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SNS COLLEGE OF NURSING

COIMBATORE-35

PROGRAMME	**: B.Sc. Nursing II Year**
COURSE	**: Pathology II and Genetics**
SEMESTER	**:III**
UNIT	**: VI**
TOPIC	**: Genetic Disorders**
PREPARED BY	**: Mrs. Sornambiga R., Asst Prof**

OBJECTIVES

After the lecture, the students will be able to describe the various genetic disorders, develop a desirable attitude and apply them in clinical practice

OVERVIEW OF GENETIC DISORDERS

Definition: Diseases caused by genetic abnormalities

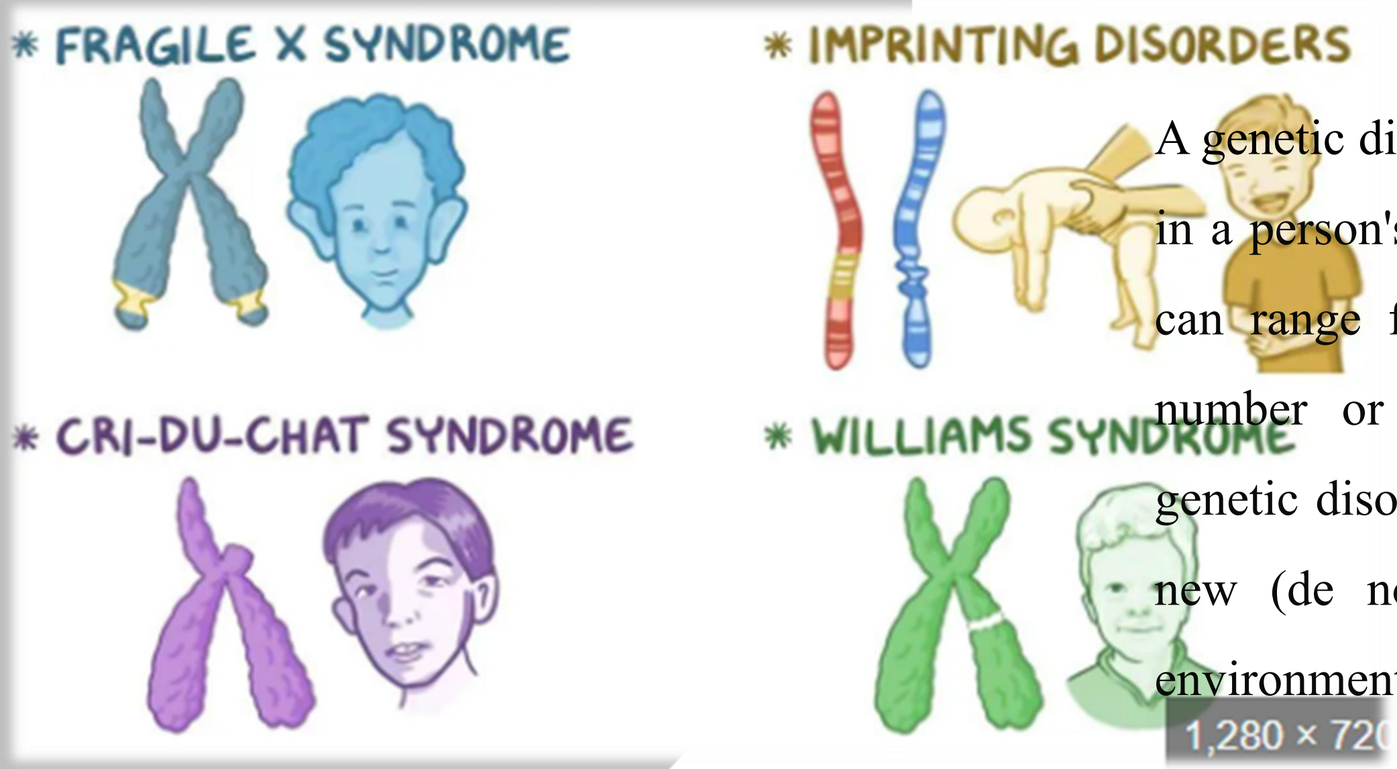
Types: Single-gene, chromosomal, multifactorial

