

CHAPTER – 8

Genetic Disorder

EXERCISES

2 Mark Questions

Q1: Explain the difference between a genetic disorder and a chromosomal disorder.

Answer: A genetic disorder is a condition caused by abnormalities or mutations in individual genes, affecting specific traits or functions. In contrast, a chromosomal disorder results from structural or numerical abnormalities in chromosomes, impacting a larger set of genes.

Q2: Define autosomal recessive inheritance and provide an example of a genetic disorder following this pattern.

Answer: Autosomal recessive inheritance means that two copies of an abnormal gene (one from each parent) are required for the disorder to manifest. An example is cystic fibrosis, where individuals with two copies of the defective CFTR gene exhibit the disorder.

Q3: What is the significance of carrier testing in the context of genetic disorders?

Answer: Carrier testing helps identify individuals who carry one copy of a mutated gene associated with a genetic disorder. While carriers typically do not show symptoms, they have the potential to pass the mutated gene to their offspring, raising awareness of the risk of the disorder in their family.

Q4: Briefly explain the role of genetic counseling in the context of genetic disorders.

Answer: Genetic counseling involves providing information and support to individuals or families who have or are at risk of genetic disorders. Counselors help clients understand the inheritance patterns, assess the risk of passing on genetic conditions, and make informed decisions regarding testing or family planning.

Q5: Define the term "genetic mutation" and explain how mutations contribute to genetic disorders.

Answer: A genetic mutation is a change in the DNA sequence. Mutations can disrupt the normal functioning of genes, leading to genetic disorders. Depending on the type of mutation (e.g., point mutation, deletion, insertion), the resulting abnormal gene may produce a non-functional protein or alter the regulation of other genes.

4 Mark Questions

Q1: Explain the difference between congenital and hereditary genetic disorders. Provide examples of each.

Answer: Congenital Genetic Disorders: These disorders are present at birth and may result from genetic, environmental, or unknown factors. Examples include Down syndrome (trisomy 21) and congenital heart defects.

Hereditary Genetic Disorders: These disorders are passed down from parents to their offspring through genetic information. Examples include cystic fibrosis, hemophilia, and Huntington's disease.

Q2: Describe the concept of incomplete dominance with reference to a genetic disorder. Provide an example.

Answer: In incomplete dominance, neither allele is completely dominant over the other, resulting in an intermediate phenotype in heterozygous individuals. An example is familial hypercholesterolemia, a genetic disorder where individuals with one normal and one mutated allele show intermediate cholesterol levels, impacting cardiovascular health.

Q3: Discuss the challenges and ethical considerations associated with genetic testing for predicting the risk of genetic disorders.

Answer:

Challenges: Challenges include the potential psychological impact of knowing one's risk, the accuracy of test results, and the uncertainty of predicting the onset and severity of disorders, and the potential for genetic discrimination.

Ethical Considerations: Ethical concerns involve issues of informed consent, confidentiality, privacy, the potential for stigmatization, and ensuring equitable access to genetic testing. Counseling and support services are crucial in addressing these considerations.

Q4: Explain the concept of X-linked recessive inheritance. Provide an example of a genetic disorder following this inheritance pattern.

Answer:

X-linked recessive inheritance involves the transmission of a genetic disorder through a recessive gene carried on the X chromosome. Males are more commonly affected because they have only one X chromosome. An example is hemophilia, where the gene for blood clotting factor is located on the X chromosome. Females are typically carriers, while males with one affected X chromosome exhibit the disorder.

7 Mark Questions

Q1: Discuss the impact of genetic disorders on individuals and families, addressing both physical and psychological aspects.

Answer:

Genetic disorders can have profound effects on individuals and their families. Physically, affected individuals may experience a range of symptoms, from mild to

severe, depending on the disorder. This can lead to challenges in daily life, medical complications, and the need for ongoing healthcare. Psychologically, individuals may face emotional distress, anxiety, depression, and challenges related to self-esteem. Families may experience stress, financial burdens, and the need for extensive caregiving. Genetic counseling and support services play a crucial role in addressing both the physical and psychological aspects, providing information, guidance, and assistance in coping with the impact of genetic disorders.

Q2: Examine the role of gene therapy in treating genetic disorders. Highlight the potential benefits, challenges, and ethical considerations associated with gene therapy.

Answer:

Benefits of Gene Therapy:

- Correction or replacement of defective genes.
- Potential for treating a wide range of genetic disorders.
- Possibility of preventing the onset or progression of diseases.

Challenges of Gene Therapy:

- Technical challenges in delivering genes to target cells.
- Immune response against the therapeutic gene.
- Off-target effects and unintended consequences.

Ethical Considerations:

- Ensuring informed consent and voluntary participation.
- Addressing concerns about germline gene editing and heritability.
- Balancing the potential benefits with the unknown long-term effects.

