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Based on **KUHS Syllabus**

Textbook of

Pediatric Nursing

for BSc Nursing Students

As per the syllabus of Kerala University of Health Sciences

Special Features

- **500+** Illustrations and Real-time Photographs
- **20+** Pediatric Procedures added
- Duly updated and evidence-based Information
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- Studded with Neonatal Problems Management Algorithms and Protocols
- Updated National Child Health Programs

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Panchali Pal

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Preface

The health problems in the developing countries are quiet challenging and require a comprehensive approach to child health problems by all health professionals including nurses. Nurses need to be trained in identifying and catering to the needs of children. Clinical expertise of a pediatric nurse is highly demanding in handling child health problems. Therefore, enriching clinical and theoretical skills by pediatric nurses is essential to contribute towards healthy childhood.

This textbook is designed for undergraduate students as per the syllabus of Kerala University of Health Sciences.

The prime objective of the book is to provide information to the nursing students in simplified and illustrated manner based on their curricular requirement.

This comprehensive book addresses domains of child development—emotional, social, physical, cognitive, language, and creative, which are readily observable and nursing considerations for developing clear concept of growth and development.

The text focuses on all major disease conditions with brief reference of rare diseases along with comprehensive nursing processes for the diseases. It explains the management of various disease condition following standardized protocols with recent advancements in the terminologies in the medical sciences. The manuscript was prepared by referring to various journals, periodicals, manuals of WHO, books and newsletters. I have tried my level best to cite all the references however, I sincerely apologize if some of them are missed out inadvertently.

I believe this book will be helpful for the students as well as staff nurses for enriching their knowledge.

Panchali Pal

Special Features of the Book

Learning Objectives

- Explain the Concept of Child Care
- Describe the Principles of Child Health Nursing
- Recognize the Difference between Child and Adult Nursing
- Describe National Policy and Child Welfare Programs

Learning Objectives enlist what the students will learn after studying the entire unit.

Chapter Outline

- History of Child Health Nursing
- Principles of Child Health Nursing
- Difference between Adult and Child Nursing
- Rights of Children
- Role of a Child Health Nurse
- Qualities of Pediatric

Chapter Outline will summarize what the students will learn after studying the chapter.

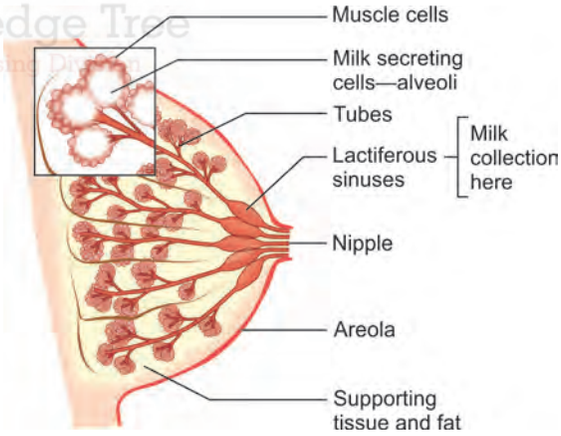

Fig. 1: Anatomy of breast

Self-explanatory images and line arts included to enhance understanding of topic.

Table 2: Types of enuresis

Type	Characteristics
Primary enuresis (80% of cases)	Enuresis in a child who has never established urinary continence for more than six months
Secondary enuresis (20% of cases)	Resumption of enuresis after at least six months of urinary continence

Important topic or concept have been represented in the form of **"Tables"** for better and easy understanding.

Various important and frequently asked facts have been highlighted in simple shortcuts under **Remember** boxes in between the relevant topics.

Remember

- Characteristics of nurse teacher includes 4 Cs
  C — Confidence C — Communication
  C — Competence C — Caring and empathy

Know it

Heat gain: Nonshivering thermogenesis:

- Brown fat metabolism causes heat production in the newborn.
- Blood becomes warm and transfer heat to other parts of the body.
- Brown fat is present near neck, scapula, axilla, kidney and adrenals.

Extra and important topic-related points are highlighted in separate boxes **"Know it"**.

At the end of each chapter **key messages** for nurses have been incorporated to inculcate the clinical importance of the relevant topic.

Key Message

- Pediatric nursing has multiple dimensions related to development of children and adolescents, and focuses on promoting health, growth and development, care and management of acute and chronic conditions and disabilities in hospital, family and the community.
- Child health protection involves maintaining child rights, formulation of child health policies and programs.

ASSESS YOURSELF

1. Define pediatric nursing.
2. List down the qualities of a pediatric nurse.
3. What are the expanded role of pediatric nurse?
4. Write short notes on:
   - Evidence-based practice
   - Functions/role of a pediatric nurse

Important questions under **Assess Yourself** have been added in the last of every chapter to help student assess their learning.

Syllabus

Child Health Nursing

Placement: III Year **Time:** 105 hours

Course description: Students identify the normal growth and development, needs/problems of children of various age groups and deviations from normal, recognize the basic concepts, principles and techniques of child care and the role of family in child rearing, develop beginning ability to plan and provide comprehensive nursing care to children suffering from diseases and disorders.