Prevalence: ~1 in 2,500 newborns have genetic disorders

Causes: Mutations, chromosomal errors, and environmental factors

Health Impact: Physical, developmental, systemic effects

INTRODUCTION OF GENETIC DISORDERS



A genetic disorder is a health condition caused by abnormalities in a person's genetic material, or genome. These abnormalities can range from a single gene mutation to a change in the number or structure of entire chromosomes. While some genetic disorders are inherited from parents, others result from new (de novo) mutations that occur randomly or due to environmental factors.

OVERVIEW OF GENETIC DISORDERS

- Examples: Cystic fibrosis, Down syndrome, diabetes
- Diagnosis: Genetic testing, clinical evaluation
- Treatment: Symptomatic, gene therapy (emerging)
- Nursing Role: Assessment, education, support
- Importance: Affects quality of life, healthcare needs

CASE STUDY: GENETIC DISORDER

- **Age:** 8-year-old female child
- **Presenting Complaints:** Delayed milestones, difficulty in learning, short stature, and webbed neck
- **Birth History:** Normal vaginal delivery, birth weight slightly below average
- **Family History:** Non-consanguineous marriage; no similar complaints in siblings

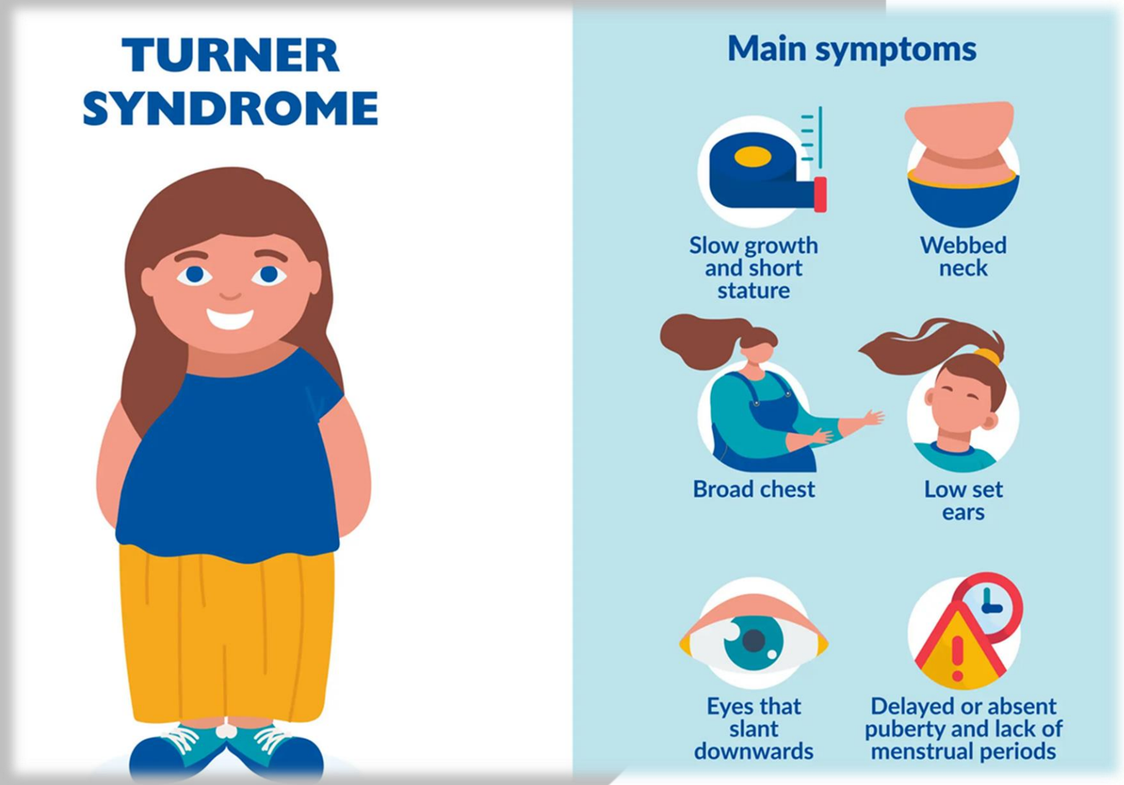
CLINICAL EXAMINATION

- Short stature (< 3rd percentile for age)
- Broad chest with widely spaced nipples
- Webbed neck
- Low hairline at the back
- Poorly developed secondary sexual characteristics
- Intelligence: Normal

INVESTIGATIONS

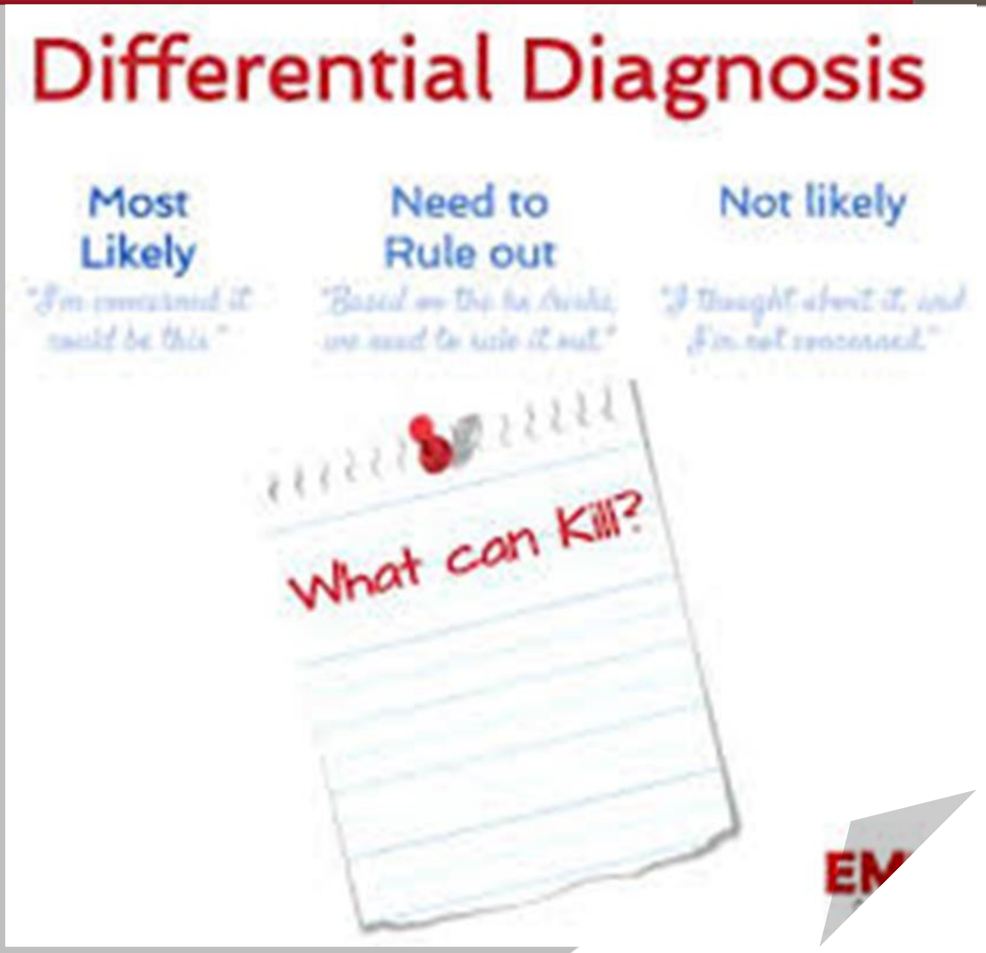
- **Karyotyping:** 45, X (Monosomy X)
- **Echocardiography:** Coarctation of aorta
- **Ultrasound abdomen:** Streak ovaries
- **Hormonal assay:** Low estrogen, elevated FSH & LH

