid sequence of the protein but does not change its function as replaced amino acid is chemically similar or the affected AA has little influence on protein function

10. Point Mutations And Enlist 3 Structural Changes In Chromosomes?

A **point mutation** or substitution is a genetic **mutation** where a single nucleotide base is changed, inserted or deleted from a sequence of DNA or RNA. **Point mutations** have a variety of effects on the downstream protein product—consequences that are moderately predictable based upon the specifics of the **mutation**. Deletion, duplication, translocation, transcription.

Structural Change in the Structure of Chromosomes. ... Chromosomes are the vehicle of hereditary material or genes. Any alteration, addition or deletion of chromosomal part leads to alteration of number, position or sequence of genes in the chromosome.

11. 5 Complications In Inheritance?

Epistasis, polygenic traits Genes & the environment - sex-influenced traits, incomplete penetrance.

12. Modification Of Epigenetica

Epigenetic modification - non coding RNA is a new mechanism for gene regulation. RNA which is not used for making proteins can be cleaved and used to inhibit protein-coding RNAs (siRNAs, microRNAs). Three common types of modification - Cytosine methylation¬ Histones modifications¬ siRNAs¬

13. Mutation Alter Genes Expression?

Non-coding mutations: Promoter/enhancer element trinucleotide repeats in 5 and 3'UTRs Splice sites. Effect of mutation on gene function. Null allele (loss of function) - no gene product.
Hypomorph – decreased activity. Gain of function - increased activity. Dominant negative-antagonizes normal product. Neomorph - novel activity. Predicting that a gene product won't do the job. Deletion, nonsense, frameshift of sequence is deleterious. Mutation in splice site usually has sever effects. Missense mutations depend on location in protein. Is it non-conservative. Is AA conserved in evolution.

14. RFLP And RAPD ?

RFLP is a technique that exploits variations in homologous DNA sequences. **RFLP** analysis was an important tool in genome mapping, localization of genes for genetic disorders, determination of risk for disease, and paternity testing.

RAPD is Random Amplification of Polymorphic DNA'. It is a type of PCR, but the segments of DNA that are amplified are random.

15. Genetic Mapping?

Gene mapping describes the methods used to identify the locus of a gene and the distances between genes. The essence of all genome mapping is to place a collection of molecular markers onto their respective positions on the genome.

16. Sickle cell anemia?

Sickle cell anemia (**sickle cell disease**) is a disorder of the blood caused by an inherited abnormal hemoglobin (the oxygen-carrying protein within the red blood **cells**). The abnormal hemoglobin causes distorted (sickled) red blood **cells**. The sickled red blood **cells** are fragile and prone to rupture.

17. Point mutation and cause?

A **point mutation** or substitution is a genetic **mutation** where a single nucleotide base is changed, inserted or deleted from a sequence of DNA or RNA. **Point mutations** have a variety of effects on the downstream protein product—consequences that are moderately predictable based upon the specifics of the **mutation**.

Cause:
The protein may lose its function, which can **result in**a disease in the organism. For example, sickle-cell disease is **caused** by a single **point mutation** (a missense **mutation**) in the beta-hemoglobin gene that converts a GAG codon into GUG, which encodes the amino acid valine rather than glutamic acid.

18. Mutation nomenclature?

Standard **nomenclature** based on genomic DNA reference sequences and RNA reference sequences is not shown. "Genomic DNA reference sequence" simply indicates any human DNA sequence in the database that is not based on a cDNA sequence. ... Example of standard **mutation nomenclature** based on a coding DNA sequence.

19. VNTR and application?

VNTR are short nucleotide sequences organized as a tandem repeats on genomes. Found on many chromosomes, Show variations in length between individuals.
Applications of VNTR Microbiology DNA fingerprinting Genetic diversity Forensics Mapping of genomes Breeding patterns of wild or domesticated animals .

20. Single And Multifactorial Mutation?

Single gene disorders are caused by DNA changes in one particular **gene**, and often have predictable inheritance patterns. Over 10,000 human disorders are caused by a change, known as a **mutation**[?], in a **single gene**[?]. These are known as **single gene** disorders

Multifactorial Disorders. Researchers are learning that nearly all conditions and **diseases** have a genetic component. Some disorders, such as sickle cell disease and cystic fibrosis, are caused by **mutations** in a single gene. ... Conditions caused by many contributing factors are called complex or **multi factorial** disorders.

21. 5 Complication Inheritance Pattern?

Complications to inheritance patterns Multiple genes - epistasis, polygenic traits Genes & the environment - sex-influenced traits, incomplete penetrance

22. Autosomal Domenet Disordered?

A single abnormal gene on one of the first 22 nonsex (**autosomal**) chromosomes from either parent can cause an **autosomal** disorder. **Dominant** inheritance means an abnormal gene from one parent can cause disease. ... Examples of **autosomal dominant disorders** include Marfan syndrome and neurofibromatosis type 1.

23. Function Of STR?

Short Tandem Repeat (**STR**) analysis is one of the most useful methods in molecular **biology** which is used to compare specific loci on DNA from two or more samples. A short tandem repeat is a microsatellite, consisting of a unit of two to thirteen nucleotides repeated hundreds of times in a row on the DNA strand.

24. During DNA Extraction How Proteins Are Denatured?

Adding proteinase

25. Define Locus?

A **locus** (plural **loci**) in **genetics** is a fixed position on a chromosome, like the position of a gene or a marker (**genetic** marker). Each chromosome carries many genes; human's estimated 'haploid' protein coding genes are 19,000–20,000, on the 23 different chromosomes.

26. Sex Linked Traits And Disorder?

Genes on the X or Y chromosome are called **sex**-**linked**. ... For example, an X-**linked** recessive allele in humans causes haemophilia, which is much more common in males than females because males are hemizygous and therefore express the**trait** when they inherit one mutant allele.

27. Define Multiple Alleles With Example?

Multiple alleles Three or more alternative forms of a gene (**alleles**) that can occupy the same locus. However, only two of the **alleles** can be present in a single organism. For example, the ABO system of blood groups is controlled by three **alleles**, only two of which are present in an individual.

28. Genetic Disorder?

A **genetic disorder** is a **genetic** problem caused by one or more abnormalities in the genome. Most **genetic disorders**are quite rare and affect one person in every several thousands or millions. **Genetic disorders** may be hereditary, meaning that they are passed down from the parents' **genes**.

29. Genetic Mapping?

Gene mapping describes the methods used to identify the locus of a gene and the distances between genes. The essence of all genome mapping is to place a collection of molecular markers onto their respective positions on the genome.

30. What Is Location Of Intronic Sequence With Respect To Exons?

Sequences that are joined together in the final mature RNA after RNA splicing are**exons**. **Introns** are **found** in the genes of most organisms and many viruses, and can be **located** in a wide range of genes, including those that generate proteins, ribosomal RNA (rRNA), and transfer RNA (tRNA).

31. Is There Any Rules For Mutation Nomenclature?

????

32. Write Down At Least Three Mitochondrial Diseases? MELAS

Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes is
MELAS. Kearns-Sayre Syndrome NARP
NARP stands for neurogenic weakness, ataxia and retinitis pigmentosa.