Unit	Time (Hrs)	Learning outcomes	Content	Teaching/learning activities	Assessment methods
I	**6**	• Explain the concept of child care • Describe the principles of child health nursing • Recognize the difference between child and adult nursing • Describe national policy and child welfare programmes	**Concepts of Child Care** • History of child health nursing • Principles of child health nursing • Difference between adult and child nursing • Rights of children • Qualities of a child health nurse • National policy and legislations in relation to child health and welfare • Child welfare programmes: State, National, International	• Lecture • Discussion	• Essay • Short answers • Very short answers
II	**2**	• Recognize universal immunization programme • Explain the activities of preventive clinics	**Preventive Pediatrics** • Child morbidity and mortality rates • Universal Immunization programme, cold chain • Under-five clinic, well baby clinic, child guidance clinic • IMNCI	• Lecture • Discussion • Field visit to child • Guidance • Clinic	• Short answers • Very short answers
III	**12**	• Describe the normal growth and development of children at different ages • Appreciate the role of play for normal and sick children	**The Healthy Child** • Principles of growth and development • Factors affecting growth and development • Growth and development, theories of development from infancy to adolescence, developmental problems and needs of children from infancy to adolescence; behavioral problems, habit disorders and management • Play needs of children	• Lecture • Discussion • Demonstration • Field visit to Anganwadi	• Essay • Short answers • Very short answers
IV	**5**	• Describe the nutritional needs of infants and children • Explain the nursing management of children with various nutritional disorders.	**Nutritional Needs and Care of Children with Nutritional Disorders** • Nutritional needs of infants and children: Breastfeeding, supplementary/artificial feeding and weaning • Baby friendly hospital initiative • Nutritional requirements in children • Nutritional deficiency • Disorders: Protein energy malnutrition • Vitamin and mineral deficiencies	• Lecture • Discussion • Demonstration • Practice session • Clinical practice	• Essay • Short answers • Very short answers

Contd...

Unit	Time (Hrs)	Learning outcomes	Content	Teaching/learning activities	Assessment methods
V	10	• Explain the nursing management of normal/high risk neonate • Perform neonatal resuscitation • Recognize and manage common neonatal problems	**Nursing Care of a Neonate** • Nursing care of a normal newborn • Essential newborn care • Kangaroo mother care • Nursing management of high risk neonate: low birth weight babies, preterm babies, • Management of common neonatal problems: Respiratory distress syndrome, hyperbilirubinemia, meconium aspiration syndrome, infant of diabetic mother, neonatal sepsis	• Lecture • Discussion • Workshop on neonatal resuscitation • Demonstration • Practice session • Clinical practice	• Essay • Short answers • Very short answers
VI	3	• Identify the reactions of child and family toward hospitalization and illness • Describe the major functions and role of pediatric nurse in caring a hospitalized child	**Caring for a Hospitalized Child** • Role of child health nurse in caring for a hospitalized child • Reaction of child and family toward illness and hospitalization and nurse's role in minimizing stress • General preoperative and postoperative management of children	• Lecture • Discussion	• Essay • Short answers
VII	5	Describe the nursing management of children with respiratory disorders	**Nursing Management of Children with Respiratory Disorders and Infections** Nursing management of children with • URTI : Tonsillitis, Croup, LRTI: Bronchial asthma, • Pneumonia, bronchiolitis, • Tuberculosis, emphysema, empyema	• Lecture cum discussion • Demonstration • Practice session • Clinical practice	• Essay • Short answers • Very short answers
VIII	11	Describe the nursing management of children with gastro-intestinal disorders	**Nursing Management of Children with Gastrointestinal disorders** • Gastroenteritis, malabsorption syndrome, hepatitis • Indian childhood cirrhosis • Cleft lip and palate • Tracheoesophageal fistula • Diaphragmatic hernia • Pyloric stenosis, intestinal • Obstruction, intussusception • Hirschsprung's disease • Anorectal malformation • Abdominal wall defects omphalocele, exomphalos, gastroschisis, hernias	• Lecture • Discussion • Clinical practice	• Essay • Short answers • Very short answers
IX	7	Describe the nursing management of children with cardiovascular disorders	**Nursing Management of Children with Cardiovascular Disorders** • Rheumatic fever • Rheumatic heart disease, heart failure, Kawasaki disease • Congenital heart defects • Patent ductus arteriosus, atrial septal defect, Ventricular septal defect, tetralogy of Fallot, TGA	• Lecture • Discussion • Clinical practice	• Essay • Short answers • Very short answers
X	6	Explain the nursing management of children with common genitourinary disorders	**Nursing Management of Children with Genitourinary Disorders** • UTI, acute glomerulonephritis, Nephrotic syndrome, • Wilms' tumor, obstructive uropathy, epispadias, hypospadias, ectopia vesica	• Lecture • Discussion • Clinical practice	• Essay • Short answers • Very short answers

Contd...

