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## References taken and adapted from:

1. “Kleigman, Behrman, Jenson, Stanton, Nelson Textbook of Paediatrics”, 1st South Asia Edition, Saunders, Elsevier

2. “Eric C. Eichenwald, Anne R. Hansen, Camila R. Martin, Ann R. Stark, Cloherty and Stark’s Manual of Neonatal Care”, 8th Edition, Wolters Kluwer

3. “Hiralal Konar, DC Dutta’s Textbook of Obstetrics”, 8th Edition, Jaypee

4. “K. Park, Park’s Textbook of Preventive and Social Medicine”, 23rd Edition, Bhanot

5. “Myung K. Park, Pediatric Cardiology for Practitioners”, 5th Edition, Mosby Elsevier

6. “Michael Glynn, William Drake, Hutchison’ Clinical Methods”, 23rd Edition, Saunders Elsevier

7. “Krishna M. Goel, Devendra K. Gupta, Hutchison’s Paediatrics”, 2nd Edition, Jaypee

8. “Taussig, Landau, Le Souef, Martinez, Morgan, Sly, Pediatric Respiratory Medicine”, 2nd Edition, Mosby Elsevier

9. “Cassidy, Petty, Laxer, Linsley, Textbook of Pediatric Rheumatology”, 6th Edition, Saunders Elsevier

10. “Denis F. Geary, Franz Schaefer, Comprehensive Pediatric Nephrology”, 1st Edition, Mosby Elsevier

11. “Kenneth F. Swaiman, Stephen Ashwal, Donna M. Ferriero, Pediatric Neurology Principles and Practice”, 4th Edition, Elsevier

12. “Sumner J. Yaffe, Jacob V. Aranda, Neonatal and Pediatric Pharmacology Therapeutic Principles in Practice”, 4th Edition, Wolters Kluver, Lippincott Williams & Wilkins

13. “Richard J. Martin, Avroy A. Fanaroff, Michele C. Walsh, Fanaroff and Martin’s Neonatal and Perinatal Medicine Diseases of the Fetus and Infant”, 4th Edition, Saunders Elsevier

14. “Somen Das, A Manual on Clinical Surgery, 9th Edition, Dr. S. Das

15. “John F. Moran, Clinical Methods: The History, Physical, and Laboratory Examinations”, 3rd edition

16. Physiology Journal, M. U. H. S. Nashik

# 1. The Field of Pediatrics

- **Quality of healthcare:** the degree to which healthcare services for individuals and populations increases the likelihood of desired health outcomes and are consistent with current professional knowledge

  A. **Safety:** avoiding preventable injuries, reducing medical errors

  B. **Effectiveness:** providing services based on scientific knowledge (clinical guidelines)

  C. **Patient-centeredness:** care that is respectful and responsive to individuals

  D. **Efficiency:** avoiding the wasting of time and other resources

  E. **Timeliness:** reducing wait times, improving the practice flow

  F. **Equity:** consistent care regardless of patient characteristics and demographics

- **Quality initiative:** systematic, data-guided activities designed to bring about immediate improvements in healthcare delivery in particular settings

- **Performance measure:** yardsticks by which all healthcare providers and organizations can determine how successful they are in delivering recommended care and improving patient outcomes

- **Performance management:** a systematic process by which an organization involves its employees in improving the effectiveness of the organization and achieving the organization's mission and strategic goals

- **Process improvement:** the systematic approach to closing of process or system performance gaps through streamlining and cycle time reduction, and identification and elimination of causes of below specifications quality, process variation, and non–value-adding activities

- **Reliability in healthcare:** the measurable capability of a process, procedure, or health service to perform its intended function in the required time under commonly occurring conditions (i.e., providing intended care on a consistent basis)

- **Screening:** the identification of a previously unrecognized disease or disease precursor, using procedures or tests that can be conducted rapidly and economically on large

numbers of people with the aim of sorting them into those who may have the condition(s)…and those who are free from evidence of the condition(s)

- **Clinical research:** studies of living human subjects, including the laboratory-based development of new forms of technology; studies of tge mechanism of human disease and evaluations of therapeutic interventions; clinical trials, outcome studies, and healthcare research; and epidemiological and behavioural studies

- **Hypothesis:** formal statements that declare what the investigator will test and then either reject or fail to reject, using appropriate statistical techniques

- **Objectives:** statements as to what the investigators plan to learn or accomplish by conducting the study

- **Prevalence:** the total number of existing cases in a defined population who developed a disease either at a point in time or during some time period

- **Incidence:** the rate at which newly diagnosed cases develop over time in a population

- **Prognosis:** the possible outcomes of a disease and the frequency with which they can be expected to occur

- **Simulation:** any exercise that allows an individual to experience a situation that, although not real, nevertheless generates authentic responses on his or her part

- **Principlism:** an approach to ethical problems that is based on the application of the four principles of beneficience, nonmaleficience, justice, and autonomy

- **Virtue:** a trait that has moral or social value

- **Spirituality:** a belief system that focuses on intangible elements that impart vitality and meaning to one's life

## 2. Growth, Development, and Behavior

- **Psychological attachment:** a biologically determined tendency of a young child to seek proximity to the parent during times of stress and their parents to reestablish a sense of well-being after a stressful experience

- **Habituation:** a basic form of learning in which repeated stimulation results in a response decrement

- **Surveillance:** ongoing monitoring (tracking over time) of such issues as parental concerns, children's progress with milestones, psychosocial risk and resilience factors, providers' efforts to both detect and address problems, and follow-up regarding child/family outcomes

- **Percentile:** the percentage of individuals in the group who have achieved a certain measured quantity or a developmental milestone

- **Normal:** 95% of a population that falls within 2 SD of the mean from any given measurement

## 3. Behavioral and Psychiatric Disorders

- **Grief:** a personal, emotional state of bereavement or an anticipated response to loss, such as a death

- **Insomnia:** difficulty initiating and/or maintaining sleep that occurs despite age-appropriate time and opportunity for sleep and results in some degree of impairment in daytime functioning for the child and/ or family (ranging from fatigue, irritability, lack of energy, and mild cognitive impairment to effects on mood, school performance, and quality of life)