33. Structul chamge in chromosome?

Structural Change in the **Structure** of **Chromosomes**. ... **Chromosomes** are the vehicle of hereditary material or genes. Any alteration, addition or deletion of **chromosomal** part leads to alteration of number, position or sequence of genes in the **chromosome**.

34.

Monosomy

Monosomy is the state of having a single copy of a chromosome pair instead of the usual two copies found in diploid cells

Causes of monosomy: followings are the causes of monosomy

Non disjunction – one gamete receives two copies of homologous chromosomes and other will have no copy.

Loss of chromosome. As it move towards pole of cell during anaphase. Fruit fly

monosomy, is the cause of some human diseases such as Turner syndrome and Cri du Chat syndrome

What are structural abnormalities?

Structural abnormalities are when part of an individual chromosome is missing, extra, switched to another chromosome, or turned upside down.
Types based on genetic material
Balanced – chromosome complement is complete.
Unbalanced – when there is incorrect amount of genetic material.
Types
Translocations Deletions Insertions Inversions Ring chromosome Isochromosome

Parents of affected individuals are asymptomatic

In most of the cases, parents of affected individuals are asymptomatic.

deletions

For point **deletions**, one nucleotide has been deleted from the sequence.

cystic fibrosis, Turner syndrome,

Results in frameshift mutation.

Suppressor mutation

Suppresses the effect of other mutation

Occurs at a site different from the site of original mutation.

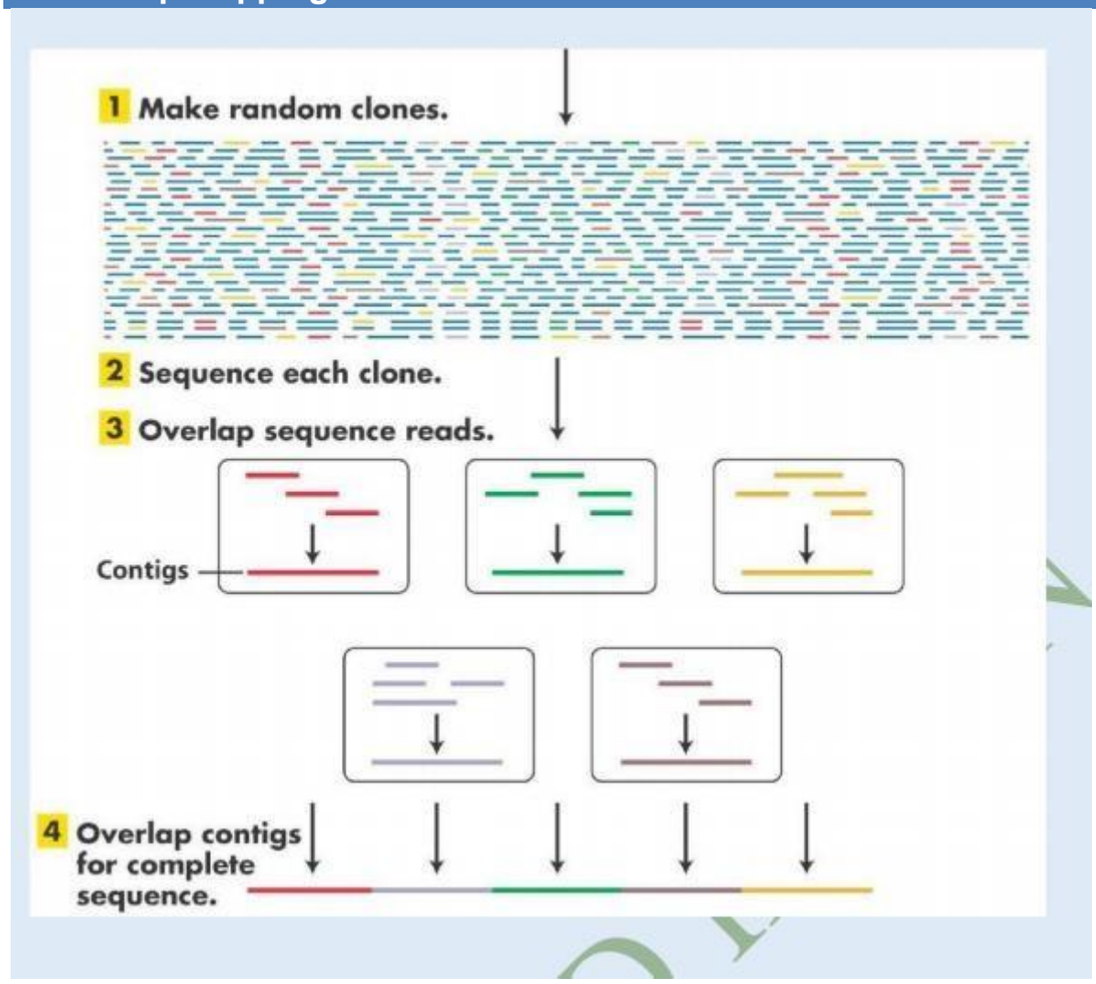
Intragenic suppression

Intragenic suppression results from suppressor mutations that occur in the same gene as the original mutation.

Intergenic suppression

Intergenic (extragenic) suppression relieves the effects of a mutation in one gene by a mutation somewhere else within the genome. Second mutation is not on same gene as original mutation

Bottom-up Mapping



Penetrance and Expressivity

Penetrance measures the proportion of individuals in a population who carry a specific gene and express the related trait. Less than 100% penetrance is a result of modifiers, epistasis, suppressors, and environmental conditions.

In **genetics**, **expressivity** is the degree to which a phenotype is expressed by individuals having a particular genotype. (Alternately, it may **refer to** the expression of particular gene by individuals having a certain phenotype.)

Low and full Penetrant

Low Penetrance An allele with low penetrance will only sometimes produce the symptom or trait with which it has been associated at a detectable level. In low penetrance, it is difficult to distinguish environmental from genetic factors.

Highly Penetrant If an allele is highly penetrant, and then the trait it produces will almost always be apparent in an individual carrying the allele.

Polyploidy

Polyploidy is the heritable condition of possessing more than two complete sets of chromosomes
Two major types

Autopolyploidy

Allopolyploidy

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.

Effect DNA sequence but not protein: Silent mutation. Mcq and definations

Trisomy

Trisomy is a type of polysomy in which there are three copies of a particular chromosome, instead of the normal two. Trisomy is a type of aneuploidy. Trisomy of chromosome 21, which is found in Down syndrome, is called trisomy 21 inversion mutation

Inversion mutations only affects a small part of the gene.

 May a single nucleotide or more than one nucleotides are involved.

X-linked recessive disorders

Duchene Muscular Dystrophy

Hunter's Disease

Menkes Disease

Hemophilia A and B

Color Blindness

Diagnostic tests for genetic disorders

Physical analysis: Physical analysis for affected individual

Ultrasonography of fetus: Ultrasonography of fetus determines malformations of head, internal organs, and extremities

Analysis of amniotic fluids: Amniotic fluid analysis to determine genetic and chromosomal disorders after 14 wks gestation. More than 200 various genetic diseases easily be detected. Maternal blood analysis: Maternal blood analysis to observe abnormal fetus.,

Polymorphism

Polymorphism, in biology, a discontinuous genetic variation resulting in the occurrence of several different forms or types of individuals among the members of a single species

Aneuploidy: Change in chromosomes number can occur by the addition/deletion of chromosome or part of a chromosome

. Euploidy: Gain of one or more complete sets of chromosomes

pattern of inheritance

Most genes have one or more versions due to mutations or polymorphisms referred to as alleles. ... There are five basic modes of **inheritance** for single-gene diseases: autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, and mitochondrial.