Unit	Time (Hrs)	Learning outcomes	Content	Teaching/learning activities	Assessment methods
XI	6	Describe the nursing management of children with neurological disorders	**Nursing Management of Children with Neurological Infections and Disorders** • Seizure disorder, meningitis, encephalitis, neural tube defects, encephalocele, hydrocephalus • Head Injury, brain tumors, cerebral palsy	• Lecture • Discussion • Clinical Practice	• Essay • Short answers • Very short answers
XII	6	Explain the nursing management of children with common hematological disorders	**Nursing Management of Children with Hematological Disorders** Anemias, thalassemia, ITP, Leukemia, Hodgkins' and nonHodgkin's lymphoma, hemophilia	• Lecture • Discussion • Clinical practice	• Essay • Short answers • Very short answers
XIII	3	Describe the nursing management of children with common endocrine disorders	**Nursing Management of Children with Endocrine Disorders** • Juvenile diabetes mellitus, congenital hypothyroidism • Growth hormone deficiency	• Lecture • Discussion • Clinical practice	• Short answers • Very short answers
XIV	2	Describe the nursing management of children with orthopedic conditions	**Nursing Management of Children with Orthopedic Disorders** Club feet, congenital hip dislocation	• Lecture cum • Discussion • Clinical practice	• Short answers • Very short answers
XV	5	Describe the nursing management of children with common genetic disorders	**Nursing Management of Children with Genetic Disorders** • Down's syndrome • Turner's syndrome • Kleinfelter's syndrome • Edwards syndrome • Inborn errors of metabolism: Phenylketonuria, albinism, galactosemia, genetic counseling	• Lecture • Discussion • Clinical practice	• Short answers • Very short answers
XVI	3	Describe the nursing management of children with disorders of skin, eye, and ears	**Nursing Management of Children with Common Disorders of Skin, Eye and Ear** • **Skin:** Scabies, pyoderma, staphylococcal scalded skin syndrome • **Eye:** Congenital glaucoma, cataract, squint, refractive errors • **Ear:** Wax, otitis externa, otitis media, mastoiditis, childhood deafness	• Lecture • Discussion • Clinical practice	• Short answers • Very short answers
XVII	5	Describe the nursing management of children with communicable diseases	**Nursing Management of Children with Communicable Diseases** • Communicable diseases in children and its prevention and management • Dengue fever, chikungunya • Nursing care of infant and children with HIV/AIDS	• Lecture • Discussion • Clinical practice	• Short answers • Very short answers
XVIII	5	Explain the nursing management of children with emergency conditions	**Child Health Emergencies** Accidents, poisoning, foreign bodies, shock, hemorrhage, burns, drowning, snake bite—nursing management	• Lecture • Discussion • Clinical practice	• Short answers • Very short answers
XIX	3	Identify the social and welfare services for challenged children	**Management of Challenged Children** • Mentally, physically, and socially challenged • Welfare services for challenged children in India	• Lecture • Discussion • Field visits to school for mentally, physically and socially challenged	• Short answers • Very short answers

Contents

UNIT VI

Genetics and Embryology

Learning Objectives

- Concepts and importance in pediatrics
- Gene structure and function
- Pattern of inheritance
- Common chromosomal disorders—Down's syndrome, Turner's syndrome, Klinefelter's syndrome, Edwards syndrome, albinism, inborn errors of metabolism, phenylketonuria, galactosemia
- Growth and development of fetus
- Maldevelopment leading to congenital anomalies
- Genetic testing in neonates and children

Unit Outline

Chapter

24

Genetics and Embryology

Chapter Outline

- Concept of Genetics
- Importance of Genetics in Pediatrics
- Gene Structure
- Functions of Gene
- Pattern of Inheritance
- Growth and Development of Fetus
- Maldevelopment Leading to Congenital Anomalies
- Genetic Testing in Neonates and Children

CONCEPT OF GENETICS

Genetics is a study of genes, gene variation and inheritance of living organism which deals with transmission of characteristics from parents to offspring. The term genetics was introduced by Bateson in 1906. The term is derived from Greek word gene, which means 'to become or to grow into'.

- **Basic terminologies:**
  - **Gene:** Gene is a discrete unit of hereditary information that usually specifies a protein; a region of DNA (locus) located on a chromosome that identifies a trait.
  - **Alleles:** They are variations of genes governing variations of the same characteristic or trait that occupy corresponding positions on homologous chromosomes.
- **Chromosome**: Structures within the nucleus of eukaryotic cells composed of chromatin and visible at cell division (condensed chromatin).
- **Homologous chromosomes:** Chromosomes that are similar in morphology (shape and form) and genetic constitution. In animals one set comes from the father and the other from the mother.
- **Gene locus**: A particular portion or region of the chromosome representing a single gene is known as gene locus.
- **Dominant allele:** It is one of a pair of allele which can express itself irrespective of homozygous or heterozygous state.
- **Recessive allele:** It is one of the pair of allele which is unable to express its effect in presence of its contrasting alleles.
- **Genotype:** It is the gene complement of an individual with regard to one character irrespective of whether the genes are expressed or not.
- **Phenotype:** It is observable or measurable distinctive structure or functional characteristics of an individual with regard to one or more characters which is a result of gene products brought to expression in a given environment.
- **Karyotype:** It is a picture showing the organization of a complete set of human chromosome.
- **Haploid:** It is the condition of having only one set of chromosomes per cell (n)
- **Diploid:** It is the condition of having two sets of chromosomes per cell (2n)
- **Carrier:** A heterozygous individual not expressing a recessive trait but capable of passing it on to its offspring
- **Recombination:** Exchange of genetic material between chromosomes
- **Crossover:** The breaking and rejoining of homologous (non-sister) chromatids during cell division i.e., early prophase I of meiosis, resulting in recombination
- **Synapsis:** The pairing of homologous chromosomes during cell division, i.e., prophase I of meiosis.
- **Disjunction**: Separation of homologous chromosomes (or sister chromatids) during anaphase.

IMPORTANCE OF GENETICS IN PEDIATRICS

- Genetics is predominantly associated with decision regarding child bearing and treating children with genetic disorders.
- It focuses on the inheritance of hereditary disorders affecting children.
- Genetic services like prenatal counselling, identification of birth defects and dysmorphology..
- It helps to understand the genetic contribution to disease risk and disease management.
- Genetic counselling is based on genetic predisposition.
- Genetic disorders account for 50% of all childhood vision and hearing impairment and mental retardation.

About 2–5% of all live births constitute for genetic disorders or congenital anomalies.

To understand genetic basis of diseases, early and effective diagnosis of the disorders and management and prevention of genetic conditions, study of genetics is very important.

