Gene Mutations and Nondisjunction

- sometimes characteristics in organisms are not traceable to their ancestors

= **mutation:** a change in the genetic makeup of a chromosome which can be passed on

- mutations may affect

i) Somatic Cells (if unchecked results in cancer)

ii) Gametes (resulting embryo would have characteristics not found in either parent)

- There are 2 types of mutations:

a) gene mutation

- caused by portions of the DNA which make up chromosomes being

1) deleted - loss of 1 or more genes on a chromosome

2) duplicated - 1 or more genes repeated

3) inverted - the order of 1 or more genes on a chromosome being changed

4) translocated - 1 or more genes interchanged with other gene(s) on the same or another chromosome

- the change is not phenotypical (visible)

- usually results in an enzyme deficiency

ie. lactose intolerant

b) chromosomal mutation

- caused by:

1) nondisjunction – failure of chromosomes to split during meiosis resulting in gametes having an extra (or too few) chromosomes

2) translocation or deletion of an arm of a chromosome

- the change is phenotypical = organism looks different from the rest of the species

ie. Downs Syndrome (results of an extra 21st chromosome = Trisomy 21)

- mutations are generally recessive and are passed on to offspring

:some mutations are beneficial, some are lethal

:beneficial mutations become adaptations enhancing the survival of an organism

= basis of Darwin's Theory of Natural Selection

- the rate spontaneous (natural) mutations is very low -- about 1 per 1,000,000 genes
- the rate of mutations can be increased as a result of mutagenic agents such as temp., chemicals, radiation, etc.

= **induced mutations** - resulting mutations are the same as those from spontaneous mutations, just more frequently

: ie) Atomic bombs at Hiroshima and Nagasaki

- **Karyotype**= a diagnostic tool used to identify chromosomal abnormalities

- a micrograph picture is taken of the chromosomes during Prophase 1 of Meiosis
- the chromosomes are arranged by size and identified by number
- used to diagnose Syndromes and Disorders

Sample Nondisjunction Disorders:

A) Down Syndrome (47 chromosomes)
= extra autosome 21
: diminished mental capacity, round face, enlarged tongue, short in height
: incidence = 1 in 600 births with risk increasing as the age of the mother increases

B) Turner Syndrome (45 chromosomes = X)
= missing 1 X chromosome caused by nondisjunction in the egg so the single X chromosome is provided by the sperm
: are females that do not develop sexually, are short, have widened necks
: incidence = only 1 in 10,000 births as most fetuses miscarry before the 20th week of pregnancy

C) Klinefelter Syndrome (47 chromosomes = XXY)
= extra X chromosome caused by nondisjunction in the egg or sperm
: are males which possess both male and female characteristics
: are sterile
: incidence = 1 in 1000 births

D) Trisomy X (47 chromosomes = XXX)
: wide range of characteristics including basic male traits, mental impairment, short, sterile

E) Trisomy Y (47 chromosomes = XYY)
: "super males"
: taller than average, learning difficulties
: once believed more aggressive and violent = genetic criminals (based on #'s in prison) = false (is due to learning)

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  = **mutation:** _____
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  1) _____- loss of 1 or more genes on a chromosome
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b**) chromosomal mutation**

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  :some mutations are beneficial, some are lethal
  :beneficial mutations become _____ enhancing the survival of an organism
  = basis of _____

- the rate spontaneous (natural) mutations is very low -- about 1 per 1,000,000 genes
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  = _____- resulting mutations are the same as those from spontaneous mutations, just more frequently
  : ie) _____

- Karyotype= _____

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Nondisjunction (Bridges)

- a.k.a. Abnormal Segregation
  : mutation caused by both chromosomes of a pair going to 1 pole of the spindle during the first division of meiosis
  = the wrong number of chromosomes in each gamete
- rarely, the gamete survives and fertilizes another gamete
- if the zygote survives, the organism is either lacking 1 chromosome or has 1 extra chromosome
  : in humans = 45 chromosomes or 47 chromosomes
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    : diminished mental capacity, round face, enlarged tongue, short in height
    : incidence = 1 in 600 births with risk increasing as the age of the mother increases
  B) Turner Syndrome (45 chromosomes = X)
    = deficient 1 X chromosome caused by nondisjunction in the egg (single X provided by the sperm)
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