37. Mutation that produce the change in the amino acid in protein product (histidine in for arginine Missense mutation

Multifactorial diseases six

: Genetic disorders controlled by many genes plus the effects of the environment

Asthma,Cancers ,Diabetes ,Heart diseases,Hypertension,Obesity

Pleiotropy

Pleiotropy The ability of a gene to affect an organism in many ways is called pleiotropy.

 Pleiotropy refers to an allele which has more than one effect on the phenotype.

Types of deletion include the following:

- Terminal deletion – a deletion that occurs towards the end of a chromosome.
- Intercalary/interstitial deletion – a deletion that occurs from the interior of a chromosome.

Microdeletion – a relatively small amount of deletion (up to 5Mb that could include a dozen genes).

Applications of molecular markers – short range

- Parentage determination
- Genetic distance estimation
- Determination of twin zygosity

Applications of Molecular Markers – Long Range

- Gene mapping & mapping of QTL by linkage
- Markers assisted selection of breeds

Method of mapping gene disorders

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

How genetic mapping performed

Scientists isolate DNA from samples and examine it for the unique patterns of bases.

Molecular techniques are referred to as genotyping polymorphisms or markers.

1. Define Epistasis

Epistasis and modifier genes Epistasis is a phenomenon that consists of the effect of one gene being dependent on the presence of one or more modifier genes Type of polygenic inheritance where alleles at one gene locus can hide or prevent expression of alleles at second gene locus.

2. VNTR and Applications of VNTR

VNTR are short nucleotide sequences organized as a tandem repeats on genomes.

Found on many chromosomes

Show variations in length between individuals.

Applications of VNTR

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

3. Genomic Imprinting

Genomic Imprinting Epigenetic marking of a locus on the basis of parental origin. Results in monoallelic gene expression. Some genes are expressed only from the maternal genome and some only from the paternal genome. It is estimated that about 40 genes are imprinted and they can be found on several different chromosomes .

4. Point mutations

- Point mutations affect single sites on DNA.
- Substitution of one base for another.

- Deletion/addition of a single base or small number of bases.

Transitions

 If purines (A/G) is replaced by purines and pyrimidines (T/C) is replaced by pyrimidines.

Transversion

 If purine substitutes for pyrimidines or pyrimidine is replaced by purines. It is called as transversions.

Result of point mutations

- Silent mutations
- Missense mutations
- Nonsense mutations

5. Frameshift mutations result in all amino acids downstream from the mutation site being completely different from wild type. These proteins are generally nonfunctional.

Mutation - Forward, Reverse and Neutral

Forward mutation

- Mutation that changes the wild type phenotype into mutant type phenotype.
- Any mutation which changes a functional wild genotype or wild phenotype to nonfunctional form.

Wild type phenotype/ genotype

- Most common phenotype in natural populations of the organism is called a wild type phenotype.

reverse mutation

- Mutation that changes a mutant phenotype back into the wild type.

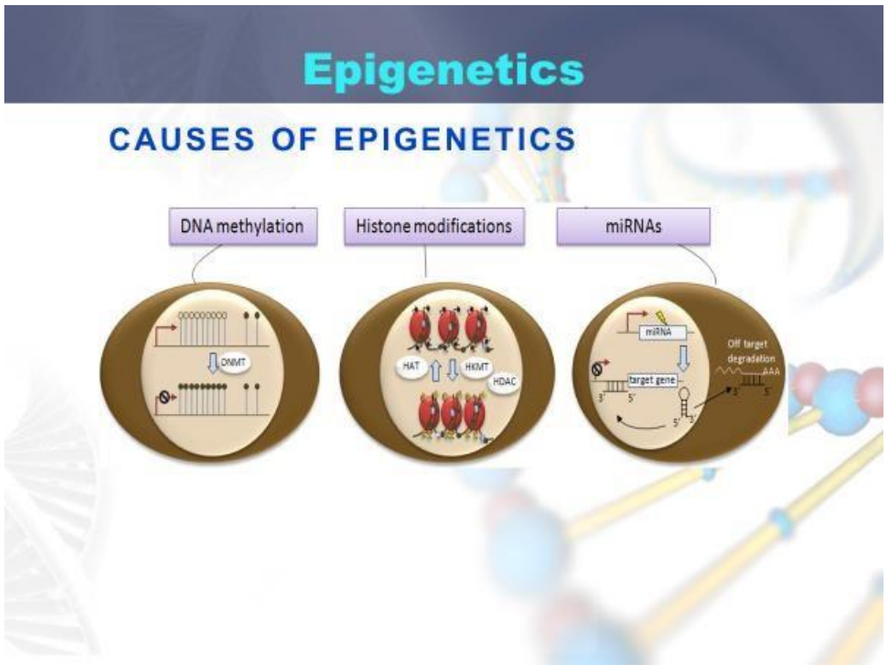
neutral mutation

- Mutation that alters the amino acid sequence of the protein but does not change its function as replaced amino acid is chemically similar or the affected amino acid has little influence on protein function.

6. Epigenetics

the study of changes in organisms caused by modification of gene expression rather than alteration of the genetic code itself. One **example** of an **epigenetic** change is DNA methylation — the addition of a methyl group, or a "chemical cap," to part of the DNA molecule, which prevents certain genes from being expressed. Another **example** is histone modification. Histones are proteins that DNA wraps around

Epigenetics An epigenetic trait is a stable heritable phenotype resulting from changes in chromosome without alterations in the DNA sequence.



Autosomal Dominant diseases

7. Marfan syndrome
8. Huntington's disease
9. Retinoblastoma
10. Waardenburg syndrome
11. Myotonic dystrophy
12. Polycystic kidney disease
13. Achondroplasia
14. Polydactyly
15. Heredity hearing loss

Examples of **autosomal** recessive **disorders** include cystic fibrosis, sickle cell anemia, and Tay-Sachs **disease**

7. Mutation nomenclature

Nomenclature

- While describing sequence variants, it should be written according to HGNC rules.
- 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)

9. How mutations alter gene function_to add some data splice site mutations have severe Effects

- Splice sites site mutations usually have severe effects on protein functions.
- Often this kind of mutation produce one or more exon skipping or larger fragments of DNA. **Effects of null allele and hypomorph** □ Null allele results in loss of function. Usually no gene product.
- Hypomorph alleles results in decreased activity of the proteins.

Effects of mutation on gene function

- Gain of function - increased activity.
- Dominant negative-antagonizes normal product.
- Neomorph - novel activity

Predicting that a gene product won't do the job

- Deletion, nonsense, frameshift of sequence is deleterious.
- Mutation in splice site usually has severe effects.

Predicting that a gene product won't do the job

□ Missense mutations have sometimes severe effects on protein function while some times less severe effects. It depends on the type of amino acid which was replaced.