GENE STRUCTURE

Gene is a structural and functional unit of heredity. There are about 30000 genes in each cell of the human body. They are mainly composed of Deoxyribonucleic Acid (DNA). A DNA molecule is composed of two chains of nucleotides that wind about each other to resemble a twisted ladder forming a double helix structure. The sides of the ladder are made up of sugars and phosphates, and the rungs are formed by bonded pairs of nitrogenous bases such as adenine (A), guanine (G), cytosine (C), and thymine (T). An A on one chain bonds to a T on the other (thus forming an A–T ladder rung); similarly, a C on one chain bonds to a G on the other. Each of these pairs of bases is called a base pair. The combination of one of these nitrogenous bases, a sugar molecule, and a phosphate molecule is called a nucleotide, the basic building block of the DNA molecule. The two strands of DNA are complementary. If the bonds between the bases are broken, the two chains unwind, and free nucleotides within the cell attach themselves to the exposed bases of the now-separated chains. The free nucleotides line up along each chain according to the base-pairing rule—A bonds to T, C bonds to G. This process results in the creation of two identical DNA molecules from one original. In this process hereditary information is passed from one generation of cells to the next (Fig. 1).

Human genes, like most genes from multi-cellular organism (eucaryotes), contain introns, i.e., stretches of DNA located within the gene, transcribed into RNA and then spliced out before the RNA is translated into protein. Regions containing the coding information which are both transcribed and translated into proteins are called exons. On either side of a gene there are regions called flanking regions that play roles in the regulation of gene expression. On the far end of

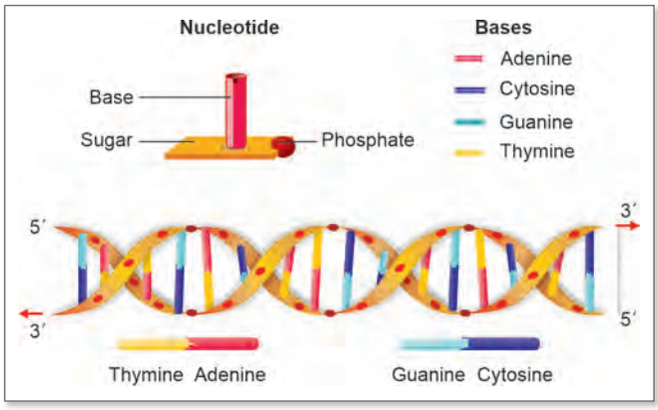

Fig. 1: Structure of DNA

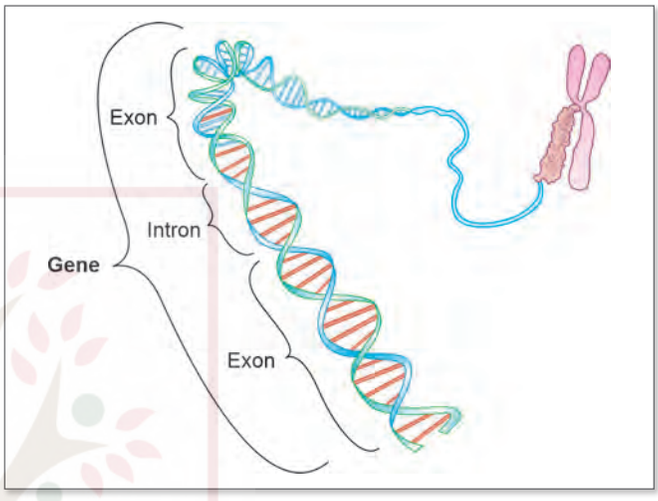

Fig. 2: Structure of Gene

the gene, past the coding region of introns and exons, is the three flanking region which largely remains untranslated (Fig. 2).

FUNCTIONS OF GENE

- Genes regulate the functions of DNA and RNA.
- Proteins are the most important materials in the human body which not only help by being the building blocks for muscles, connecting tissue and skin but also takes care of the production of the enzyme.
- These enzymes play an important role in conducting various chemical processes and reactions within the body. Therefore, protein synthesis is responsible for all activities carried on by the body and are mainly controlled by the genes.
- Genes consist of a particular set of instructions or specific functions. For example, the globin gene was instructed to produce hemoglobin. Hemoglobin is a protein that helps to carry oxygen in the blood.
- Replication of gene causes cell division.

- They control structure and metabolism of the body.
- They determine the morphology of the phenotype of individual.

PATTERN OF INHERITANCE

Refer chapter 19: **Nursing Management of Children with genetic disorders**

GROWTH AND DEVELOPMENT OF FETUS

Prenatal development is divided into germinal period, embryonic period and fetal period. 1st and 2nd week is germinal period when fertilization occurs and zygote forms which undergoes cell division. Embryonic period lasts from 3rd week to 8th week of conception. In this period zygote gets implanted in uterus and called embryo.

Developments during Embryonic Period

- **3–4 weeks:** The embryo changes from a flat disc to a curved, C-shaped form. Organs begin forming. Tube forms from embryo's length which will develop brain and spinal cord.Heart begins to beat. Limb buds appear.
- **5–6 weeks**: About half of the embryo's length is the head, due to the rapid growth of the brain. The heart starts to form the normal four chambers. A heartbeat can be seen on ultrasound. Eye and ear starts to form. Leg buds appears. Hand appears as paddles. Lungs and kidneys begin to form.
- **7–8 weeks:** Fingers and toes are formed. Facial structures are formed. Early bones are formed Hair follicles start to develop. Face looks as human.

Development during Fetal Period

At nine weeks of age, the embryo is called a fetus. At this stage, the fetus is about the size of a kidney bean and begins to take on the recognizable form of a human being.