Gene therapy holds promise in revolutionizing the treatment of genetic disorders, but ongoing research and careful consideration of ethical issues are crucial to its responsible development and application.

Q3: Discuss the concept of carrier screening in the context of genetic disorders. Outline its purpose, methods, and potential impact on family planning.

Answer:

Purpose of Carrier Screening:

- Identifying individuals carrying one copy of a mutated gene.
- Assessing the risk of having a child with a recessive genetic disorder.

Methods of Carrier Screening:

- Blood tests or DNA analysis to identify specific genetic mutations.
- Testing both partners to assess the risk of passing on recessive genes.

Impact on Family Planning:

- Providing information for informed decision-making about having children.
- Offering options like pre-implantation genetic diagnosis or prenatal testing.
- Facilitating discussions about potential risks and alternatives.

Fill in the Blanks

1. _____ disorders are present at birth and may result from genetic, environmental, or unknown factors.

Answer: Congenital

2. In autosomal recessive inheritance, an individual must inherit _____ copies of an abnormal gene to manifest the disorder.

Answer: Two

3. _____ testing helps identify individuals who carry one copy of a mutated gene associated with a genetic disorder.

Answer: Carrier

4. Gene therapy involves the correction or replacement of _____ genes to treat genetic disorders.

Answer: Defective

5. X-linked recessive inheritance involves the transmission of a genetic disorder through a recessive gene located on the _____ chromosome.

Answer: X

Multiple Choice Questions

1. What is the primary cause of genetic disorders?

- a) Environmental factors
- b) Lifestyle choices
- c) Mutations in genes
- d) Bacterial infections

Answer: c) Mutations in genes

2. Which term describes a genetic disorder that is present at birth?

- a) Congenital
- b) Hereditary
- c) Acquired
- d) Somatic

Answer: a) Congenital

3. In autosomal recessive inheritance, an individual must inherit the mutated gene from:

- a) Only the mother
- b) Only the father
- c) Both parents
- d) Either parent

Answer: c) Both parents

4. What is the role of carrier testing in the context of genetic disorders?

- a) Diagnosing affected individuals
- b) Identifying individuals carrying one copy of a mutated gene
- c) Predicting the severity of disorders
- d) Determining the gender of the affected individual

Answer: b) Identifying individuals carrying one copy of a mutated gene

5. Which of the following disorders exhibits X-linked recessive inheritance?

- a) Down syndrome
- b) Cystic fibrosis
- c) Hemophilia
- d) Huntington's disease

Answer: c) Hemophilia

6. Gene therapy primarily aims to:

- a) Prevent the onset of genetic disorders
- b) Correct or replace defective genes
- c) Induce mutations in genes

d) Enhance the expression of normal genes

Answer: b) Correct or replace defective genes

7. What is the purpose of genetic counseling in the context of genetic disorders?

a) Diagnosing disorders

b) Administering gene therapy

c) Providing information and support

d) Conducting carrier testing

Answer: c) Providing information and support

8. Which term describes the situation where neither allele is completely dominant, resulting in an intermediate phenotype in heterozygous individuals?

a) Recessive

b) Dominant

c) Incomplete dominance

d) Co dominance

Answer: c) Incomplete dominance

9. The central dogma of molecular biology outlines the flow of genetic information as:

a) DNA replication -> Translation -> Transcription

b) Translation -> DNA replication -> Transcription

c) Transcription -> Translation -> DNA replication

d) DNA replication -> Transcription -> Translation

Answer: d) DNA replication -> Transcription -> Translation

SUMMARY:

These chapter likely dives into the fascinating world of inherited diseases caused by variations in our genetic makeup. Let's take a quick peek:

1. Chromosomal Abnormalities:

- Extra or missing chromosomes, like in Down syndrome or Turner syndrome, disrupt gene balance and cause developmental problems.
- Structural changes, like translocations or deletions, can alter gene function and lead to various disorders.

2. Single-Gene Disorders:

- Caused by mutations in a single gene, such as cystic fibrosis or sickle cell anemia.
- These mutations can be dominant or recessive, influencing inheritance patterns and disease severity.

3. Multifactorial Disorders:

- Complex diseases like cancer or heart disease arise from interplay between multiple genes and environmental factors.
- Understanding these interactions is crucial for risk assessment and developing preventive strategies.

4. Genetic Testing:

- Techniques like karyotyping, DNA sequencing, and gene panels help diagnose genetic disorders and predict disease risk.
- Ethical considerations and genetic counseling play a vital role in utilizing these tests responsibly.

5. Future of Genetic Medicine:

- Gene therapy, gene editing, and personalized medicine hold promise for treating and potentially curing genetic disorders.