Predicting that a gene product won't do the job

- Missense mutations depends on location in protein.
- Is it in non-conservative
- Is amino acid is conserved in evolution

10. Genetic Disorders: A genetic disease or disorder appears as a result of mutations in an individual's DNA. Change in DNA sequence may be small or large like deletions/addition of entire chromosome.

Multifactorial diseases: Genetic disorders controlled by many genes plus the effects of the environment.

11. Common X-linked dominant diseases

- Rett syndrome

- Alport syndrome
- Goltz syndrome
- Fragile-X Syndrome

12. *Short Tandem Repeat Polymorphism* Short tandem repeat sequences

- STR are repeats of nucleotide sequences.
- AAAAAA… mononucleotide
- ATATAT… dinucleotide
- TAGTAGTAG…trinucleotide
- TAGTTAGTTAGT… tetranucleotide
- TAGGCTAGGCTAGGC… pentanucleotide
- Different alleles contain different numbers of repeats.

13. complications in basic inheritance pattern

Complications in basic inheritance pattern

Single genes -Non-mendelian inheritance

Differences in gene actions can generate more complex inheritance patterns.

Single gene can give codominance, incomplete dominance, overdominance.

Complications to inheritance patterns

Multiple genes - epistasis, polygenic traits Genes & the environment - sex-influenced traits, incomplete penetrance

Complications

- A common recessive inheritnace can give dominant pattern
- Autosomal dominant inheritance with non-penetrance
- Autosomal dominant inheritance with variable expression.

Pedigrees with complicated inheritance pattern

Complications

- A common recessive can give dominant pattern
- Autosomal dominant inheritance with non-penetrance
- Autosomal dominant inheritance with variable expression

14. Frequency of autosomal dominant disorders

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

Point mutations

Point mutations

- Point mutations affect single sites on DNA.
- Substitution of one base for another.

- Deletion/addition of a single base or small number of bases.

Transitions

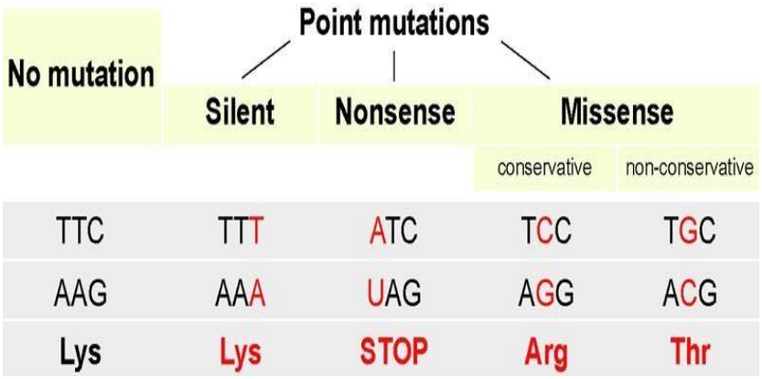
- If purines (A/G) is replaced by purines and pyrimidines (T/C) is replaced by pyrimidines.

Transversion

- If purine substitutes for pyrimidines or pyrimidine is replaced by purines. It is called as transversions.

Result of point mutations

- Silent mutations
- Missense mutations
- Nonsense mutations



Point mutations

- Point mutations affect single sites on DNA.
- Substitution of one base for another.

Common mitochondrial diseases

16. Myoclonic epilepsy
17. Mitochondrial recessive ataxia
18. Leber hereditary optic neuropathy
19. Sensory ataxia neuropathy

17.

Q. Define linkage?

Linkage is defined genetically as the failure of two genes to assort independently. Linkage occurs when two genes are close to each other on the same chromosome

Q. What is the difference between karyotype and idiotype

Karyotype	Idiotype
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 in an individual organism	the karyotype of a species may be represented diagrammatically, showing all the morphological features of the chromosome; such a diagram is known as Idiotype

Q. What is polygenic inheritance?

Condition when two or more genes influence the expression of one trait. Polygenic inheritance is controlled by multiple genes

Q. What is epistasis?

Epistasis is a phenomenon that consists of the effect of one gene being dependent on the presence of one or more modifier genes Type of polygenic inheritance. Where alleles at one gene locus can hide or prevent expression of alleles at second gene locus.

Q. What is linked gene?

:Genes that are found on the same chromosomes are called linked gene.

Q. What is physical mapping?

: A physical map is primarily base on the location of landmark along a DNA molecule.

Q. write Name of different mapping technique?

1 : Cytogenetic map 2 : In situ hyberdization 3 : Genetic mapping 4 : Restriction enzyme 5 : Radiation hybrid mapping.

Q. What is position independent?

It is a forward genetics. It started at the phenotype, determining which protein was involvednand getting to the gene through the protein.

Q. How gene – species oligonucleotide are prepared?

Oligonucliotide isolating the protein product of the gene. These genes are usually produced large amount of protein. Then gene – species oligonucleotide are prepared.

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Q. How polygenic inheritance are controlled by multiple genes?

When two or more genes influence the expression of one trait. Then polygenic inheritance is controlled by multiple genes.

Q. What is multifactorial?

Many of traits are effected by the person,s environment, so they are called multifactorial.

Q. Give example of polygenic trait?

1 : Stature 2 : Shape of face 3 : Finger prints

Q. How many genes are controlled the chicken combs shapes? And where these found?

Comb shapes are controlled by two genes .These genes are found on two different chromosomes. Gene 1 : R , Gene 2 : p.

Q. Short note on hair colour?

Hair color is controlled by alleles on chromosomes 3, 6, 10 and 18. When more dominant alleles appear in genotype, the darker will be the hair.

Q. What is role of positive epistasis?

Positive epistasis is occurring between beneficial mutations. It generates improvement in function.

Q. What is negative effect?

When two mutation has a smaller effect than expected from their effects when alone.It is called negative effect.

Q. How sign epistasis occur?

It occur when one mutation has the opposite effect when in the presence of another mutation.

Q. What is dominant epistasis?

It is fruit color in summer squash. Epistasis gene exerts is effect with the presence of a dominant allele.

Q. Write duplicate dominant epistasis and its shape?

fruit shape in shephered,s purse . A – or B - =heart shape . aa and bb = narrow shape

Q. What is epigenetics?

Its literally means above the genetics. Epigenetics trait is a stable and heritable phenotype resulting from changes in chromosomes without alternation in DNA sequences.

Q. Write molecular mechanism of epigentics?

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1 : Methylation of cytosine posttranslational modification to histone protein , include acetylation, phosphorylation .

Q. how many epigenetic modifications are there write their name?

Three types of epigenetic modifications are there these are

1) Cytosine methylation 2) Histones modification 3)Non-coding RNAs (siRNAs)

Q. describes histones in Histone modification?

Histones are the proteins that organize the genetic material. Have a high percentage of basic amino acids, which gives histones an overall positive charge, while DNA is negative charged.

Q. Which two enzymes involved in Histone accyitylation?

1) HAT: Histone acetyltransferase 2) HDAC: Histone deacetylase.

Q. what is role of histones in accetylation?

When histones are acetylated, chromatin is open and genes are potentially active. When histones not tagged, deacetylated, the chromatin condenses and genes silenced.