PROVISIONAL DIAGNOSIS



👉 *Turner Syndrome (Monosomy X – Genetic Disorder)*

DIFFERENTIAL DIAGNOSIS



- Noonan syndrome
- Klinefelter syndrome (in males, 47, XXY)
- Constitutional short stature

MANAGEMENT-SUPPORTIVE CARE

Growth hormone therapy to improve height

Estrogen replacement therapy for secondary sexual characteristics & bone health

Cardiac surgery if required (for coarctation of the aorta)

OUTCOME & COUNSELING

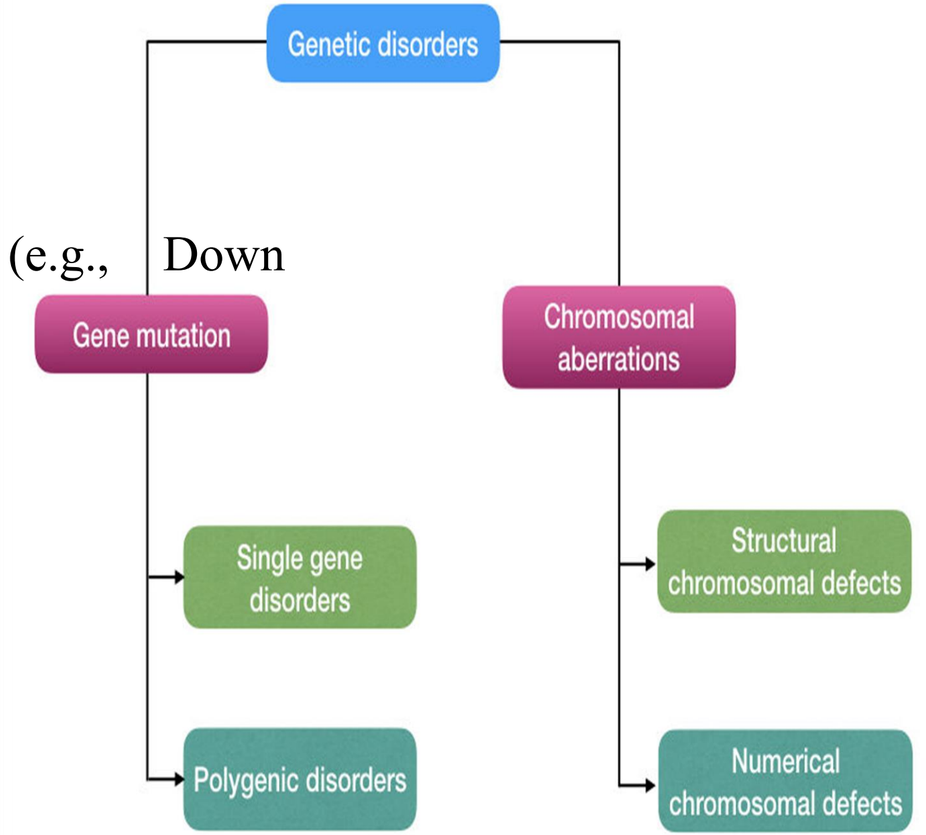
•With early diagnosis and hormone therapy, quality of life improves significantly

•Normal intelligence allows independent living with proper medical care

•Genetic counseling for family – recurrence risk is very low (usually sporadic, not inherited)