- **Primary snoring:** snoring without associated ventilatory abnormalities on overnight polysomnogram (e.g., apneas or hypopneas, hypoxemia, hypercapnia) or respiratory-related arousals, and is a manifestation of the vibrations of the oropharyngeal soft tissue walls that occur when an individual attempts to breathe against increased upper airway resistance during sleep

- **Parasomnias:** episodic nocturnal behaviors that often involve cognitive disorientation and autonomic and skeletal muscle disturbance

- **Restless leg/Willis Ekbom syndrome:** a chronic neurologic disorder, characterized by an almost irresistible urge to move the legs, often accompanied by uncomfortable sensations in the lower extremities

- **Hypersomnia:** a group of disorders characterized by recurrent episodes of excessive daytime sleepiness, reduced baseline alertness, and/or prolonged nighttime sleep periods that interfere with normal daily functioning

- **Narcolepsy:** a chronic lifelong CNS disorder, typically presenting in adolescence and early adulthood, that is characterized by profound daytime sleepiness and resultant significant functional impairment

- **Sleep attacks:** irresistible state in that the child or adolescent is unable to stay awake despite considerable effort, and they occur even in the context of normally stimulating activities (e.g., during meals, in the middle of a conversation)

- **Hypnogogic/Hypnopompic hallucinations:** vivid visual, auditory, and sometimes tactile sensory experiences occurring during transitions between sleep and wakefulness, primarily at sleep offset (hypnopompic) and

sleep onset (hypnogogic)

- **Sleep paralysis:** the inability to move or speak for a few secs or mins at sleep onset or offset, and often accompanies the hallucinations

- **Delayed sleep phase disorder:** a circadian rhythm disorder, involves a significant, persistent, and intractable phase shift in sleepwake schedule (later sleep onset and wake time) that conflicts with the individual's normal school, work, and/or lifestyle demands

- **Rumination disorder:** the repeated regurgitation of food, where the regurgitated food may be rechewed, reswallowed, or spit out, for a period of ≥1 mth following a period of normal functioning

- **Pica:** the persistent eating of nonnutritive, nonfood substances (e.g., paper, soap, plaster, charcoal, clay, wool, ashes, paint, earth) over a period of ≥1 mth

- **Tourette disorder, persistent (chronic) motor or vocal tic, and provisional tic disorders:** disorders characterized by involuntary, rapid, repetitive, single or multiple motor

and/or vocal/phonic tics that wax and wane in frequency but have persisted for >1 yr since first tic onset (<1 yr for provisional tic disorder)

- **Stereotypic movement disorder:** a neurodevelopmental disorder characterized by repetitive, seemingly driven, and apparently purposeless motor behavior (stereotypy) that interferes with social, academic, or other activities that may result in self-injury

- **Habit:** an action or pattern of behavior that is repeated often

- **Anxiety disorder:** pathologic anxiety in which anxiety becomes disabling, interfering with social interactions, development, and achievement of goals or quality of life, and can lead to low self-esteem, social withdrawal, and academic underachievement

- **Childhood-onset social phobia (social anxiety disorder):** a disorder characterized by excessive anxiety in social settings (including the presence of unfamiliar peers, or unfamiliar adults) or performance situations, leading to social isolation and is associated with social scrutiny and fear of doing something embarrassing

- **Panic disorder:** a syndrome of recurrent, discrete episodes of marked fear or discomfort in which patients experience abrupt onset of physical and psychologic symptoms called panic attacks

- **Obsessions:** specific repetitive thoughts that invade consciousness

- **Compulsions:** repetitive rituals or movements that are driven by anxiety

- **Phobia:** excessive and unreasonable fear which can be cued by the presence or anticipation of the feared trigger, with anxiety symptoms occurring immediately

- **Systematic desensitization:** a form of behavior therapy that gradually exposes the patient to the fear-inducing situation or object, while simultaneously teaching relaxation techniques for anxiety management

- **Post traumatic stress disorder:** an anxiety disorder resulting from the long- and short-term effects of trauma that cause behavioral and physiologic sequelae

- **Mood disorders:** interrelated sets of psychiatric symptoms characterized by a core deficit in emotional self-regulation

- **Major depressive disorder:** a disorder characterized by a distinct period of ≥2 wk in which there is a depressed or irritable mood and/or loss of interest or pleasure in almost all activities that is present for most of the day, nearly every day

- **Persistent depressive disorder (dysthymia):** a disorder characterized by depressed or irritable mood for more days than not, for ≥1 yr

- **Manic episode:** a distinct period of ≥1 wk in which there is an abnormally and persistently elevated, expansive, or irritable mood and abnormally and persistently increased goal-directed activity or energy that is present for most of the day, nearly every day (or any duration if hospitalization is necessary)

- **Cyclothymic disorder:** a disorder characterized by a period of ≥1 yr in which there are numerous periods with hypomanic and depressive symptoms that do not meet criteria for a hypomanic episode or a major depressive episode, respectively

- **Eating disorders:** the disorders characterized by body dissatisfaction related to overvaluation of a thin body ideal associated with dysfunctional patterns of cognition and weight-control behaviors that result in significant biologic, psychological, and social complications

- **Anorexia nervosa:** a significant overestimation of body size and shape, with a relentless pursuit of thinness that typically combines excessive dieting and compulsive exercising in the restrictive subtype; in the binge-purge subtype, patients might intermittently overeat and then attempt to rid themselves of calories by vomiting or taking laxatives, still with a strong drive for thinness

- **Bulimia nervosa:** a disorder characterized by episodes of eating large amounts of food in a brief period, followed by compensatory vomiting, laxative use, and exercise or fasting to rid the body of the effects of overeating in an effort to avoid obesity

- **Attention deficit hyperactivity disorder:** a disorder characterised by impulsivity, hyperactivity, and a decreased ability to maintain attention

- **Avoidant Restrictive Food Intake Disorder (ARFID):** a group

of conditions in which food intake is restricted or avoided due to adverse feeding or eating experiences or the sensory qualities of food, resulting in significant nutritional deficiencies and problems with social interactions

- **Oppositional defiant disorder:** a disorder characterized by a pattern lasting ≥6 mths of angry, irritable mood, argumentative/defiant behavior, or vindictiveness exhibited during interaction with ≥1 individual who is not a sibling

