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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.
                                                                                                                                       Spontaneous mutation
                                                                                                                                       -mutations that occur naturally which caused by nondisjunction.
                                                                                                                                       -caused by mutagens present in the environment.
       Gene mutation = A change occurs in the base pairs in the nucleotide sequences
                                                                                                                                       -mutagens include cosmic 宇宙 rays, radioactive compounds, heat and caffeine
       in a single gene of a DNA. Amino acid sequences change, protein produced will
       be differ, differ protein may not function as normal.
                                                                                                                                       -Due to errors during DNA replication.
       Type of gene mutation
       1)Base substitution
                                                                                                                                       Induced mutation
       -One or more base pairs in the nucleotide sequence in a gene are replaced with
                                                                                                                                       -mutations where organism is artificially exposed to mutagen physically and chemically.
                                                                                  4
                                                                                                                                       -caused by 2 types of mutagens
       other base pairs.
       -No effect bouz the change to a base pair may transform (change) one
                                                                                                                                       -physical agents : X-rays , UV rays, alfa beta gamma rays
                                                                                                                                       -chemical agents: colchicine
       codon into another that encodes 编码 for the same amino acids. (silent
                                                                                                                                       -Lead to genetic variations.
       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
                                                                                                                                       Why base deletion and base insertion more harmful than base substitution?
                                                                                    3
       -One or more base pairs are inserted into the nucleotide sequences of DNA.
                                                                                                                                       -Base deletion is a mutation where:one or more base pairs deleted in the nucleotide sequences of DNA.
       -Results in frameshift mutation
                                                                                                                                       -Base insertion is a mutation where one or more base pairs inserted in the nucleotide sequences of
       -That changes all amino acids at and after the point of insertion.
                                                                                                                                       DNA,
       3)Base deletion
                                                                                                                                       -Both result in frameshift mutations.
       -One or more base pairs deleted in the nucleotide sequences of DNA
                                                                                                                                       -That change all amino acids at and after the point of deletion and insertion.
       -Results in frameshift mutation
                                                                                                                                      -Differ amino acid sequences produce differ protein.
       -That changes all amino acids at and after the point of deletion.
                                                                                                                                       -Differ protein may not function as normal.
       Sickle-cell anaemia
       -caused by missense mutation
       -bcuz of base substitution
                                                                 1(P)
       -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 裏成一块 together and clog the capillaries and less
       oxygen is transported.
       =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
                                                                                                                                   P/2 .
       chromosome 22.
                                                                                                                       = Abnormal larynx development
       2)Deletion
                                                                                      Cr. du chor Syndrowe = Caused by chromosome 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.
                                                                                                                       =small head
      -The remaining ends of the chromosome will join again and the chromosome
                                                                                                                       =distinctive cry resembling mewing kitten
       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.
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                                                  chromo some number
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Obnorman 1:ty
atteration
chromoso me
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NO. that occur
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X meiosis II,
                                                                                      in plants,
                 autoseme
                                                                    whole
Ohronosome
                   number.
                                           chromosome.
                                                                                                                              M on osomy
                                                                                                                                                                        Trisomy
                                             number
                                                                                                                                                                       (2n+1)
                                                                                                                                (2n-1)
                                                               k1:ne-telter
     down syndrome
/Trisomy 21
                            K-4 mascher
                                                                                                                                                                                                                                           Sperm will hv abnormal sex chromosome with genotype (22+XY)
                                                                                                                                                                                                                                           -If abnormal sperm with genotype (22+XY) is fertilized with normal ovum with genotype (22+X)
                                                                  (47, KXT)
       (47,×Y)
                                                (45, \chi 0)
                             (45, XX)
                                                                                                                                                                                                                                          Klinefelter syndrome is produced.
                            ( 45, 27)
                                                                                                                                                                          auto soma
      (47, KK)
                                                                                                                     autosona
                                                                                                                                               chronosem
                                                                                                                                                                           chromosome
                                                                                                                      chromosone
                                                                                                                                                                                                         Klinefelter
                                 Mo hosom,
                                                                                                                                                                                                                                      Characteristics
                                                                                                                                                                           Down-syndrong/
Trisonny 21
                                                                                                                                                                                                                                      -Small testis
                                                                                                                                                                                                            Syndrom
                                                                                                                                                                                                                                      -soft voice
                                                                                                                   Norosomy 21
                                                                                                                                                                                                                                      -big breast
                                                                                                                                                                                                                                      -long legs and arms
                                                                                                                                                                                                     ection of chromosomes 21 during
                                                                                                                                                                                -Occurs when nondisju-
                                                                                                                                                                                -Homologous chromosomes fail to separate during
                                                                                                                                             Characteristics
                                                                                                                                                                                maphase or sister chromatid fail to separate during
                                                                                                               May not exist in live born.
                                                                                                                                             -Underdeveloped ovary
                                                                                                                                                                               anaphase II
                                                                                                                                              -Poor breast development
                                                                                                                                                                                -The gametes formed by extra chromosome
                                                                                                                                             -short stature たる.
                                                                                                                                                                                -A normal gamete (n) fertilize with an abnormal gamete
                                                                                                                                              -webbed neck
                                                                                                                                                                                with an extra chromosome 21 (n+1)
                                                                                                                                                                                -The zygote would have an extra chromosome 21
                                                                                                                                             If nondisjunction of sex
                                                                                                                  dlm. Kandungan)
                                                                                                                                                                                (genome is 2n+1)
                                                                                                                                              chromosome occurs during
                                                                                                                                                                                -Individual with 47 chromosome (instead of 46),
                                                                                                                                              meiosis I, ovum not carry any
                                                                                                                                                                                appearance of 3 chromosome 21
                                                                                                                                             chromosome X or may carry 2
                                                                                                                                             chromosome X.
                                                                                                                                                                             Characteristics
                                                                                                                                             If abnormal ovum w/o X
                                                                                                                                             chromosome fertilize with normal
                                                                                                                                                                                -A wide rounded face
                                                                                                                                             sperm(X), Turner syndrome will be
                                                                                                                                                                                -stubby fingers and toes
                                                                                                                                             produced.
                                                                                                                                                                                 49.3
                                                                                                                                              If abnormal ovum w/o X
                                                                                                                                             chromosome fertilize with normal
                                                                                                                                             sperm(Y), YO will dies.
                                                                                             alteration
                                                                                                                Nondisjunction
      Down syndrome and Klinefelter /Turner syndrome = Both genetic disorder involve chromosomal