Q. what are Common Epigenetics regulated phenomena?

1) Cellular Differentiation 2) X Chromosome inactivation 3) Genomic imprinting

Q. Describe Identical twins?

Identical twins are from the same zygote, so they begin life with the same genetic information. While infants they experience the same or very similar environments, so there is little variation in the epigenome. Over time the twins' environments will diverge, resulting in individual epigenetic tags to form for each twin. The difference in the twins' epigenomes is what makes them become different when they are older. The epigenetic tags can have such an effect on the twins that one can develop a disease while the other is fine.

Q. what is Vorinostat ?

Vorinostat is a histone deacetylase inhibitor.

Q. what environmental factors affected by epigenetics?

Nutrition of mother, stress mother, social interactions and physical activities, Exposure to toxins and diet.

Q. defines penetrance?

Proportion of individuals carrying a particular variant of a gene (allele or genotype) that also expresses an associated trait (phenotype)

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Q. what is variable expressivity?

When phenotype is expressed to a different degree among individuals with the same genotype it is called variable expressivity.

Q. MEN 1 stand for?

Multiple endocrine neoplasia 1 it is a hereditary disorder

Q. Write any two types of penetrance?

Complete penetrance, highly penetrance, incomplete or reduced penetrance, low penetrance.

Q. What is chromosomal aberration?

Any change in normal number or structure of chromosomes. Affects individuals may be due altering the amounts of products of the genes involved. Altered amounts may cause anomalies directly or may alter the balance of genes

Q. How many types of chromosomal aberration are there describe them?

1) Numerical (usually due to de novo error in meiosis).

2) tructural (may be due to de novo error in meiosis or inherited).

Q. Which structural abnormalities may occurs due to change in chromosomes?

Structural abnormalities may occurs due to

1) Translocations 2) Deletions 3) Duplications 4) Inversions

Q. chromosomal identifications dependent upon which factors? Write their name?

1. Size 2. Location of the centromere 3. Banding patterns

Q. how many chromosome are present in down syndrome?

Ans: 47 chromosome

Q. Write name of Common numerical Autosomal and sex chromosome Abnormalities?

Ans Autosomal 1) Downsyndrome (Trisomy 21) 2) Edwards syndrome (trisomy 18) 3) patau syndrome (trisomy13)

sex chromosomes abnormalities 1) Turner syndrome 45,x 2) klinefelter syndrome 47,xxy

Q. wite the characteristics of Down syndrome

Ans it is a trisomy of chromosomes no 21. The frequency of trisomy increases with increasing maternal age. Sometimes, robertssonian translocation involving chromosome 21. 3-4 %, not related to maternal age.

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Q write characteristic of turner syndrome.

Ans Relatively common disorder caused y the loss of genetic material from one of the sex chromosomes. Affects only females and they have only one x chromosome.

Q. Define Triploidy ? how many chromosomes that deseas contain?

Ans This is phenomena in which all the set of chromosomes contain one extra chromosome like a normal person have 2 pair of chromosomes and triploidy person have 3 chromosomes so they become 69 chromosomes and a normal person has 46 chromosomes.

Q. Define structural abnormalities?

Ans rearrangement because of chromosome breakage and subsequent reunion in a different configuration.

what are the types of structural abnormalities.

Ans there are 6 types i.e 1) Trasnlocation 2) Deletions 3) insertions 4) inversions 5) ring chromosomes 6) isochromosomes

Q. Define Translocation? Write its name?

Ans Transfer of genetic material from one chromosome to another chromosome called translocation. There are two types of translocations 1) Reciprocal translocation and 2) Robertosnian Translocation.

Q. Defien Reciprocal Translocation?

Ans An exchange of genetic material between two different chromosomes is called a reciprocal translocation in which no loss or gain of chromosomal material.

Q. Define Robertsonian translocations.?

Ans Break occurs at the extreme end of the short arms of two non homologuous acrocentric chromosomes and the small acentric fragments are lost so the the larger fragments fuse at their centromeric regions to for a single chromosome. this type of translocation occurs between chromosomes no 13 and 14 in humans.

Q write common human diesease caused by translocations?

Ans the common disease in humans caused by translocations are 1) cancer 2) infertility 3) down syndrome 4) leukemia 5) XX male syndrome

Q. What is difference between disjunction and non-disjunction?

Ans separation of two homologous chromosomes during cell division is called disjunction. When two homologous chromosomes or sister chromatids fails to separate during cell division it is called as non-disjunction.

Q differentiate between meiotic and mitotic non disjuntion ?

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Ans if non-disjunction occurs during 1st meotic division then all the sperms/ova derived from primary spermatocyte /oocytes will be abnormal.

Mitotic non-disjuntion results in cells with differetn number of chromosomes due to breaking of the spindle fibers during metaphse or anaphase

Q. Explain the failure of chromosomes separation?

Ans 1) Failure of a pair of homologous chromosomes to separate in meisis1.

2) Failure of sister chromatids to separate during meiosis II.

3) Failure of sister chromatids to separate during mitosis.

Q what will be the results in Mitotic non-disjuntion?

Ans Mitotic non-disjunction results in trisomy and monosomy. it causes trisomic and monosomic daughter cells to be formed.

Q. What are the causes of Non-Disjunciton ?

Ans there are several cause of non-disjuntion but the most are 1) Aging effect on primary oocyte 2) radiation from the environment 3) delayed fertilization after ovulation

Q. What is the difference between Trisomy and Monosomy?

Trisomy is a type of polysomy in which there are three copies of a particular chromosome, instead of the normal two. Trisomy is a type of aneuploidy (an abnormal number of chromosomes).

Monosomy occurs when there is loss of a single chromosome. monosomy of autosomes is lethal during sex chromosome monosomy will be Turner syndrome XO in which loss of sex chromosomes and they don't survive and died

Q. Define Trisomy Types

. Ans There are 4 types of Trisomy. 1) Primary Trisomy - An entire etra chromosome has been copied it is called primary trisomy. 2) Partial trisomy- in which an extra copy of part or fragment of a chromosomes present called partial trisomy. 3) Secondary trisomy- the extra chromosome has duplicated arms so it is called secondary trisomy. 4) Tertiary trisomy - extra chromosome is made up of copies of arms from two other chromosomes

Q. What are the common Trisomy in humans?

Ans Trisomy 21 , Trisomy 18 trisomy 13 and Klinefelter's syndrome (47xxy)

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Q. What are the causes of Monosomy?

Ans during Non-disjunction one gamete receives two copies of homologous chromosomes and other will have no copy. loss of chromosome as it move towards pole of cell during anaphase.

Q. Write the name of Monosomy diseases in humans.

Ans Turner syndrome and Cri Du chat syndrome

Q Define polyploidy?

Ans Multiples of haploid cells or no of chromosomes are called polyploidy . it can be in form of triploidy, tetraploidy , pentaploidy etc. during polyploidy fetus does not survive this phenomena is seems in many crops.

Q. write types of polyploidy in plants.

Ans Triploid (3x) Watermelon, Tetraploid(4x) cotton , Pentaploid (5x) Kenai Birch , Hexaploid (6x) Wheat

Q. Write the causes of Polyploidy

Ans 1) Retention of polar body. 2) Formation of diploid sperms 3) Di-spermy- fertilization by two sperms.

Q. Write polyploidy examples in plants.

Ans Triploid - Apple , Bannana, citrus fruits , ginger watermelons .