- **Intermittent explosive disorder:** a disorder characterized by recurrent verbal or physical aggression that is grossly disproportionate to the provocation or to any precipitating psychosocial stressors

- **Conduct disorder:** a disorder characterized by a repetitive and persistent pattern over ≥12 mths of serious rule-violating behavior in which the basic rights of others or major societal norms or rules are violated
- **Autism spectrum disorder:** a disorder characterized by a persistent impairment in reciprocal social communication and interaction, and restricted, repetitive patterns of behavior or interests

- **Psychosis:** a severe disruption of thought, perception, and behavior resulting in loss of reality testing

- **Delusions:** fixed, unchangeable, false beliefs even in light of conflicting evidence

- **Hallucinations:** vivid and clear perception-like experiences that occur without external stimulus and have the full force and impact of normal perceptions

- **Illusions:** misinterpretations of actual sensory stimuli

- **Delirium:** an activated mental state that may include disorientation, irritability, fearful responses, and sensory misperception

- **Brief psychotic disorder:** a disorder characterized by the sudden onset (within 2 wk from baseline function) of these symptoms in the context of emotional turmoil or overwhelming confusion, followed by complete resolution

- **Neurodevelopmental dysfunctions:** disruptions of neuro-anatomic structure or psychophysiologic function and place

a child at-risk for developmental, cognitive, emotional, behavioral, psychosocial and adaptive challenges

- **Intellectual function:** the capacity to think in the abstract, reason, problem solve and comprehend

- **Memory:** the cognitive mechanism by which information is acquired, retained, and recalled

## 4. Learning Disorder

- **Dyslexia/Reading disability:** an unexpected difficulty in reading, that is, unexpected in relation to intelligence, chronological age/grade level, education, or professional status

- **Phonemic awareness:** the ability to focus on and manipulate phonemes (speech sounds) in spoken syllables and words

- **Phonology:** the correct use of speech sounds to form words

- **Semantics:** the correct use of words

- **Syntax:** the appropriate use of grammar to make sentences

- **Pragmatic abilities:** verbal and nonverbal skills that facilitate the exchange of ideas, including the appropriate choice of language for the situation and circumstance and the appropriate use of body language (i.e., posture, eye contact, gestures)

- **Developmental dysphasia/Developmental language disorder/Specific language impairment:** a disorder characterized by a significant discrepancy between the child's overall cognitive level (typically nonverbal measures of intelligence) and functional language level

- **Selective mutism:** a failure to speak in specific social situations despite speaking in other situations, and it is typically a symptom of an underlying anxiety disorder

- **Hyperlexia:** the precocious development of reading single words that spontaneously occurs in some young children (ages 2-5 yr) without specific instruction

- **Intellectual disability:** deficits in cognitive abilities and adaptive behaviors

- **Stuttering:** a disorder in the rhythm of speech in which the person knows precisely what he or she wishes to say but at the same time may have difficulty saying it because of an involuntary repetition, prolongation, or cessation of sound

## 5. Children with Special Needs

- **Adoption:** a social, emotional, and legal process that provides a new family for a child when the birth family is unable or unwilling to parent

- **Bullying:** the assertion of power through social, emotional, or physical means of aggression that involves a bully repeatedly and intentionally targeting a weaker victim

- **Cyberbullying:** an emerging form of bullying that takes place using electronic technology (text messaging, mass emailing, Internet chat rooms, social networking sites, etc.)

- **Abuse:** acts of commission, resulting in actual or potential harm

- **Neglect:** omissions in care, resulting in actual or potential harm

- **Sexual abuse:** the involvement of dependent, developmentally immature children and adolescents in sexual activities which they do not fully comprehend, to

which they are unable to give consent, or that violate the social taboos of family roles

- **Psychological abuse:** verbal abuse and humiliation and acts that scare or terrorize a child

- **Palliative care for children:** the active total care of the child's body, mind and spirit, and also involves giving support to the family….Optimally, this care begins when a life-threatening illness or condition is diagnosed and continues regardless of whether or not a child receives treatment directed at the underlying illness

# 6. Nutrition

- **Estimated average requirement:** the average daily nutrient intake level estimated to meet the requirements for 50% of the population, assuming normal distribution

- **Recommended dietary allowance:** an estimate of the daily average nutrient intake to meet the nutritional needs of >97% of the individuals in a population, and it can be used as a guideline for individuals to avoid deficiency in the population

- **Estimated energy requirement:** the average dietary energy intake predicted to maintain energy balance in a healthy individual and accounts for age, gender, weight, stature, and physical activity level

- **Glycemic index:** a measure of the height of blood sugar levels 2 hrs following ingestion against the reference standard (a slice of white bread)

- **Fibers:** nondigestible carbohydrates mostly derived from plant sources, such as whole grain, fruits, and vegetables, that escape digestion and reach the colon

nearly 100% intact

- **Vegetarianism:** the practice of following a diet that excludes animal flesh foods, including beef, pork, poultry, fish, and shellfish

- **Food security:** the state when all people, at all times, have access to sufficient, safe, nutritious food to maintain a healthy and active life

- **Severe acute malnutrition:** severe wasting and/or bilateral edema.

  A. **Severe wasting:** extreme thinness diagnosed by a weight-for-length (or height) <3 SD of the WHO Child Growth Standards. In children ages 6-59 mths, a mid-upper arm circumference <115 mm also denotes extreme thinness

  B. Bilateral edema is diagnosed by grasping both feet, placing a thumb on top of each, and pressing gently but firmly for 10 secs. A pit (dent) remaining under each thumb indicates bilateral edema

- **Failure to thrive:** is a sign, not a diagnosis. FTT is generally used to describe children younger than 2 or 3

yrs who meet any of the following criteria:

A. Growth under the third percentile on World Health Organization (WHO) weight for age growth charts (<3% = less than 3 standard deviations below mean)

B. Weight for height or body mass index (BMI) less than the 5th percentile

C. Growth patterns that have crossed two major percentiles downward on the weight for age charts within 6 mths

D. Growth velocity less than normal for age

- **Craniotabes:** a softening of the cranial bones and can be detected by applying pressure at the occiput or over the parietal bones

- **Trace element:** an element with <0.01% of the body weight

- **Acid:** a substance that releases ("donates") a hydrogen ion (H+)