- **9–10 weeks:** The ears move up from around the neck to their normal position. Fetal movements and heartbeat can be seen on ultrasound. Various glands begin to function. The kidneys start producing urine. The crown-rump length is 61 mm. The fetus weighs 14 grams.
- **11–12 weeks:** The sex of the fetus can be visible at this age. The fetus starts swallowing fluid from the amniotic sac. The fluid is replaced with urine. The placenta is well developed. Blood cells are produced in the bone marrow. The neck can be clearly visible between the head and body. The crown-rump length is 86 mm and the weight is 45 grams.
- **13–14 weeks:** The fetal head is still large as the body straightens out. The arms and legs are formed, and can move and bend. Sex organs are almost completely developed. Toenail and fingernail starts growing. The ears reach normal position and the face is also well formed. Tooth buds may start appearing. The crown-rump length becomes 120 mm.The fetus weighs 110 grams.
- **15–16 weeks:** Growth begins to speed up. The legs grow longer, making fetal head less large. Slow fetal eye movements can be identified by ultrasound. The ears stand out from the head. The crown-rump length reaches 140 mm and the fetus weighs 200 grams.
- **17–18 weeks:** The fetal skin is covered by "vernix caseosa". Fine downy hair called "lanugo" covers the fetal body. The crown-rump length goes to 160 mm. The fetus weighs 320 grams.
- **19–20 weeks:** The skin becomes red and wrinkled. Blood vessels can be visible clearly beneath it. Eyebrow and eyelashes start to develop. The crown-rump length is 190 mm. The fetus weighs 460 grams.
- **21–22 weeks:** Lung grows and some gas exchange sacs are formed. The heartbeat can be heard with a stethoscope. The crown-rump length is 210 mm. The fetus weighs 630 grams. At this time, there is a chance the fetus may live if delivered.
- **23–24 weeks:** The lungs continue to grow and surfactant is developed. Fat gradually builds up under the skin. The fetus can suck on fingers or hands. The fetus will blink and act startled in response to loud noises. The crown-rump length reaches 230 mm. The fetus weighs 820 grams.
- **25–26 weeks:** The fetus continues to gain weight. The brain grows and starts to do more complex tasks. Fetal eyes will open slightly. Eyelashes are formed. The crown-rump length reaches 250 mm. The fetus weighs 1000 grams.
- **27–28 weeks:** The fetal brain can now regulate body temperature and direct regular breathing. The fetus can weakly grasp at things. The eyes open wide. Toenails begin to form. Blood cells starts producing in the bone marrow. The crown-rump length is around 270 mm. The fetus weighs 1300 grams.
- **29–30 weeks:** The fetus starts to look like a newborn baby. The lanugo hairs on the face disappears. The pupils of the eyes react to light. The fetus may now hiccup. The crown-rump length is around 280 mm. The fetus weighs around 1700 grams.
- **31–32 weeks:** Fat is still building up under the skin as the fetus grows. The lungs keep growing and produce more surfactant. The ear holds it shape when moved. Fetal muscle tone increases. The crown-rump length is around 300 mm. The fetus weighs around 2100 grams.
- **33–34 weeks:** The lungs and the nervous system keep growing. Hair on the head begins looking normal. Testes in male fetuses start to move from the abdomen into the scrotum. The labia in female fetuses begin to cover the clitoris. The fetus moves into a head-down position to prepare for delivery. The average crown-rump length is over a foot. The fetus weighs around 2500 grams.
- **35–36 weeks:** In almost all cases, fetal lungs are matured. The fetus drops lower into the mother's pelvis. Lanugo hairs

Fig. 3: Stages of fetal development

are almost disappeared except for around the shoulders and upper arms. The fetus may be born now or may stay in the womb while more fat builds up under the skin.

- **37–38 weeks:** This is full term in pregnancy. Most babies are delivered during this time. The average crown-rump length is 360 mm. On average, a full-term baby weighs 3400 grams (Fig. 3).

MALDEVELOPMENT LEADING TO CONGENITAL ANOMALIES

Congenital anomalies are those that appear due to an interruption or deviation from the normal development of the fetus during the embryological and fetal period of development. Congenital abnormalities appearing during embryonic period is known as embryopathies and deformities arising during fetal period are fetopathies.

Terminologies Related to Congenital Anomalies

- **Primary Malformations:** Formation during organogenesis (3rd–8th week of development) of the embryonic period (2nd–8th week of development), whereby the organ can completely or partially fail, or exhibit structural defects due to genetic or external factors. During this time, the embryo is highly vulnerable to teratogens.
- **Secondary Malformations:** Formation via destruction or alteration of already developing organs, such as intestinal atresia due to a vascular change, or defects in the amniotic bands.

- **Agenesis:** The absence of an organ due to failure to develop
- **Aplasia:** Though the organ has developed but has defective features.
- **Atresia:** Absence of the physiological opening or the lumen of a (hollow) organ, or occlusion thereof.
- **Stenosis:** Constriction within an organ.
- **Syndrome:** Include multiple malformations with the same cause and in a characteristic combination, e.g., resulting from trisomy 21 – Down's syndrome.

Factors Responsible for Malformations

- Maternal infections like TORCH leading to vision disorders, heart defects and mental impairments.
- Exposure to radiations may lead to micro or anencephaly, cleft palate, etc.
- Chemical factors like smoking and alcohol may lead to fetal alcohol syndrome.
- Medicines like Thalidomide (amelia/meromelia, and heart disorders), phenytoin (organ malformation and mental impairment) and tetracycline (dental diseases or deafness).
- Hormones may lead to reproductive organ abnormalities.
- Maternal illness like diabetes, seizures, thyroid disorders may lead to abnormalities.
- Family history of malformations increases the risk of developing anomalies.

