

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

L Reisser



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Table of Contents Genetic Disorders Concept Mapping Answers Key

1. Understanding the eBook Genetic Disorders Concept Mapping Answers Key
 - The Rise of Digital Reading Genetic Disorders Concept Mapping Answers Key
 - Advantages of eBooks Over Traditional Books
2. Identifying Genetic Disorders Concept Mapping Answers Key
 - Exploring Different Genres
 - Considering Fiction vs. Non-Fiction
 - Determining Your Reading Goals
3. Choosing the Right eBook Platform
 - Popular eBook Platforms
 - Features to Look for in an Genetic Disorders Concept Mapping Answers Key
 - User-Friendly Interface

4. Exploring eBook Recommendations from Genetic Disorders Concept Mapping Answers Key
 - Personalized Recommendations
 - Genetic Disorders Concept Mapping Answers Key User Reviews and Ratings
 - Genetic Disorders Concept Mapping Answers Key and Bestseller Lists
5. Accessing Genetic Disorders Concept Mapping Answers Key Free and Paid eBooks
 - Genetic Disorders Concept Mapping Answers Key Public Domain eBooks
 - Genetic Disorders Concept Mapping Answers Key eBook Subscription Services
 - Genetic Disorders Concept Mapping Answers Key Budget-Friendly Options
6. Navigating Genetic Disorders Concept Mapping Answers Key eBook Formats
 - ePub, PDF, MOBI, and More
 - Genetic Disorders Concept Mapping Answers Key Compatibility with Devices
 - Genetic Disorders Concept Mapping Answers Key Enhanced eBook Features
7. Enhancing Your Reading Experience
 - Adjustable Fonts and Text Sizes of Genetic Disorders Concept Mapping Answers Key
 - Highlighting and Note-Taking Genetic Disorders Concept Mapping Answers Key
 - Interactive Elements Genetic Disorders Concept Mapping Answers Key
8. Staying Engaged with Genetic Disorders Concept Mapping Answers Key
 - Joining Online Reading Communities
 - Participating in Virtual Book Clubs
 - Following Authors and Publishers Genetic Disorders Concept Mapping Answers Key
9. Balancing eBooks and Physical Books Genetic Disorders Concept Mapping Answers Key
 - Benefits of a Digital Library
 - Creating a Diverse Reading Collection Genetic Disorders Concept Mapping Answers Key
10. Overcoming Reading Challenges
 - Dealing with Digital Eye Strain
 - Minimizing Distractions
 - Managing Screen Time
11. Cultivating a Reading Routine Genetic Disorders Concept Mapping Answers Key
 - Setting Reading Goals Genetic Disorders Concept Mapping Answers Key
 - Carving Out Dedicated Reading Time

12. Sourcing Reliable Information of Genetic Disorders Concept Mapping Answers Key
 - Fact-Checking eBook Content of Genetic Disorders Concept Mapping Answers Key
 - Distinguishing Credible Sources
13. Promoting Lifelong Learning
 - Utilizing eBooks for Skill Development
 - Exploring Educational eBooks
14. Embracing eBook Trends
 - Integration of Multimedia Elements
 - Interactive and Gamified eBooks

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