- **Base:** a substance that accepts a hydrogen ion

- **Buffers:** substances that attenuate the change in pH that occurs when acids or bases are added to the body

# 7. Pediatric Drug Therapy

- **Pharmacogenetics:** the study or clinical testing of genetic variations that give rise to interindividual differences in the response to drugs

- **Pharmacogenomics:** the broader application of genome-wide technologies and strategies to identify both disease processes that represent new targets for drug development and factors predictive of efficacy and risk of adverse drug reactions

- **Metabolomics:** the study of the complete set of low-molecular weight molecules (metabolites) present in a living system (cell, tissue, organ or organism) at a particular developmental or pathological state

- **Metabonomics:** the study of how the metabolic profile of biological systems changes in response to alterations because of pathophysiologic stimuli, toxic exposures, dietary changes, etc

- **Pharmacometabonomics:** the prediction of the outcome, efficacy or toxicity, of a drug or xenobiotic intervention

in an individual based on a mathematical model of preintervention metabolite signatures

- **Pharmacokinetics:** the study of the movement of a drug throughout the body and the concentrations (or amounts) of a drug that reach a given body space and/or tissue and its residence time therein

- **Pharmacodynamics:** the relationship between drug dose or drug concentration and response

- **Elimination half life:** the time necessary for the drug concentration to decrease by 50%

- **Metabolism:** the biotransformation of an endogenous or exogenous molecule by ≥1 enzymes to moieties that are more hydrophilic and thus, can be more easily eliminated by excretion, secretion or exhalation

- **Agonist:** a drug that binds to and activates the receptor, directly or indirectly achieving the desired effect

- **Partial agonist:** a drug that binds to and activates the

receptor, but maximal effect is not achieved even in the presence of receptor saturation

- **Antagonist:** a drug that binds to a receptor preventing binding of other molecules thereby preventing activation of the receptor

- **Biomarker:** a characteristic that is objectively measured and evaluated as an indicator of normal biological processes, pathogenic processes, or pharmacologic responses to a therapeutic intervention

- **Surrogate end point:** a biomarker that is intended to substitute for a specific clinical end point

- **Therapeutic drug monitoring:** a retrospective, reactive approach whereby drug concentrations in plasma (primarily) or other biologic fluids are measured at some point during either a constant rate intravenous infusion or during a dosing interval for drugs given by intermittent dosing schedules

- **Therapeutic range:** the range of drug concentrations associated with a high degree of efficacy and a low risk of

dose-related toxicity in the majority of patients

- **Absorption:** the translocation of a drug from its site of administration into the bloodstream

- **Drug distribution:** the movement of drugs and other compounds from the systemic circulation into various body compartments, tissues, and cells

- **First-pass effect:** rapid metabolization or alteration of a drug when it passes through the intestinal mucosa or liver for the first time

- **Clinical pharmacokinetics:** a prospective, proactive approach where plasma drug concentrations are used to estimate pharmacokinetic parameters which are then used to calculate a dosing regimen required to attain a desired level of systemic exposure that would portend a desired pharmacologic response

- **Adverse drug reaction:** a response to a drug that is noxious and unintended, and occurs at doses normally used in man for the prophylaxis, diagnosis or therapy of disease or for the modication of physiological function

- **Adherence/Compliance:** the extent to which a person's behaviour (taking medication, following a diet, and/or executing lifestyle change) correspond with agreed recommendations from a healthcare provider

- **Noncompliance:** deviations from medical advice on the part of the patient (such as using the wrong route of administration, taking too low or too high a dose, or taking the medication at the wrong times or for an inappropriate length of time)

- **Malignant hyperthermia:** an acute hypermetabolic syndrome that is triggered by inhalational anesthetic agents and succinylcholine

- **Unconventional analgesic medication:** a wide number of drugs that were developed for other indications but that have been found to have analgesic properties

- **Cognitive-behavioral strategies:** techniques that teach children how to manage pain by learning new ways to think about the pain and to change behaviors associated with the pain

## 8. The Acutely Ill Child

- **Gastric lavage:** placing a tube into the stomach to aspirate contents, followed by flushing with aliquots of fluid, usually water or normal saline

- **Prehospital care:** emergency assistance rendered by trained emergency medical personnel before a child reaches a treating medical facility

- **Regionalization (emergency medical services):** a geographically organized system of services that ensures access to care at a level appropriate to patient needs while maintaining efficient use of available resources

- **Brain death:** the irreversible cessation of all functions of the entire brain, including the brainstem

- **Apnea:** the absence of respiratory effort in response to an adequate stimulus

- **Syncope:** a transient loss of consciousness and muscle tone that result from inadequate cerebral perfusion,

followed by spontaneous recovery

A. **Stretch syncope:** syncope which occurs while stretching the neck and the trunk backward and the arms outward, or during flexion of the neck, the presumed mechanism is mechanical disruption of brain perfusion caused by compression of the vertebral arteries

B. **Reflex/Situational syncope:** syncope that is triggered by specific factors or events

- **Shock:** an acute process characterized by the body's inability to deliver adequate oxygen to meet the metabolic demands of vital organs and tissues

- **Systemic inflammatory response syndrome:** an inflammatory cascade that is initiated by the host response to an infectious or noninfectious trigger

- **Respiratory failure:** state when oxygenation and ventilation are insufficient to meet the metabolic demands of the body

- **Tension pneumothorax:** a state when air accumulates under pressure in the pleural space

- **Lacerations:** tears of the skin caused by blunt or shearing forces

- **Cut:** an injury inflicted by a sharp object

- **Abrasion:** a scrape to the epidermis, and sometimes the dermis, that is usually caused by friction of the skin against a rough surface

- **Ventilation:** the process of carbon dioxide removal

- **Weaning (from ventilator):** the process in which the work of breathing is shifted from the ventilator to the patient

# 9. Human Genetics

- **Genetics:** the science of genes, heredity, and the variation of organisms

- **Linkage testing:** tracking a genetic trait through a family using closely linked polymorphic markers as a surrogate for the trait

- **Clinical validity:** the degree to which the test correctly predicts presence or absence of disease

- **False-negative results:** an inability to detect a mutation in an affected patient

- **Clinical utility:** the degree to which the results of a test guide clinical management

- **Genetic counseling:** a communication process in which the genetic contribution to health is explained, along with specific risks of transmission of a trait and options to manage the condition and its inheritance