Maldevelopments in Various Systems

- **CNS (3rd–32nd week of development):** The CNS develops from the neural tube, which protrudes from the neurectoderm. Impairment may lead to neural tube defects, such as spina bifida and mental disability.
- **Heart (3rd–7th–9th week of development):** The heart develops from the lateral plate mesoderm. The most common heart defects are persistent truncus arteriosus, atrial septal defect (ASD), and ventricular septum defect (VSD).
- **Extremities (4th–6th–9th week of development):** The extremities are mesenchymal in origin, and also arises from the lateral plate mesoderm. The symptoms of meromelia/amelia, or the lack of parts or all of one or more extremities, are due to impairments during this period.
- **Ears (4th–10th–32nd week of development):** If the ear, developing from the ectodermal inner ear and Eustachian tube, as well as the eardrum from the endoderm, is damaged during this phase, may lead to deafness, auricular dysplasia, or a deep base of the ear.
- **Respiratory tract (4th–16th–40th week of development):** The respiratory tract combines with the mucosa of the mouth and nose from the surface ectoderm, and the respiratory epithelium from the endoderm. Impairments may cause fistulas, stenoses, and atresia, or fetal acute respiratory distress syndrome (ARDS) (Fig. 4).
- **Urogenital system (4th–16th–40th week of development):** The shared mesodermal precursor is the pronephric duct, from which the kidneys and the deviating urinary passages, and the sex organs (via the Wolffian and Müllerian ducts), develop. One common malformation is the urachal fistula after persistent connection of the urinary bladder with the navel, upon which urine is discharged by the navel of the infant upon activation of the abdominal press.
- **Gastrointestinal tract (GIT; 5th–32nd–40th week of development):** The mucosa of the gastrointestinal tract is allocated to the endoderm, and the stomatodaeum and anal region to the ectoderm; the glands flowing into them, including the liver, gallbladder, and pancreas, originate in the endoderm (Table 1). Along with stenoses and atresia of all systems involved, impairments may also result in rotational disabilities or lead to umbilical hernias as the result of temporary, extra-fetal localization of the intestine, such as in the form of an omphalocele.
- **Face, lips, palate (5th/6th–7th/8th–16th week of development):** the face develops from five mesenchymal facial processes, which form from neural crest cells. If the epithelia are unable to fully make contact, or if there is a lack of proliferation or a break-up of already forming coalescence zones, such as the medial nasal prominence and the maxillary process, the relatively common cleft lip and palate may develop.

Prevention

- Ensuring adolescent girls and mothers have a healthy diet including a wide variety of vegetables and fruit, and maintain a healthy weight.
- Ensuring an adequate dietary intake of vitamins and minerals, and particularly folic acid in adolescent girls and mothers.
- Reducing or eliminating environmental exposure to hazardous substances (such as heavy metals or pesticides) during pregnancy.
- Increasing and strengthening education of health staff and others involved in promoting prevention of congenital anomalies.
- Screening for infections, especially rubella, varicella, and syphilis, and consideration of treatment.
- Vaccination of the mother against rubella.
- Folic acid substitution during pregnancy to lower the incidence of neural tube defects.
- Iodation of drinking water and table salt prevents developmental impairments of the thyroid (cretinism).
- Abstinence from alcohol and cigarettes.
- Good assortment of medication for metabolic diseases of the mother.
- Consider teratogenicity when administering medication for pregnant women.

