Topic 7

2022年11月13日 星期日　　下午9:48

Mutation : A sudden permanent change in gene or chromosome in organism and can be passed to next generation.

Gene mutation = A change occurs in the base pairs in the nucleotide sequences in a single gene of a DNA. Amino acid sequences change , protein produced will be differ, differ protein may not function as normal.
Type of gene mutation
1)Base substitution
-One or more base pairs in the nucleotide sequence in a gene are replaced with other base pairs.
-No effect bcuz the change to a base pair may transform (change) one codon into another that encodes 编码 for the same amino acids. (silent mutation)
-Results in the change of amino acids in the polypeptide (missense mutation)
-change a codon into a stop codon which leads to nonfunctional protein. (nonsense mutation)
2)Base insertion
-One or more base pairs are inserted into the nucleotide sequences of DNA.
-Results in frameshift mutation
-That changes all amino acids at and after the point of insertion.
3)Base deletion
-One or more base pairs deleted in the nucleotide sequences of DNA
-Results in frameshift mutation
-That changes all amino acids at and after the point of deletion.

Sickle-cell anaemia
-caused by missense mutation
-bcuz of base substitution
-Base T substituted by base A
-cause the DNA sequence changes from CTC to CAC
-changes the codon GAG to GUG
-The amino acid in polypeptide chain becomes valine (Val) instead of glutamic acid (Glu)
-Haemoglobin become abnormal
-not efficient in transporting oxygen.
-Results in S-shape red blood cells.
-Red blood cells may clump 裹成一块 together and clog the capillaries and less oxygen is transported.

Spontaneous mutation
-mutations that occur naturally which caused by nondisjunction.
-caused by mutagens present in the environment.
-mutagens include cosmic 宇宙 rays, radioactive compounds, heat and caffeine
-Due to errors during DNA replication.

Induced mutation
-mutations where organism is artificially exposed to mutagen physically and chemically.
-caused by 2 types of mutagens
-physical agents : X-rays , UV rays, alfa beta gamma rays
-chemical agents: colchicine
-Lead to genetic variations.

Why base deletion and base insertion more harmful than base substitution?
-Base deletion is a mutation where:one or more base pairs deleted in the nucleotide sequences of DNA.
-Base insertion is a mutation where one or more base pairs inserted in the nucleotide sequences of DNA,
-Both result in frameshift mutations.
-That change all amino acids at and after the point of deletion and insertion.
-Differ amino acid sequences produce differ protein.
-Differ protein may not function as normal.

Chromosomal mutation
=changes in chromosome structure (chromosomal aberration)
=changes in chromosome number / alteration of chromosome number (aneuploidy/euploidy)

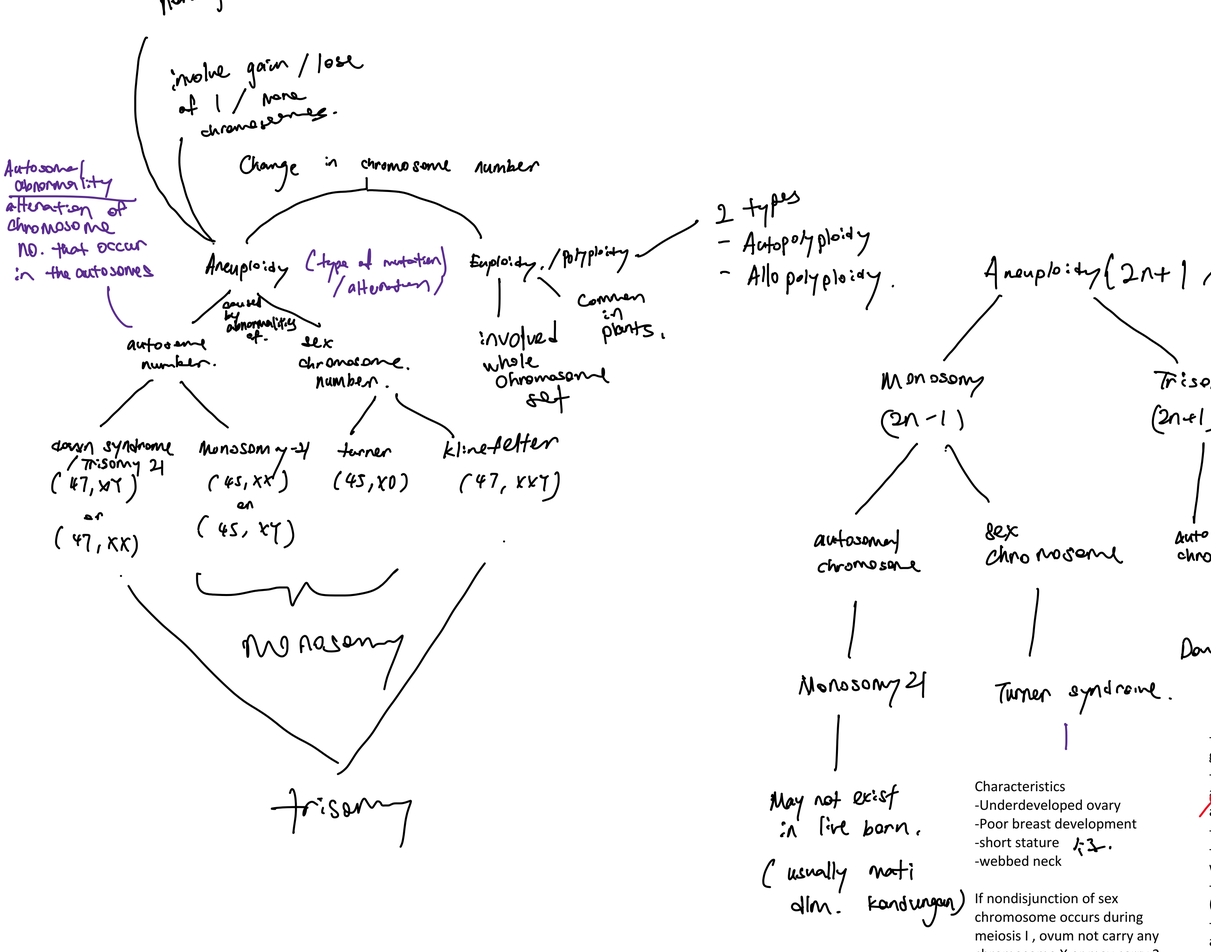
Changes in chromosomal structure due to error during meiosis