- **Founder effect:** a specific change affecting a disease-causing gene becomes relatively common in a population derived from a small number of founders

- **Genomic disorders:** a group of diseases caused by alterations in the genome, including deletions (copy number loss), duplications (copy number gain), inversions (altered orientation of a genomic region) and chromosomal rearrangements (altered location of a genomic region)

- **Contiguous gene disorders:** a group of diseases caused by changes that affect two or more genes that contribute to the clinical phenotype and are located near each other on a chromosome

- **Translocation:** a type of chromosomal rearrangement in which a genomic region from 1 chromosome is transferred to a different location on the same chromosome or on a nonhomologous chromosome

- **Mosaicism:** a condition in which only a portion of cells that make up a person's body are affected by the single gene defect, the genomic disorder or the chromosomal defect

- **Gonadal/Germline mosaicism:** occurrence of more than one genetic constitution in the precursor cells of eggs or sperm

- **Polygenic disorders:** the disorders caused by the cumulative effects of changes or variations in more than 1 gene

- **Multifactorial disorders:** the disorders caused by the cumulative effects of changes or variations in multiple genes and/or the combined effects of both genetic and environmental factors

- **Loss-of-function mutation:** a reduction in the level of protein function as a result of decreased expression or production of a protein that does not work as efficiently

- **Haploinsufficiency:** the situation in which maintenance of a normal phenotype requires the proteins produced by both copies of a gene, and a 50% decrease in gene function results in an abnormal phenotype

- **Gain-of-function mutations:** the mutations which result in production of a protein molecule with an increased ability to perform a normal function or they can confer a novel

property on the protein

- **Polymorphisms:** the genetic variants in a single gene that occur at a frequency of >1% in a population

- **Anticipation:** occurrence of a genetic disorder at earlier age of onset and/or at increased severity in successive generations

- **Alternative splicing:** formation of diverse mRNAs through differential splicing of an mRNA precursor

- **Codon:** three consecutive bases/nucleotides in DNA/RNA that specify an amino acid

- **Allele:** one of a series of alternative DNA sequences for a particular gene

- **Exon:** segment of a gene (usually protein coding) that remains after splicing of the primary RNA transcript

- **Intron:** segment of a gene transcribed into the primary RNA

transcript but excised during exon splicing, thus does not code for a protein

- **Genome:** the individual's (or cell's) total genetic information

- **Transcription:** production of mRNA from DNA template

- **Translation:** the process by which protein is synthesized from an mRNA sequence

- **Karyotype:** classified chromosome complement of an individual or a cell

- **Domain:** segment of a protein associated with a specialized structure or function

- **Crossover:** exchange of genetic material between homologous chromosomes during meiosis

- **Conservation:** sequence similarity for genes present in two distinct organisms or for gene families; can be detected by measuring the sequence similarity at nucleotide or amino

acid level

- **Concordant:** both members of a twin pair show the trait
- **Discordant:** only one member of a twin pair shows the trait
- **Compound:** individual with two different mutant alleles at a locus
- **Genotype:** the internally coded, heritable information of an individual and can also be used to refer to which particular alternative version (allele) of a gene is present at a specic location (locus) on a chromosome
- **Phenotype:** the observed structural, biochemical, and physiologic characteristics of an individual, determined by the genotype, and can also refer to the observed structural and functional effects of a mutant allele at a specific locus
- **Germline:** cell lineage resulting in eggs or sperm
- **Germline mutation:** any detectable, heritable variation in the

lineage of germ cells transmitted to offspring while those in somatic cells are not

- **Heterozygote:** person with one normal and one mutant allele at a given locus on a pair of homologous chromosomes

- **Homozygote:** person with identical alleles at a given locus on a pair of homologous chromosomes

- **Pedigree:** a graphic depiction of a family's structure and medical history

- **Proband:** the child or adolescent who is being evaluated

- **First-degree relatives:** the relatives who share 1/2 of their genetic information, such as a parent, full sibling, or child

- **Horizontal transmission:** the observation of multiple affected members of a kindred in the same generation, but no affected family members in other generations

- **Digenic inheritance:** two genes interacting to produce a

disease phenotype

- **Lyon hypothesis/X inactivation:** principle of inactivation of one of the two X chromosomes in normal female cells

- **Pleiotropy:** multiple effects of a single gene

- **Consanguinity:** the existence of a relationship by a common ancestor and increases the chance that both parents carry a gene affected by an identical mutation that they inherited

- **Heteroplasmy:** a condition in which a cell can have a mixture of normal and abnormal mitochondrial genomes

- **Homoplasmy:** a condition in which a daughter cell receives all wild-type or all mutant mtDNA

- **Epigenetics:** the study of the causal interactions between genes and the phenotype

- **Polyploid cells:** the euploid cells with more than the

normal diploid number of 46 (2n) chromosomes: 3n, 4n

- **Aneuploid cells:** abnormal cells that do not contain a multiple of haploid number of chromosomes

- **Nondisjunction:** the failure of chromosomes to disjoin normally during meiosis

- **Inversion:** a single chromosome break at 2 points; the broken piece is then inverted and joined into the same chromosome

- **Microdeletion:** a loss of small chromosome regions, the largest of which are detectable only with prophase chromosome studies and/or molecular methods

- **Duplication:** the presence of extra genetic material from the same chromosome

- **Contiguous gene deletion:** a deletion involving more than a single gene

- **Insertions:** a condition when a piece of a chromosome broken at 2 points is incorporated into a break in another part of a chromosome

- **Turner syndrome:** a condition characterized by complete or partial monosomy of the X chromosome and defined by a combination of phenotypic features

- **Uniparental disomy:** a condition when both chromosomes of a pair or areas from 1 chromosome in any individual have been inherited from a single parent

- **Genomic imprinting:** a condition when the phenotypic expression of a gene depends on the parent of origin for certain genes or in some cases entire chromosome regions

- **Familiality:** the ratio of the prevalence rate in siblings and/or parents to the prevalence rate in the general population

- **Heritability of a trait:** the estimate of the fraction of the total variance contributed by genetic factors

- **Locus heterogeneity:** the situation in which a trait results

from the independent action of >1 gene

- **Allelic heterogeneity:** the presence of >1 variant in a particular gene contributing to disease risk

