

**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:

- 21<sup>st</sup> 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

**Denise R. Ferrier**



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