TYPES OF GENETIC DISORDERS

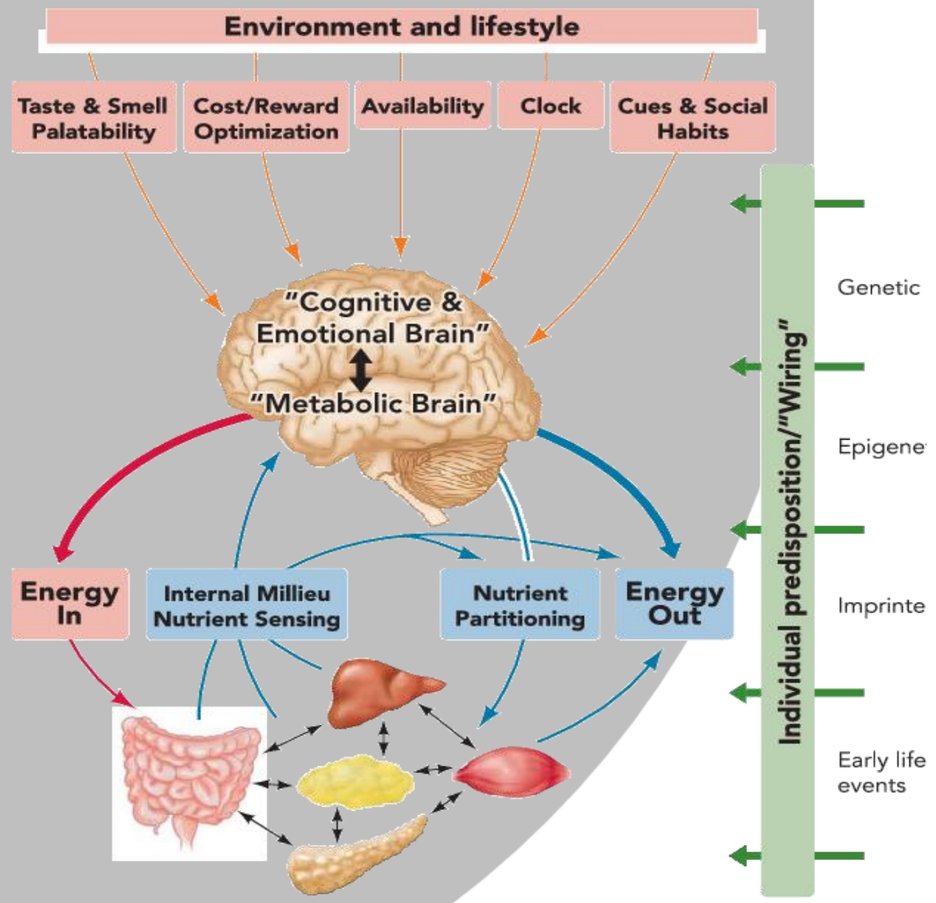
- Single-Gene: Mutations in one gene (e.g., sickle cell anemia)
- Chromosomal: Abnormal chromosome number/structure (e.g., Down syndrome)
- Multifactorial: Genes + environment (e.g., heart disease)
- Mitochondrial: Mutations in mitochondrial DNA (e.g., LHON)
- Autosomal Dominant: One mutated allele (e.g., Huntington's)
- Autosomal Recessive: Two mutated alleles (e.g., cystic fibrosis)



CAUSES OF GENETIC DISORDERS

- Mutations: Point mutations, insertions, deletions
- Chromosomal Errors: Aneuploidy, translocations
- Environmental: Teratogens, radiation exposure
- Germline Mutations: Inherited from parents
- Somatic Mutations: Acquired, not inherited
- Epigenetic Changes: Altered gene expression

HEALTH IMPACTS



- Developmental: Intellectual disability (e.g., Down syndrome)
- Systemic: Multi-organ dysfunction (e.g., cystic fibrosis)
- Metabolic: Enzyme deficiencies (e.g., PKU)
- Neurological: Seizures, motor deficits (e.g., Huntington's)
- Reproductive: Infertility (e.g., Turner syndrome)