- **Phenocopy:** the development of a trait or disease from a nongenetic mechanism

- **Penetrance:** a phenomenon that any variant or allele (inherited unit, DNA segment, or chromosome) in a gene has a certain probability of being affected with a specific gene variant-associated disease

- **Empiric risk:** recurrence risk based on experience rather than calculation

- **Telomeres:** a series of tens to thousands of TTAGGG repeats at the ends of chromosomes that are important for stabilizing the chromosomal ends and limiting breakage, translocation, and loss of DNA material

# 10. The Fetus and the Neonatal Infant

- **Family planning:** a way of thinking and living that is adopted voluntarily, upon the basis of knowledge, attitudes and responsible decisions, by individuals and couples, in order to promote the health and welfare of the family group and thus contribute effectively to the social development of a country

- **High-risk pregnancies:** those pregnancies that increase the likelihood of abortion, fetal death, preterm delivery, intrauterine growth restriction, poor cardiopulmonary or metabolic transitioning at birth, fetal or neonatal disease, congenital malformations, or mental retardation and other handicaps

- **Maternal death:** the death of a woman while pregnant or within 42 days of termination of pregnancy, irrespective of the duration and site of pregnancy, from any cause related to or aggravated by the pregnancy or its management but not from accidental or incidental causes

- **Maternal mortality ratio:** the number of maternal deaths (during pregnancy and within 42 days postpartum) per 1,00,000 live births

- **Maternal mortality rate:** the number of maternal deaths (during pregnancy and within 42 days postpartum) per 1,00,000 pregnant females

- **Still birth:** fetal deaths greater than 1000 g or more than 28 completed wks per 1000 total births

- **Perinatal mortality rate:** stillbirths plus early neonatal deaths (upto 1st wk of life) per 1000 total births

- **Neonatal mortality rate:** deaths at less than 28 days per 1000 live births

- **Postneonatal mortality rate:** deaths from 28 days until 1 yr per 1000 live births

- **Infant mortality rate:** deaths in the first yr of life per 1000 live births

- **Superfecundation:** the fertilization of an ovum by an insemination that takes place after 1 ovum has already been fertilized

- **Superfetation:** the fertilization and subsequent development of an embryo when a fetus is already present in the uterus

- **Labor:** a series of events that take place in the genital organs in an effort to expel the viable products of conception (fetus, placenta and the membranes) out of the womb through the vagina into the outer world

- **Delivery:** the expulsion or extraction of a viable fetus out of the womb

- **Normal labor/Eutocia:** labor is called normal if it fulfills the following criteria

  A. Spontaneous in onset and at term

  B. With vertex presentation

  C. Without undue prolongation

  D. Natural termination with minimal aids

  E. Without having any complications affecting the health of the mother and/or the baby

- **Abnormal labor/Dystocia:** any deviation from the definition of normal labor

- **Polyhydramnios:** a state where liquor amnii >2,000 mL or AFI >25 cm or maximum vertical pocket of liquor >8 cm (>95th percentile)

- **Oligohydramnios:** a state where liquor amnii <200 mL at term or the maximum vertical pocket of liquor <2 cm or when amniotic fluid index (AFI) <5 cm (<5th percentile)

- **Prolonged labor:** a state when the combined duration of the first and second stage is more than the arbitrary time limit of 18 hrs

- **Erythroblastosis fetalis:** a condition caused by the transplacental passage of maternal antibody active against paternal RBC antigens of the infant and is characterized by an increased rate of RBC destruction

- **Chorioamnionitis:** the clinical syndrome of intrauterine infection, which includes maternal fever, with or without local or systemic signs of chorioamnionitis (uterine tenderness, foul-smelling vaginal discharge/amniotic fluid, maternal leukocytosis, maternal and/or fetal tachycardia)
- **Fetal/Intrauterine growth restriction:** any fetus that does not reach his or her intrauterine growth potential or

fetuses weighing <10th percentile for gestational age

- **Chronic hypertension:** hypertension preceding pregnancy or first diagnosed before 20 wks' gestation or that persists 6 to 12 wks postpartum

- **Chronic hypertension with superimposed preeclampsia:** worsening hypertension and new-onset proteinuria, in addition to possible concurrent thrombocytopenia, or transaminase derangements after the 20th wk of pregnancy in a woman with known chronic hypertension

- **Gestational hypertension:** new onset of sustained elevated blood pressure (≥140 mm Hg systolic or ≥90 mm Hg diastolic) in a previously normotensive woman, detected after 20 wks' gestation

- **Preeclampsia:** new onset elevated blood pressures (≥140 mm Hg systolic or ≥90 mm Hg diastolic) with proteinuria (≥300 mg of protein in a 24-hr period or protein/ creatinine ratio ≥0.3 mg/mg), detected after 20 wks' gestation
- **Eclampsia:** a generalized tonic-clonic seizure activity in a pregnant woman with preeclampsia, that cannot be

attributed to other causes, with no prior history of a seizure disorder

- **Gestational diabetes mellitus:** glucose intolerance beginning or diagnosed for the first time during pregnancy, after 20 wks of gestation

- **Hydrops fetalis:** the presence of extracellular fluid in ≥2 fetal body compartments. These fluid colections include skin edema (>5 mm thickness), pericardial effusion, pleural effusions, and ascites

- **Fetal growth discordance (twin pregnancy):** a difference in birth weight >20% of the larger twin's weight

- **Intrauterine fetal demise (twin pregnancy):** fetal demise after 20 wks' gestation but before delivery and is confirmed by ultrasonographic evidence of absent fetal cardiac activity

- **Perinatal period:** the period from the 28th wk of gestation through the 7th day after birth

- **Neonatal period:** the 1st 28 days after birth

- **Transitional period:** the first 4 to 6 hrs after birth

- **Infancy:** the 1st yr after birth

- **Fontanel:** a wide gap in the suture line

- **Molding:** the alteration of the shape of the forecoming head while passing through the resistant birth passage during labor

- **Caput succedaneum:** a diffuse, sometimes ecchymotic, edematous swelling of the soft tissues of the scalp involving the area presenting during vertex delivery

- **Cephalohematoma:** a subperiosteal hemorrhage occurring due to injury during delivery, hence always limited to the surface of 1 cranial bone