    Autosomal abnormalities is the type of mutation (aneuploidy)

                                                                                                                 -Nondisjuction occurs when Homologous chromosomes fail to separate during anaphase I or sister
                                                                                                                                                                                                                                      that occurs to the autosomal chromosomes (other than sex
       number alteration (change of chromosomal number)
                                                                                                                chromatid fail to separate during anaphase II
                                                                                                                                                                                                                                      chromosomes)
      Down syndrome = involve nondisjunction of chromosome 21 = autosomal chromosome
                                                                                                                -If homologous chromosomes fail to separate during meiosis I, produce 2 gametes that lack that
                                                                                                                                                                                                                                      • Due to nondisjunction that occurs during meiosis
                                                                                                                chromosome (n-1) and 2 gametes with 2 copies of the chromosome (n+1). (2:2)
                                                                                                                                                                                                                                      •This will produce a gamete that is lack or has extra
      Klinefelter /Turner syndrome = involve nondisjunction of chromosome 23
                                                                                                                -If sister chromatids fail to separate during meiosis II, produce 1 gamete that lacks that chromosome
                                                                                                                                                                                                                                      chromosomes
                                  = sex chromosome
                                                                                                                (n-1) and 2 normal gametes with 1 copy of the chromosome (n) and 1 gamete with 2 copies of the
                                                                                                                                                                                                                                      •When this gamete fertilize with a normal gamete, it will produce a
                                                                                                                chromosome (n+1). (1:2:1)
                                                                                                                                                                                                                                      mutant individual
      Both sterile. Not able to produce
                                                  RMOOK
                                                                                                                -Individual with an error in chromosome number is described as an uploidy, a term that includes
                                                                                                                                                                                                                                      • The most common examples for an uploidy in autosomal
       Klinefelter syndrome
                                               Turner syndrome
                                                                                                                monosomy (loss of one chromosome) and trisomy (gain of one chromosome).
                                                                                                                                                                                                                                      chromosomes are monosomy 21 syndrome and trisomy 21
      Due to trisomy (2n+1) of sex chromosome Due to monosomy (2n-1) of sex chromosome
                                                                                                                                                                                                                                      (Down syndrome)
                                                                                                                                                                                                                                      • In monosomy 21, the individual will lack one chromosome 21
                                               Genotype is XO
       Genotype is XXY
                                                                                                                                                                                                                                      • The genome for this individual is 2n - 1
       Individual has 47 chromosomes (44+XXY) Individual has 45 chromosomes (44+XO)
                                                                                                                                                                                                                                      • It is characterised by short distance between eyes, large ears and
                                                                                                                                                                                                                                      contracted muscle, short neck and generally lethal (mostly die
       Phenotype is male
                                                                                                                                                                                                                                      between 3 weeks and 20 months of living)
       -Underdeveloped testes
                                               -Underdeveloped ovary
                                                                                                                                                                                                                                      • In trisomy 21 or also known as Down syndrome, the individual
       -produce little or no sperm
                                                                                                                                                                                                                                      will have extra chromosome 21
                                                                                                                                                                                                                                      • The genome for this individual is 2n + 1
       -soft voice
                                               -Poor breast development
       -big breast
                                               -short stature
                                                                                                                                                                                                                                      • It is characterised by short stature, broad hands, stubby fingers
       -long hand and leg
                                               -webbed 厚 neck
                                                                                                                                                                                                                                      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.
                        2092
                                                                                ( V:31)
                                    (U:30)
          G:
           F. :
                                            - Herile hyurid
                                                no hamplegons de meierras
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- who has homologous paid - In: 122

MMAA

- Fertile hybrid

- n+n >61

Chromosone.

Doubling the chronosom so. (by Entury col chicines)