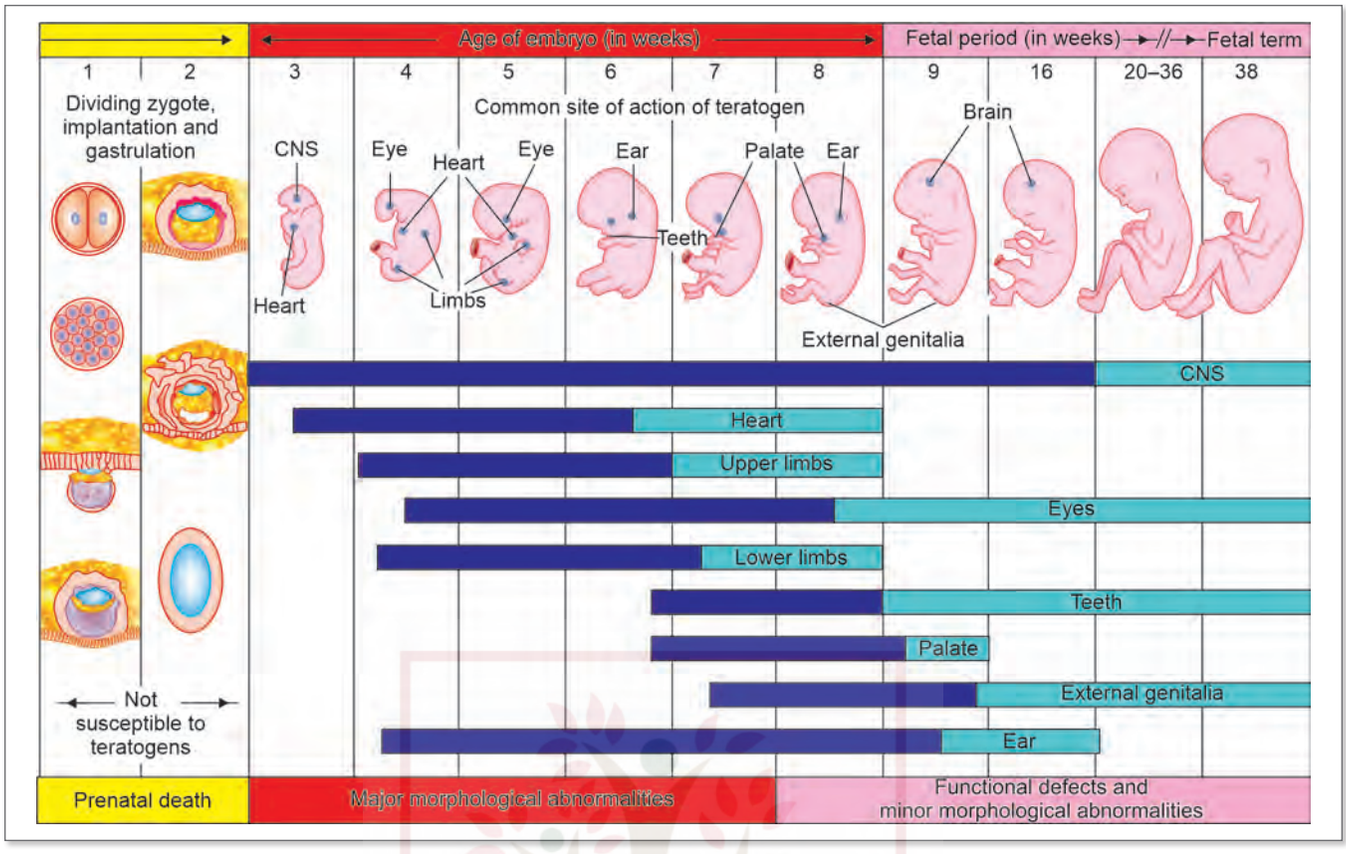

Fig. 4: Maldevelopments during fatal period

Table 1: Common congenital anomalies

System	Congenital abnormality
Central nervous system	• Anencephaly • Craniorachischisis • Iniencephaly • Encephalocele • Spina bifida • Microcephaly
Eye/ear/face/neck	• Microtia/Anotia • Cleft lip and cleft Palate
Cardiovascular	• Common truncus (truncus arteriosus) • Hypoplastic left heart syndrome • Interrupted aortic arch • Pulmonary valve atresia • Tetralogy of Fallot • Transposition of the great arteries • Tricuspid valve atresia and stenosis
GI	• Esophageal agenesis/hypoplasia • Large intestinal atresia/stenosis • Rectal atresia/stenosis
Musculoskeletal	• Talipes equinovarus • Reduction defects of upper and lower limbs (longitudinal, transverse, and intercalary) • Exomphalos (omphalocele) • Gastroschisis
GU	• Hypospedias • Epispedias • Renal agenesis

GENETIC TESTING IN NEONATES AND CHILDREN

Genetic testing is defined as examining a sample of blood or other body fluids or tissue for bio-chemical, chromosomal, or genetic markers that indicate the presence or absence of genetic disease.

Types of Genetic Testing

Diagnostic Testing

DNA based tests are used to identify or rule out a specific genetic or chromosomal condition when clinical signs and symptoms suggest the diagnosis.

Uses:

- Duchenne muscular dystrophy, spinal muscular atrophy, Friedriech ataxia, Fragile X syndrome, Pompe disease, etc.
- In management of cystic fibrosis, for use of specific and safe drugs pharmacogenetic testing is done.

Carrier Testing

It is used to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder.

This type of testing is usually requested when a relative is affected with an autosomal recessive or an X-linked disease or when parents are carriers. Testing may allow the carriers to be well prepared to choose their partners and take appropriate reproductive decisions.

Presymptomatic Testing

For early onset disorders: It is done when early institution of preventive and intervention measures can affect the prognosis. **Ex.:** Juvenile hemochromatosis where HFE2 and HAMP genes are mutated. All the family members of an affected individual should be tested for the mutations identified and treatments can be initiated if the disease can be detected early.

Presymptomatic diagnosis of Wilson disease and retinoblastoma in childhood can aid in medical management of at risk child.

For late onset disorders like adult-onset conditions pre-symptomatic testing helps to plan for the future, availing life insurance, and making reproductive decisions. Ex.: Huntington disease, neuronal problems, Breast cancer, Diabetes, etc.

Cancer Susceptibility Testing

In some familial cancers it may be done to ensure effective prevention and early diagnosis.

Karyotyping

Karyotyping is the process by which photographs of chromosomes are taken in order to determine the chromosome complement of an individual, including the number of chromosomes and any abnormalities. The chromosomes are depicted (by rearranging a photomicrograph) in a standard format known as a karyogram or idiogram: in pairs, ordered by size and position of centromere for chromosomes of the same size (Fig. 5).