- **Subgaleal hemorrhage:** a collection of blood beneath the aponeurosis that covers the scalp and serves as the insertion for the occipitofrontalis muscle

- **Trophic feeding/Minimal enteral nutrition/Gut priming:** the nonnutritive use of very small volumes of human milk or formula, for the intended purpose of preservation of gut maturation rather than nutrient delivery

- **Anoxia:** the consequences of complete lack of oxygen as a result of a number of primary causes

- **Hypoxemia:** a decreased arterial concentration of oxygen

- **Hypoxia:** a decreased oxygenation to cells or organs

- **Ischemia:** the blood flow to cells or organs that is insufficient to maintain their normal function

- **Diaphragmatic hernia:** a communication between the abdominal and thoracic cavities with or without abdominal contents in the thorax

- **Eventration of the diaphragm:** an abnormal elevation, consisting of a thinned diaphragmatic muscle that causes elevation of the entire hemidiaphragm or, more commonly, the anterior aspect of the hemidiaphragm

- **Plethora:** a ruddy, deep red-purple appearance associated with a high hematocrit, is often due to polycythemia, defined as a central hematocrit ≥65%

- **Omphalocele:** a herniation or protrusion of the abdominal contents into the base of the umbilical cord

- **Hyperthermia in a newborn:** the elevations in temperature (38-39°C [100-103°F]) occasionally noted on the 2nd or 3rd day after birth in infants whose clinical course has been otherwise satisfactory

- **Dysmorphology:** the study of abnormalities of human form and the mechanisms that cause them

- **Malformation:** a primary structural defect arising from a localized error in morphogenesis and resulting in the abnormal formation of a tissue or organ

- **Dysplasia:** an abnormal organization of cells into tissues

- **Deformation:** an alteration in shape or structure of a structure or organ that has differentiated normally

- **Disruption:** a structural defect resulting from the destruction of a structure that had formed normally before the insult

- **Syndrome:** a pattern of multiple abnormalities that are related by pathophysiology and result from a single, defined etiology

- **Sequences:** multiple malformations that are caused by a single event that can have many etiologies

- **Association:** a nonrandom collection of malformations in which there is an unclear or unknown relationship among the malformations such that they do not fit the criteria for a syndrome or sequence

- **Cephalization:** a diversion of blood flow away from nonvital organs such as kidney (resulting in oligohydramnios) toward vital organs such as the brain, in response to increasing hypoxia

- **Neonatal tetanus (surveillance case definition):** the ability of a newborn to suck at birth and for the 1st few days of life, followed by an inability to suck starting between 3 and 10 days of age, difficulty swallowing, spasms, stiffness,

seizures, and death

- **Birth injury:** an impairment of the infant's body function or structure due to adverse influences that occurred at birth. Injury may occur antenatally, intrapartum, or during resuscitation and may be avoidable or unavoidable

- **Resuscitation:** efforts at delivery designed to help the newborn make the respiratory and circulatory transitions that must be accomplished immediately after birth

- **Small for gestational age:** a neonate whose birth weight or birth crown-heel length is <10th percentile for GA

- **Large for gestational age:** a neonate whose birth weight or birth crown-heel length is >90th percentile for GA.
- **Epstein pearls:** small white inclusion cysts clustered about the midline at the juncture of the hard and soft palate

- **Excessive high-pitched cry:** the cry when the infant is unable to self-console in 15 secs or continuous up to 5 mins despite intervention

- **Excessive sucking:** rooting with more than three attempts noted to suck fist, hand, or pacifier before or after feeding

- **Kangaroo care:** skin-to-skin holding technique consistently associated with improved infant outcomes (i.e., fewer respiratory complications, improved weight gain, and temperature regulation) and maternal outcomes (i.e., improved maternal competence and longer breastfeeding duration)

- **Hyperthermia:** an elevated core body temperature, may be caused by a relatively hot environment, infection, dehydration, CNS dysfunction, or medications

- **Radiation:** heat dissipates from the infant to a colder object in the environment

- **Convection:** heat is lost from the skin to moving air

- **Evaporation:** heat is lost through conversion of water to gas

- **Conduction:** heat is lost due to transfer of heat from the infant to the surface on which he or she lies

- **NICU discharge readiness:** the attainment of technical skills and knowledge, emotional comfort, and confidence with infant care by the primary caregivers at the time of discharge

- **NICU discharge preparation:** the process of facilitating discharge readiness to successfully make the transition from the NICU to home

- **Engorgement (breasts):** a severe form of increased breast fullness that usually presents on day 3 to 5 postpartum signaling the onset of copious milk production

- **Neonatal hyperglycemia:** a whole blood glucose level >125 mg/dL or plasma glucose values >145 mg/dL

- **Neonatal hypocalcemia:** a total serum calcium concentration <7 mg/dL or an ionized calcium concentration <4 mg/dL (1 mmol/L)

- **Neonatal hypercalcemia:** a serum total calcium level >11 mg/dL or serum ionized calcium level >5.8 mg/dL

- **Neonatal hypermagnesemia:** a serum magnesium level >3 mg/dl

- **Neonatal hypomagnesemia:** a serum magnesium level <1.6 mg/dl

- **Neonatal hyperbilirubinemia:** a serum total bilirubin >95th percentile on the hr specific Bhutani nomogram

- **Severe hyperbilirubinemia:** a serum total bilirubin >25 mg/dL in term and late preterm infants, and presumably lower in preterm infants

- **Kernicterus:** the chronic and permanent sequelae of bilirubin toxicity that develop during the first yr of age

- **Necrotising enterocolitis:** an acute inflammatory injury of the distal small and often proximal large intestine

- **Neonatal proteinuria:** urinary proteins >250 mg/m2/day

- **Neonatal massive proteinuria:** urinary proteins >1.5g/m2/day

- **Renal tubular acidosis:** metabolic acidosis resulting from the inability of the kidney to excrete hydrogen ions or to reabsorb bicarbonate

- **Capnography:** the noninvasive measurement of the partial pressure of carbon dioxide in exhaled breaths, expressed as the CO2 concentration over time

- **Apnea in neonate:** the cessation of airflow. Apnea is pathologic (an apneic spell) when absent airflow is prolonged (usually ≥20 sec) or accompanied by bradycardia (heart rate <100 bpm) or hypoxemia that is detected clinically (cyanosis) or by oxygen saturation monitoring