DIAGNOSIS AND SCREENING

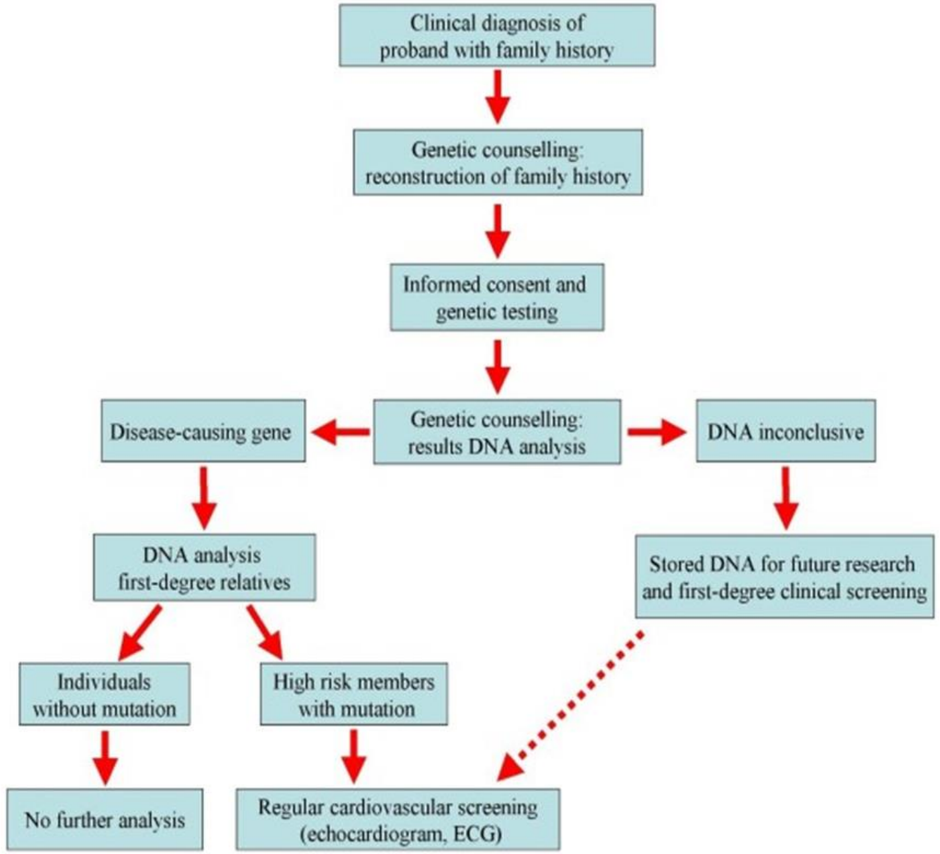
Genetic Testing: Detects mutations, chromosomal errors