Tetraploid. Cotton, patato, tobacco, peanut

Hexaploid - Oat, Kiwifruit

Octaploid- Straberry

Decaploid- Some sugar cane hybrids

Q. What is difference between Autopolyploidy and Allopolyploidy.

Ans Autopolyploids are polyploids with multiple chromosome sets dervied from a single species. in case of allopolyploids are polypoloids with chromosomes derived from different species in the result of cross between two different species. Interspecies crosses can generate alloploids and their offspring generally sterile.

Q. Which chemical is used to promote the polyploidy in plants?

Ans is Colchicine is used to promote polyploidy in the plants to avoid non disjunction.

Q. What does the sterile means?

Ans The word sterile means that the specie cannot reproduce.

Q. What is the difference between Aneuploidy and Euploidy?

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Ans Aneuploidy:- change in chromosomes number can occur by the addition /deletion of chromosome or part of a chromosome. In case of Euploidy :- gain of one or more complete sets of chromosomes

Q. what is the difference between Terminal and interstitial deletions?

Ans in the Terminal the part of the chromosomes that is at the end like tips of the chromosomes been deleted and in interstitial the part of chromosomes which is at middle part or two breaks.

Q. Define Gene or chromosomal duplication?

ANs Gene duplication or chromosomal duplication is a major mechanism through which new genetic material is generated during molecular evolution

Q. Define Replication Slippage?

Ans Replication slippage is an error in DNA replication that can produce duplication of short genetic sequences. during replication DNA polymerase begins to copy the DNA.

Q. Write the causes of chromosomal duplications?

Ans 1) DNA Replication - Gene duplications can arise as products of several types of errors in DNA replication and repair machinery. 2) Ectopic homologous recombination;- common sources of gene duplications include ectopic homologous recombination 3) ectopic recombination Misaligned homologous - Duplicatoin arise from an event termed unequal crossing-over that occurs during meiosis between misaligned homologous chromosomes. 4) others causes of duplications - Retro-transposition event, aneuploidy, polyploidy, and replication silpage are also cause of duplications

Q. Define Dispersed and Tandem Duplications.

Ans. Some duplications are dispersed found in different locations from each other. Tandem duplications found next to each other

Q What is called pseudogenes?

Ans pseudogenes are inactive forms of gene forms by mutations, these genes cannot produce messenger RNA.

Q. What is called Chromosomal inversions?

Ans it involves two break re-arrangement and segment is reversed there are two types Pericentric inversions in which centromere is involved and paracentric in which only chromosomes Arms like P or Q is involved.

Q. What are the chromosomal inversions affects?

Ans it affects gene expression gene is re-positioned in a way that alters its gene expression 2 % of the human population carries karyotipically detectable inversions.

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Q. Define Hetrozygote inversion?

Ans individual with one copy of a normal chromosome and one copy of an inverted chromosome called inversion heterozygotes usually phenotypically they are normal.

Q. what are the effects of chromosomal inversion in humans?

Ans The most common inversion seen in humans is on chromosome 9 , at inv 9 (p12q13) this inversion is generally considered to have no harmful effects but increased risk of miscarriage or infertility.

Q. Differentiate between ring chromosome and Philadelphia chromosome?

Ans **A ring chromosome** is a chromosome whose arms have fused together to form a ring. in humans it is denoted by small r and in drosophila it is denoted by capital R.

philadelphia chromosome:- is a specific chromosomal abnormality that is related with chronic myelogenous leukemia(CML). reciprocal translocation between chromosome 9 and chromosome 22 which is specifically designated t(9:22)(q34:q11)

Q. Mutations are source of genetic variation? Justify

Mutation and variation Mutations are a major source of genetic variation in a population increasing biodiversity. Some variations may help to survive better and longer. Mutations in gametes (egg & sperm) are passed onto offspring. Mutations in body cells only affect the organism in which they occur and are not passed onto offspring.

Q. write name of major types of mutations.

Major types of mutations

1) Points Mutations 2) Variations in chromosomes number 3) Variations in chromosomes structure

Q. writes differences between Somatic and Germline mutation

Somatic mutation: Arise in the somatic cells. Passed on to other cells through the process of mitosis. Effect of these mutations depends on the type of the cell in which they occur.

Germline mutation: They occur in the cells that produce gametes. This type of mutation usually passed on to future generation. In multicellular organisms, the term mutation is generally used for germ line mutations..

Q. Explain - Forward, Reverse and Neutral mutations

Wild type most common phenotype in natural populations of the organism is called as wild type phenotype. The effect of mutation is considered with reference to wild type phenotype.

Forward mutation: Mutation that alters the wild type phenotype into mutant phenotype.

Reverse mutation: Mutation that changes a mutant phenotype back into the wild type.

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Neutral mutation: Mutation that alters the amino acid sequence of the protein but does not change its function as replaced amino acid is chemically similar or the affected AA has little influence on protein function..

Q What is difference between loss of function of gain of function ?

Loss of function, Gain of function Loss of function: Complete or partial loss of the normal function. Structure of protein is so altered that it is no longer works correctly. Mutation can occur in regulatory regions that affect transcription, translation or splicing of the protein. Frequently recessive

Gain of function: Produces an entirely new trait. Such a mutation causes a trait to appear in inappropriate tissues or at inappropriate times in development. It is frequently dominant.

Q. Differentiate between conditional and lethal mutants?

Conditional mutation: Expressed only under certain conditions. E.g. A temperature-sensitive mutation can cause cell death at high temperature, but might have no deleterious effects at a lower temperature.

Lethal mutation: Causes the death of the organism. Death does not have to occur immediately, it may take several months or even years. Longevity of an individual is significantly reduced.

Q. write difference between spontaneous and induced mutations?

Spontaneous mutation: Mutations that result from natural changes in DNA.

Induced mutation results from changes caused by environmental, chemicals and radiations.

Q. Define mutagens?

Mutagen: Any environmental agent that increases the rate of mutation above the spontaneous is called a mutagen such as chemicals & radiations, X-rays, Mustard gas etc.

Q. Define Base analogs?

Base analogs: Chemicals with structures similar to that of any of the four standard bases of DNA. DNA polymerases cannot distinguish these analogs. They may be incorporated into newly synthesized DNA molecules.

Q. Define Point mutations:

Point mutations affect single sites on DNA and it is Substitution of one base for another. Or Deletion/addition of a single base or small number of bases.

Q. what is difference between Transition and transversion mutation.

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if purine (A/G) is replaced by purines and pyrimidine (T/C) is replaced by pyrimidines it is called transitions mutation.

If purine substitutes for pyrimidine or vice versa is called transversion.

Q. Define Silent Mutation

Silent Mutation: These types of mutations have affected the DNA sequence but not the protein. They have no effect on the organism's phenotype. There is change at DNA level but no change in protein level.

Q. Define Missense mutation and its effects on protein formation?

Missense mutation: Missense mutations substitute one amino acid for another. Some missense mutations have very **large effects**. Some have minimal or no effect. It depends on where the mutation occurs in the protein's structure and how big a change in the type of amino acid it is. Missense mutation produces a change in amino acid sequence in protein product (e.g. Histidine in for Arginine). May change function of protein or may not!