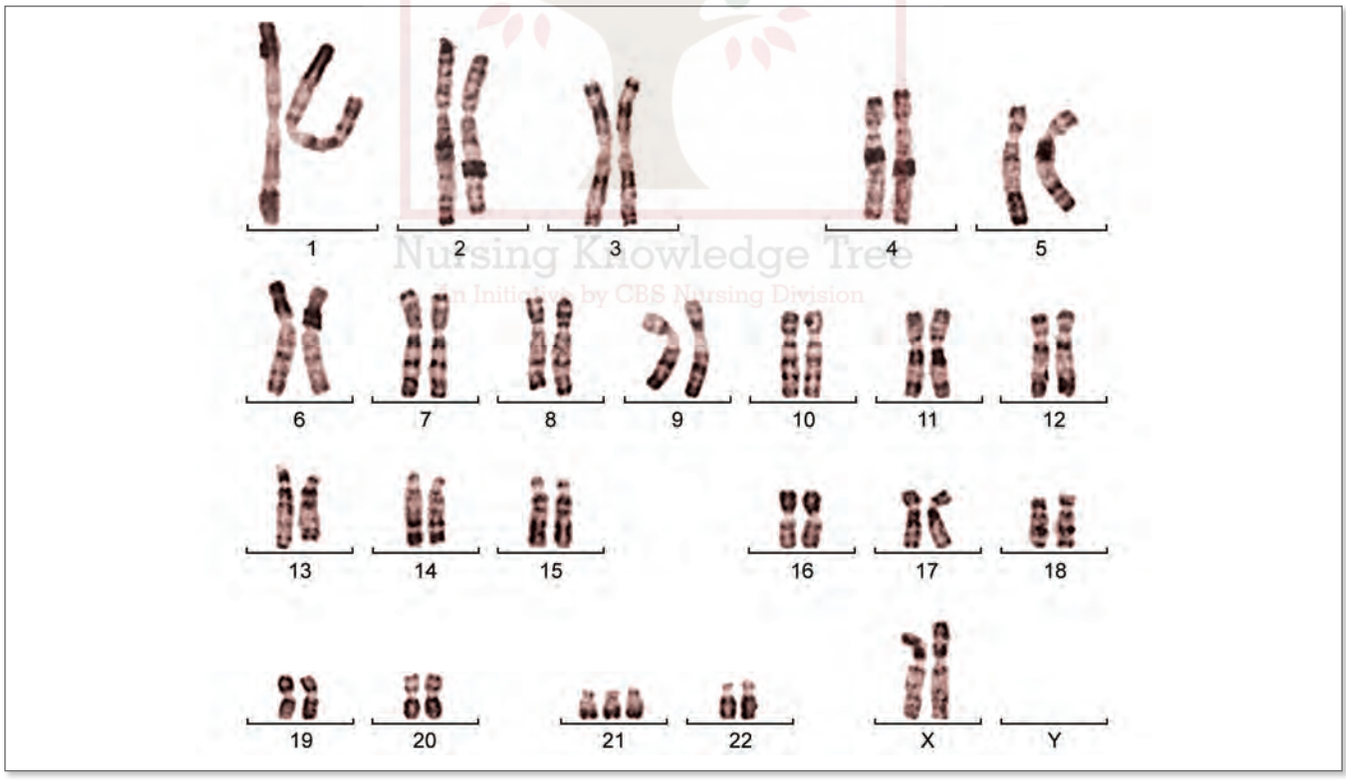

Fig. 5: Karyotyping of chromosomes

Uses:

- It is used to diagnose a genetic disorder in a newborn or child with symptoms such as Trisomy at 21st chromosome (Down syndrome)—Trisomy at sex chromosome XXY (Klinefeltners syndrome)—Monosomy at sex chromosome- XO (Turner syndrome).
- It helps to undergo preconception testing before deciding to become pregnant. Parents have an option for prenatal diagnosis during the next pregnancy.
- Karyotype also helps in detection of some cancers.

Fluorescence *in situ* Hybridization (FISH)

This is a molecular cytogenetic technique that uses fluorescent probes that binds to specific regions of a chromosome to understand where and when a specific DNA sequences exist in cells by detecting the fluorescent group.

Chromosomal Microarray Analysis (CMA) or Microarray

This is a whole genome test to identify a wide variety of genetic syndromes caused by microdeletions and duplications in the genome. It is highly useful technique for detection of submicroscopic deletions and duplications across the genome. It is indicated to diagnose ailments related with development delays, autism or any kind of genetic abnormalities.

Benefits of Genetic Testing

- **Medical benefits:** DNA testing plays a major role in arriving at definitive diagnosis in diseases such as spinal muscular atrophy, Friedreich ataxia, Fragile X syndrome, etc.
- **Psychological issues:** The report can reduce the uncertainty and offer emotional relief if negative. Positive result on the other hand helps in preparing for the future as in planning education, managing finances and allows time to adjust and avoid emotional problems.
- **Reproductive issues:** It will only be useful in reproductive or family planning decisions.

Potential Harms in Genetic Testing

- Genetic tests may prompt further investigations and unnecessary treatments with no proven benefits thus causing distress and escalating the cost.
- A positive test may cause unwarranted anxiety about the possible early signs of the disorder before any genuine manifestations actually set in.
- Revealing the results can impair the self-esteem and lead to discrimination in education, employment, insurance etc.
- Testing at an early age deprives the child's right to decide about pre-symptomatic diagnosis as an adult.

Preconception Screening

It is required to identify those at risk of conceiving a child with a birth defect since inherited disorders tend to cluster within families, using family history to identify individuals at risk of having affected children. Carrier screening is done for common recessive disorders—β thalassemia and sickle cell anemia

Periconception screening is done during 1st and 2nd trimester of pregnancy. Common tests are Nucchal transluscency test, beta hCG level test, triple screen test (Alpha-fetoprotein test, human chorionic gonadotrophin, and estriol estimation), amniocentacis and chorionic villus sampling etc.

Common Chromosomal Disorders

***Refer* to Chapter 19**

Key Message

Embryonic development is very crucial for having a healthy child. Study of genetics and genetic disorders are the key to prevent genetic abnormalities. Timely screening and counseling can reduce the burden of genetic abnormalities.

ASSESS YOURSELF

1. What is the importance of genetics?
2. Discuss the structure and function of gene.
3. Write short notes on development in fetal period, types of congenital anomalies, karyotyping.

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CBS Publishers & Distributors Pvt. Ltd.
4819/XI, Prahlad Street, 24 Ansari Road, Daryaganj, New Delhi 110 002, India
E-mail: feedback@cbspd.com, **Website:** www.cbspd.com
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ISBN: 978-93-90619-80-1