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 Genetic Disorders disease in your notes, you won't have to know it for the exam.) Two categories—chromosomal aberrations & single gene disorders Chromosomal aberrations Key characteristics (XC): Single gene disorders (next page) · one or more chromosomes damaged often caused by clastogens such as xravs, drugs, viruses, free radicals Alterations in structure KC: 2 sub-categories: alterations in number (aneuploidy) when parts of chromosomes are deleted, duplicated, or rearranged one type of rearrangement is called translocation alterations in structure an example of translocation-- Philadelphia chromosome. Alterations in number (aneuploidy) KC: Philadelphia chromosome -> CML caused by non-disjunction-failure to divide properly KC: non-disjunction results in too few chromosomes (45) or too results from translocation. many (47): part of a particular chromosome "breaks off" & attaches to another. · the left over, deformed, piece is known as the Philadelphia too fewtoo manychromosome monosomy polysomy If a person has this chromosome, will more likely have. CML—chronic myelocytic leukemia · CML occurs because the genes on the once-healthy Turner's Syndrome Trisomy 21 (Down's syndrome) chromosome control WBC production-now those genes KCC: RC: damaged -> WBC production "goes crazy" in bone marrow -> 31" chromosome set has 3. presence of only: huge number of WBCs in blood (leukocytosis)-> diagnosis of instead of two & this causes. 45 chromosomes leukemia. -one X instead of XX malfunction of the genes that code for normal development. 58/5 (phenotypel): mental retardation epicanthic fold. short limbs large tongue-

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**Jin-Ying Zhang** 

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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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