Q. Define nonsense mutation and its effects on protein formation

Nonsense mutation: Nonsense mutations convert an amino acid into a stop codon. **The effect** is to shorten the resulting protein. Sometimes this has only a little effect, as the ends of proteins are often relatively un-important to function. However, often nonsense mutations result in completely non-functional proteins.

Q. Define Addition mutation or frame shift mutations

Additions mutation: Addition mutation is actually results in a frame shift mutation. Often, it has severe effects on protein function. It is also called as insertions.

Q. Define Deletion Mutations and its effects.

Deletions mutations:- Deletion of one or more nucleotides in DNA sequence is called deletion mutations. These types of mutations have severe effects on the phenotype.

Q. How mutations alter gene function

Null allele (loss of function) - no gene product.

Hypomorph – decreased activity.

Gain of function - increased activity.

Dominant negative-antagonizes normal product

Neomorph - novel activity

Predicting that a gene product won't do the job

Deletion, nonsense, frameshift of sequence is deleterious.

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Mutation in splice site usually has sever effects.

Missense mutations depend on location in protein.

Q. Explain the 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)

There is no nucleotide 0 (zero).

Nucleotide 1 is A of the ATG-translation initiation codon.

The nucleotide 5' of the ATG-translation initiation codon is -1, the previous -2, etc. e.g. in 5' UTR c.-12G>A

The nucleotide 3' of the translation stop codon is *1, the next *2, etc. e.g. in 3' UTR c.*70T>A

Nucleotide numbering is purely arbitrary and starts with 1 at the first nucleotide of the database reference file.

No +, - or other signs are used.

The sequence should include all nucleotides covering the sequence (gene) of interest and should start well 5' of the promoter of a gene.

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BIO-301 GENETICS NOTES FOR FINAL TERM

Q. Define Genetic Disorders and Its classification.

Genetic Disorders: A genetic disease or disorder appears as a result of mutations in an individual's DNA. Change in DNA sequence may be small or large like deletions/addition of entire chromosome.

Fully penetrant and low penetrant

Fully penetrant, where other genes and environmental factors have no effect

Low-penetrance, genes play a small part, along with other environmental factors

Classification of genetic disorders

1) Multifactorial disorders **2) Single gene disorders** **3) Chromosomal disorders**

4) Mitochondrial disorders **5) Somatic mutations (cancers)**

Q. Explain Inheritance patterns of genetic disorders

Autosomal Dominant: Affected individuals are heterozygote. One allele is mutated, while other one is normal.

Autosomal recessive: Homozygous, when both alleles of the gene are mutated.

X-linked recessive: Mutated alleles are present on either of the sex chromosomes.

Multifactorial: Genes and environment both contribute. Multifactorial frequent, single gene disorders less frequent: Multifactorial are common. Single gene less common like dominant/recessive pedigree patterns

Chromosomal disorders:- Thousands of genes may be involved. Multiple organ systems affected at multiple stages in gestation

Q. Explain Autosomal recessive inheritance / diseases

Autosomal Recessive diseases: Diseases occurs in individuals with two mutant alleles. In general, an individual inherit one mutant allele from each of the parent.

Recessive diseases: Homozygotes with two copies of the altered gene

Recessive diseases: Since each parent has two alleles. Chance of inheriting a mutant allele from one parent is 50% and for the other parent is also 50%. Net chance of inheritance of two mutant alleles is 25%.

Generally, the disease appears in the progeny of unaffected parents.

Affected progeny include both males and females equally.

Phenotypic proportions are equal – autosomal recessive: When we know that both male and female phenotypic proportions are equal, we can assume that we are dealing with autosomal inheritance.

Q. Write name of Common recessive disorders

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1) Bloom syndrome	2) Carpenter syndrome	3) Cystic fibrosis
4) Thalassemia	5) Many forms of mental retardation	6) Gaucher's disease
7) Glycogen storage diseases	8) Rotor syndrome	9) Many of eye diseases

Q. Properties of Autosomal recessive inheritance

Consanguinity: These diseases appear where parents have common ancestors.

Same allele increases chances of inheriting disorders: The chance of inheriting identical alleles by an individual from both parents increases the chance of inheriting a recessive disorder.

Common genetic background: Small populations of individuals with a common genetic background may have increased risk of recessive diseases. Ashkenazi: Gaucher disease. Phenotype found more likely in siblings of proband than in other relatives. Males and females are equally affected. In most of the cases, parents of affected individuals are asymptomatic. Risk for each of the sibling being affected. Risk of disease for each sib is 25 %.

Q. Explain Autosomal dominant inheritance/ disorders

Autosomal dominant disorders: In autosomal dominant disorders, the normal allele is recessive and the abnormal allele is dominant. In a typical pedigree, every affected person has one affected parent. Equal numbers of affected females/males are expected. Male to male transmission is possible. One-half of the children of an affected individual are expected to have inherited the dominant allele.

Autosomal Dominant disorders

Pedigree analysis, main clues for identifying a dominant disorder is that the phenotype tends to appear in every generation of the pedigree. Affected fathers/mothers transmit the phenotype to both sons and daughters. Trait is common in the pedigree. Trait is found in every generation.

Autosomal Dominant diseases

1) Marfan syndrome	2) Huntington's disease	3) Retinoblastoma
4) Waardenburg syndrome	5) Myotonic dystrophy	6) Polycystic kidney disease
7) Achondroplasia	8) Polydactyly	9) Heredity hearing loss

Q. Explain Dominance and Recessiveness? Define Hemizygous

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Dominance and recessiveness are the properties of characters/traits, not genes. A character is dominant if it is expressed in the heterozygote and recessive if not.

Recessiveness: Cystic fibrosis is recessive because only homozygotes manifest it, whereas heterozygotes show the normal phenotype.

Hemizygous: Males are hemizygous for loci on the X and Y chromosomes, where they have only a single.

Q. Explain X-linked recessive inheritance and disorders? Their Characteristics and disease.

X-linked recessive: Traits which are due to the genes which are present on either of the sex chromosomes. Many of the genes have a disease phenotype.

Males are hemizygous and express the disease: Males are hemizygous, will express the disease phenotype if one mutation is present.

Homozygous Females manifest disease: Females may be homozygous or heterozygous. Homozygous may manifest the disease.

General characteristic

1) Trait is rare in pedigree. 2) Trait skips generations. 3)Affected fathers do not pass to their sons.

4) Males are more often affected than females.

X-linked recessive disorders:-X-linked recessive: X-linked recessives expressed in all males but only in homozygous females.

Common X-linked disorders

1) Duchene Muscular Dystrophy 2) Hunter's Disease 3) Menkes Disease

4) Hemophilia A and B 5) Color Blindness

Q. Write name of Common X-linked dominant diseases

1) Rett syndrome 2) Alport syndrome 3) Goltz syndrome 4) Fragile-X Syndrome

Q. Explain maternal inheritance of mitochondrial DNA

Mitochondrial DNA (MTDNA) is inherited through ovum not sperm. Mother could pass it to all children. Father will pass it to none of the children. More than one copy of mtDNA is passed. Mutations are common.More than one type of mtDNA genome is passed.

Q. What is difference between mtDNA and Nuclear DNA ?

Nuclear DNA is inherited from all ancestors while mtDNA is inherited from a single lineage i.e from mother only.