1)Translocation (tukar tempat)
-Involves a region of chromosome breaking off and rejoining either the other end of the same chromosome or to another non-homologous chromosome.
-The exchange of a large portion of chromosome 22 with a small fragment from a tip-off chromosome 9 produce a much shortened and easily recognized chromosome 22.
2)Deletion
-Involves a region of chromosome breaks at two places and leads to the loss of the middle segment.
-1 or more genes will be missing from chromosome structure.
-The remaining ends of the chromosome will join again and the chromosome becomes shorter.
3)Inversion
-Involves a region of chromosome breaking off and rotate through 180 before rejoining the chromosomes.
4)Duplication
-Involves a region of chromosome becomes duplicated in the same chromosome
-Results in the addition of a segment in the same chromosome.

Changes in chromosome number
-Chromosomes fail to segregate properly during meiosis due to nondisjunction.
-Nondisjunction : Homologous chromosomes fail to separate during anaphase I or sister chromatid fail to separate during anaphase II.

Cri du chat Syndrome,
= Abnormal larynx development
=Caused by chromosome deletion
=Due to loss of small part of the short arm of chromosome 5
=small head
=distinctive cry resembling mewing kitten

nondisjunction of chromosome.



Down syndrome and Klinefelter /Turner syndrome = Both genetic disorder involve chromosomal number alteration (change of chromosomal number)
Down syndrome = involve nondisjunction of chromosome 21
= autosomal chromosome
Klinefelter /Turner syndrome = involve nondisjunction of chromosome 23
= sex chromosome

Both sterile.

Klinefelter syndrome	Turner syndrome
Due to trisomy (2n+1) of sex chromosome	Due to monosomy (2n-1) of sex chromosome
Genotype is XXY	Genotype is XO
Individual has 47 chromosomes (44+XXY)	Individual has 45 chromosomes (44+XO)
Phenotype is male	Female
-Underdeveloped testes -produce little or no sperm	-Underdeveloped ovary
-soft voice -big breast -long hand and leg	-Poor breast development -short stature -webbed 厚 neck

Nondisjunction
-Nondisjuction occurs when Homologous chromosomes fail to separate during anaphase I or sister chromatid fail to separate during anaphase II
-If homologous chromosomes fail to separate during meiosis I, produce 2 gametes that lack that chromosome (n-1) and 2 gametes with 2 copies of the chromosome (n+1). (2:2)
-If sister chromatids fail to separate during meiosis II, produce 1 gamete that lacks that chromosome (n-1) and 2 normal gametes with 1 copy of the chromosome (n) and 1 gamete with 2 copies of the chromosome (n+1). (1:2:1)
-Individual with an error in chromosome number is described as aneuploidy, a term that includes monosomy (loss of one chromosome) and trisomy (gain of one chromosome).

• Autosomal abnormalities is the type of mutation (aneuploidy) that occurs to the autosomal chromosomes (other than sex chromosomes)
• Due to nondisjunction that occurs during meiosis
•This will produce a gamete that is lack or has extra chromosomes
•When this gamete fertilize with a normal gamete, it will produce a mutant individual
• The most common examples for aneuploidy in autosomal chromosomes are monosomy 21 syndrome and trisomy 21 (Down syndrome)
• In monosomy 21, the individual will lack one chromosome 21
• The genome for this individual is 2n - 1
• It is characterised by short distance between eyes, large ears and contracted muscle, short neck and generally lethal (mostly die between 3 weeks and 20 months of living)
• In trisomy 21 or also known as Down syndrome, the individual will have extra chromosome 21
• The genome for this individual is 2n + 1
• It is characterised by short stature, broad hands, stubby fingers and toes, a wide rounded face, a large protruding tongue that makes speech difficult.

Euploidy/ Polyploidy
-A condition where an organism possesses more than 2 complete sets of chromosomes.
-Occurs due to the fertilization of an abnormal gamete with a normal gamete.
-2 types = autopolyploidy and allopolyploidy
-Autopolyploidy = results from same species interbreeding/self-fertilize and combining their chromosomes to become more than 2 chromosomes set. Can be induced by using colchicines. Exp: bananas (triploid,3n) , potato (tetraploidy,4n) , strawberry (octaploid,8n)
-Allopolyploidy = chromosome number in a sterile hybrids become doubled and produce fertile hybrids. F1 hybrid produced from differ species are usually sterile.