

CONCEPT MAP OF GENETIC DISORDERS (most of this info is also in your notes. Some information is added here so you can understand examples of each category. If you don't see a disease in your notes, you won't have to know it for the exam.)

Genetic Disorders

Two categories—chromosomal aberrations & single gene disorders

Single gene disorders (next page)

Chromosomal aberrations

Key characteristics (KC):

- one or more chromosomes damaged
- often caused by clastogens such as x-rays, drugs, viruses, free radicals
- 2 sub-categories:
 - alterations in number (aneuploidy)
 - alterations in structure

Alterations in structure

KC:

- when parts of chromosomes are deleted, duplicated, or rearranged
- one type of rearrangement is called translocation
- an example of translocation—Philadelphia chromosome

Alterations in number (aneuploidy)

KC:

- caused by non-disjunction—failure to divide properly
- non-disjunction results in too few chromosomes (45) or too many (47)

too few—
monosomy

too many—
polysomy

Turner's Syndrome

KC:

- presence of only 45 chromosomes—
one X instead of XX

Trisomy 21 (Down's syndrome)

KC:

- 21st chromosome set has 3 instead of two & this causes malfunction of the genes that code for normal development.

S&S (phenotype):

- mental retardation
- epicanthic fold
- short limbs
- large tongue

Philadelphia chromosome → CML

KC:

- results from translocation
- part of a particular chromosome "breaks off" & attaches to another
- the left over, deformed, piece is known as the Philadelphia chromosome
- if a person has this chromosome, will more likely have CML—chronic myelocytic leukemia
- CML occurs because the genes on the once-healthy chromosome control WBC production—now those genes damaged → WBC production "goes crazy" in bone marrow → huge number of WBCs in blood (leukocytosis) → diagnosis of leukemia.

Genetic Disorders Concept Mapping Answers Key

S Nieto



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