Q. Write the Properties of Mitochondrial inheritance

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1) Heteroplasy 2) Variable expression 3) Pleiotropy

4) Reduced penetrance 5) Functional somatic mosaic

Q. writes the name of Common mitochondrial diseases.

1) Myoclonic epilepsy 2) Mitochondrial recessive ataxia 3) Leber hereditary optic neuropathy

4) Sensory ataxia neuropathy

Q. What is Multifactorial inheritance and disorders?

Multifactorial diseases: Genetic disorders controlled by many genes plus the effects of the environment.

Q. What is difference between genetic and environmental disorders in Multifactorial?

They are rare, genetically simple, unifactorial and high recurrence rate while environmental they are common genetically very complex, multifactorial and low recurrence rate.

Q. writes the name of Multifactorial diseases

1) Asthma 2) Cancers 3) Ciliopathies 4)Cleft palate 5) Diabetes 6) Heart diseases

7) hypertension 8) inflammatory bowel disease 9) intellectual disability

10) mood disorder 11) Obesity 12) infertility

Q. Explain Diagnostic tests for genetic disorders

Physical analysis: Physical analysis for affected individual.

Ultrasonography of fetus: Ultrasonography of fetus determines malformations of head, internal organs, and extremities.

Analysis of amniotic fluids: Amniotic fluid analysis to determine genetic and chromosomal disorders after 14 wks gestation. More than 200 various genetic diseases easily be detected.

Maternal blood analysis: Maternal blood analysis to observe abnormal fetus

Q. Define pedigree and its use.

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.

Q. Define Gene therapy

Gene therapy a procedure that involves identification, manipulation, and transference of genetic segments into a host to replace defective genes and to perform desired genetic activities.

Q. Explain Complications in basic inheritance pattern

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BIO-301 GENETICS NOTES FOR FINAL TERM

Single genes -Non-mendelian inheritance

Differences in gene actions can generate more complex inheritance patterns.

Single gene can give codominance, incomplete dominance, overdominance.

Complications to inheritance patterns

Multiple genes - epistasis, polygenic traits Genes & the environment - sex-influenced traits, incomplete penetrance

Complications

1) A common recessive inheritance can give dominant pattern
2) Autosomal dominant inheritance with non-penetrance
3) Autosomal dominant inheritance with variable expression.

Q. What are the Goals/Reasons of pedigree analysis?

1) Determine the mode of inheritance: dominant, recessive, sex-linked.

2) Determine the probability of an affected offspring for a given cross

Q. Define DNA polymorphism and its phenotypic effects.

DNA polymorphism is a sequence difference compared to a reference standard present in at least 1–2% of a population. DNA Polymorphism may be single nucleotide or thousands of BASES. DNA Polymorphism may or may not have phenotypic effects

Q. Polymorphic sequences can serve as markers? Justify

If the location of a polymorphic sequence is known, it can serve as a marker for locating other genes or genetic regions. Each polymorphic marker has different versions or alleles. Polymorphisms can be silent, or be exhibited at levels of Proteins.

linked SNPs outside of gene :- have no effect on protein production or function.

Causative SNPs in gene:-

Non coding SNP: change amount of protein produced.

Co:ding SNP change amino acid sequence

Q. why to study- Polymorphism

Essential to study inheritance patterns. Map phenotypes and anchor genes to the genetic map by co-segregation analysis. Determine change in function: resistant/sensitive populations genetically determined variability among humans is due to a difference in 0.1% of the genomic sequence.

Q. what are the Types of Polymorphic DNA Sequences

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BIO-301 GENETICS NOTES FOR FINAL TERM

RFLP: Restriction Fragment Length Polymorphism

VNTR: Variable Number Tandem Repeats (8 to > 50 base pairs)

STR: Short Tandem Repeats (1– 8 base pairs).

SNP: Single Nucleotide Polymorphisms

AFLP: Amplified Fragment Length Polymorphism

Q. write the Applications of molecular markers

– short range

1) Parentage determination 2) Genetic distance estimation 3) Determination of twin zygosity 4) Sexing of pre-implanted embryos 5) Identification of disease carriers

– Long Range

1) Gene mapping & mapping of QTL by linkage 2) Markers assisted selection of breeds

Q. Explain Restriction Fragment Length Polymorphism and their uses in detailed

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.

Methodology

- Genomic DNA digested with restriction enzymes.
- DNA fragments separated via electrophoresis and transfer to nylon membrane.
- Membranes exposed to probes labeled with P32.
- Film exposed to X-Ray

Uses

- RFLPs can be used in paternity cases or criminal cases to determine the source of a DNA sample. (i.e. it has forensic applications).
- RFLPs can be used to measure recombination rates which can lead to a genetic map with the distance between RFLP loci.
- Direct method for selecting desirable genes such as disease resistance genes

RFLP used to determine disease status

- Determine disease status of an individual, i.e.

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- Huntington
- Cystic fibrosis
- Sickle cell anemia

Mutation detection

- RFLPs can be used to determine the disease status of an individual; (e.g. it can be used in the detection of particular mutations)

Q. Define VNTR

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

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

Q. Write the 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.

Q. writes the Applications of VNTR

1) Microbiology 2) DNA fingerprinting 3) Genetic diversity

4) Forensics 5) Mapping of genomes 6) Breeding patterns of wild or domesticated animals

Q. Explain the Difference between AFLP & RAPD

Amplified Fragment Length Polymorphism	**Random Amplified Polymorphic DNA**
Restriction endonuclease digestion of DNA	PCR based marker with 10-12 base pairs
Ligation of adaptors	Random amplification of several fragments
Amplification of ligated fragments	Amplified fragments run in agarose gel detected by ethidium bromide.
Separation of the amplified fragments via electrophoresis and visualization	Unstable amplification leads to poor repeatability
AFLPs have stable amplification and good repeatability	

Q. Write uses of Molecular Markers

1) Different types of markers used to study diseases 2) DNA forensics

3) QTL traits in animals and plants

Q. Explain SNP- Screening and its importance

People can be grouped based on SNP profile

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BIO-301 GENETICS NOTES FOR FINAL TERM

1) Genome of each individual contains distinct SNP pattern.

2) People can be grouped based on the SNP profile.

SNP - found in coding and noncoding regions

1) SNPs are found in coding and (mostly) noncoding regions.

2) Occur with a very high frequency about 1 in 1000 bases to 1 in 100 to 300 bases.

SNP SCREENING methods: Two different screening strategies

1) Many SNPs in a few individuals 2) A few SNPs in many individuals

SNP Importance

1) In disease diagnosis 2) In finding predisposition to diseases 3) In drug discovery and development

4) In drug responses 5) Investigation of migration patterns 6) Medication and diagnosis at individual level

Q. What is the Frequency of autosomal dominant disorders

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

Q. What is the Location of intronic sequence with respect to exons

Beginning of intron

The number of the last nucleotide of the preceding exon, a plus sign and the position in the intron, like c.88+2T>G

End of intron

The number of the first nucleotide of the following exon, a minus sign and the position upstream in the intron, like c.89-1G>T

Q. What is the function of STRP

STR is simple repeats of nucleotide sequences.

1) Mononucleotide, dinucleotide 2) Used in linkage analysis 3) Used in DNA forensics

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