Newborn Screening: Identifies disorders like PKU

Prenatal Testing: Amniocentesis, chorionic villus sampling

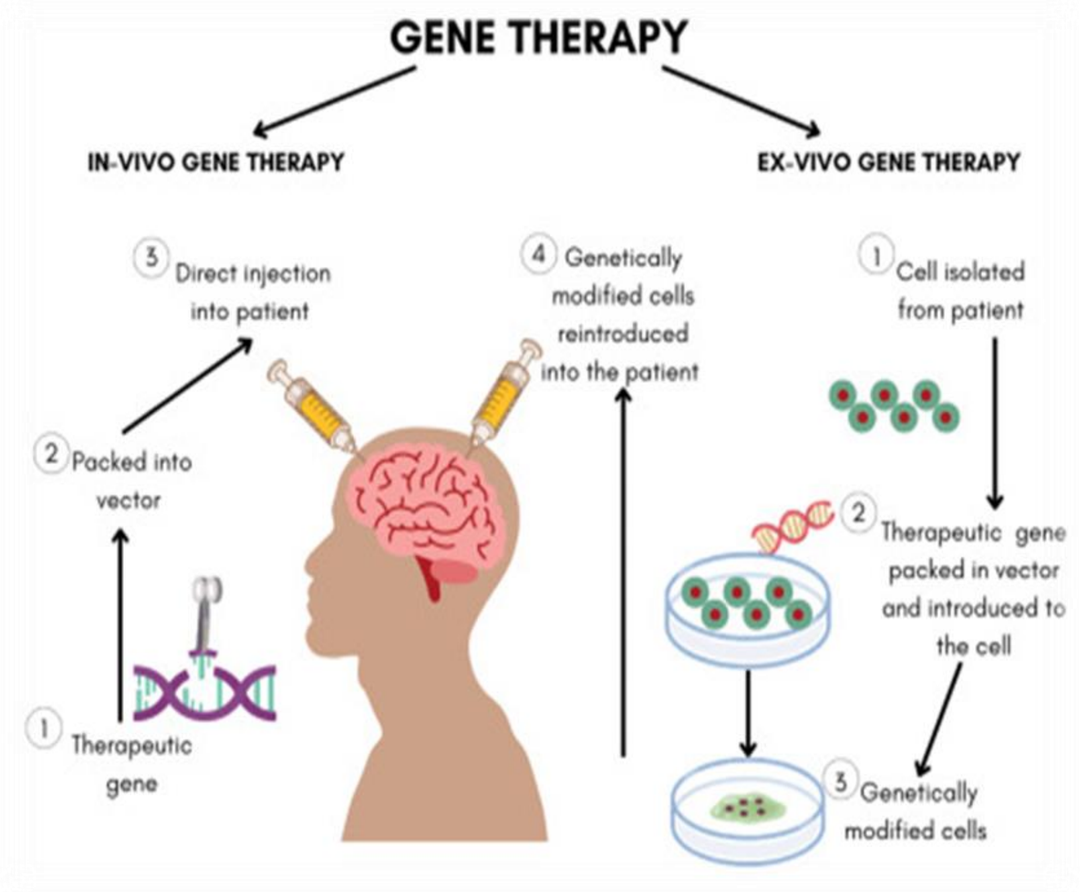
Family History: Key to identifying risk

DNA Sequencing: Identifies specific mutations



TREATMENT AND MANAGEMENT

- Symptomatic: Treat symptoms (e.g., insulin for diabetes)
- Gene Therapy: Emerging for single-gene disorders
- Surgery: Correct malformations (e.g., cleft palate)
- Psychosocial Support: Address emotional needs
- Follow-Up: Monitor for complications



NURSING ROLE IN GENETIC DISORDERS

- Assessment: Collect detailed family history
- Education: Explain disorders, treatment options
- Advocacy: Ensure informed consent for testing
- Emotional Support: Address patient/family concerns
- Documentation: Record genetic findings, care plans
- Ethical Practice: Maintain confidentiality
- Patient Empowerment: Encourage informed decisions

CONCLUSION

- Genetic disorders significantly impact health
- Early diagnosis and management improve outcomes
- Nurses are vital in assessment, education, and support
- Ethical care ensures patient trust
- Advances in genetics enhance treatment options

COMPETITIVE EXAM QUESTIONS (MCQS)

- **Question 1**: Cystic fibrosis is an example of a:
  - A) Chromosomal disorder
  - B) Single-gene disorder
  - C) Multifactorial disorder
  - D) Mitochondrial disorder
  - **Answer**: B) Single-gene disorder
- **Question 2**: Down syndrome is caused by:
  - A) Point mutation
  - B) Trisomy 21
  - C) Deletion
  - D) X-linked mutation
  - **Answer**: B) Trisomy 21
- **Question 3**: A key diagnostic tool for genetic disorders is:
  - A) ECG
  - B) Genetic testing
  - C) X-ray
  - D) Spirometry
  - **Answer**: B) Genetic testing

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Thank
You