  A. **Central apnea:** inspiratory efforts are absent

  B. **Obstructive apnea:** inspiratory efforts persist in the presence of airway obstruction, usually at the pharyngeal level

  C. **Mixed apnea:** airway obstruction with inspiratory efforts precedes or follows central apnea

- **Transient tachypnea of the newborn:** a benign, self-limited process resulting from delayed clearance of fetal lung fluid characterized by tachypnea with signs of mild respiratory distress including retractions and cyanosis;

decreased oxygen saturation is usually alleviated by supplemental oxygen with FiO2 <0.04

- **Bronchopulmonary dysplasia:**

  A. For infants born at <32 wks' gestation who received supplemental O2 for their first 28 days, BPD is defined at 36 wks' postmenstrual age (PMA) as

  1. **Mild:** no supplemental O2 requirement
  2. **Moderate:** supplemental O2 requirement <30%
  3. **Severe:** supplemental O2 requirement ≥30% and/or continuous positive airway pressure (CPAP) or ventilator support

  B. For infants born at ≥32 wks, BPD is defined as supplemental O2 requirement for the first 28 days with severity level based on O2 requirement at 56 days

- **Persistent pulmonary hypertension of the newborn:** disruption of the normal perinatal fetal to neonatal circulatory transition characterized by sustained elevation in pulmonary vascular resistance (PVR) rather than the decrease in PVR that normally occurs at birth

- **Pulmonary hemorrhage:** the presence of hemorrhagic fluid

in the trachea accompanied by respiratory decompensation that requires increased respiratory support or intubation within 60 mins of the appearance of fluid

- **Cyanosis:** bluish tinge of the skin and mucous membranes

  A. **Central cyanosis:** cyanosis that is associated with arterial desaturation

  B. **Peripheral cyanosis:** cyanosis associated with normal arterial saturation

  C. **Circumoral cyanosis:** cyanosis around the mouth

  D. **Acrocyanosis:** a bluish or red discoloration of the fingers and toes of normal newborns in the presence of normal arterial oxygen saturation

- **Polycythemia:** venous hematocrit ≥65%

- **Hyperviscosity:** viscosity >2 SD greater than the mean

- **Neonatal thrombocytopenia:** a platelet count <150 × 10^3/μL

  A. **Mild** (100 to 149 × 10^3/μL)

  B. **Moderate** (50 to 99 × 10^3/μL)

  C. **Severe** (<50 × 10^3/μL)

- **Omphalitis:** a condition characterized by erythema and/or induration of the periumbilical area with purulent discharge from the umbilical stump

- **Ophthalmia neonatorum:** an inflammation of the conjunctiva within the first mth of life

- **Congenital syphilis:** demonstration of *T. pallidum* by darkfield microscopy, polymerase chain reaction, or immunohistochemical test, or special stains of specimens from lesions, placenta, umbilical cord, or autopsy material

- **Latent syphilis:** the period after infection when patients are seroreactive but demonstrate no clinical manifestations of disease

- **Tuberculosis exposure:** a condition when an individual has had contact with a case of contagious tuberculosis disease in the past 3 mths

- **Tuberculosis infection:** a condition when an individual has a positive tuberculin skin test result or a positive interferon gamma release assay result, a normal physical exam, and a chest radiograph that is either normal or shows evidence of

healed calcifications

- **Tuberculosis disease:** a condition when an evident illness (signs, symptoms, and/or radiographic changes) is caused by *Mycobacterium tuberculosis*

- **Congenital tuberculosis disease:** a condition when a neonate is infected with *M. tuberculosis* in utero or during delivery and develops disease afterward. This is determined by having a positive acid-fast bacillus stain or culture from the neonate, with exclusion of possible postnatal transmission, and either lesions in the first wk of life, primary hepatic complex or caseating hepatic granulomas, or tuberculosis infection of the placenta or maternal genital tract

- **Postnatally acquired tuberculosis disease:** a condition when an infant is infected after delivery, either through inhalation of *M. tuberculosis* from a contagious caregiver or ingestion of *M. tuberculosis* via infected breast or cow milk, and develops signs, symptoms, and/or radiographic evidence of tuberculosis disease

- **Perinatal asphyxia:** a condition during the first and second stage of labor in which impaired gas exchange leads to fetal acidosis, hypoxemia, and hypercarbia

- **Congenital anomaly:** an internal or external structural defect that is identifiable at birth

- **Developmental:** a deviation that occurs over time; one that might not be present or apparent at birth

- **Dysmorphisms:** anomalous external physical features

- **Myelomeningocele:** a saccular outpouching of neural elements (neural placode), typically through a defect in the bone and the soft tissues of the posterior thoracic, sacral, or lumbar regions

- **Encephalocele:** a defect of anterior neural tube closure is an outpouching of dura with or without brain, noted in the occipital region

- **Anencephaly:** a defect in the cranial vault and posterior occipital bone, exposing the derivatives of the neural tube, including both brain and bony tissue

- **Meningocele:** herniation of the meninges through a defect in the posterior vertebral arches or the anterior sacrum

- **Congenital muscular torticollis:** a disorder characterized by limited motion of the neck, asymmetry of the face and skull, and a tilted position of the head

- **Congenital scoliosis:** a lateral curvature of the spine secondary to a failure either of formation of a vertebra or of segmentation

- **Sacral agenesis:** the absence of part or all of 2 lower vertebral bodies

- **Osteopenia:** a condition when postnatal bone mineralization is inadequate to fully mineralize bones

- **Vernix caseosa:** a greasy substance that protects the fetal skin during the lengthy immersion in amniotic fluid

- **Inborn errors of metabolism:** a group of disorders each of which results from deficient activity of a single enzyme in a metabolic pathway

# 11. Adolescent Medicine

- **Puberty:** the biologic transition from childhood to adulthood

- **Gender identity:** a person's basic sense of being a boy/man, girl/ woman, or other gender (e.g., transgender)

- **Gender role:** one's role in society, typically either the male or female role

- **Social sex role/Gender expression:** characteristics in personality, appearance, and behavior that are, in a given culture and time, considered masculine or feminine

- **Sexual orientation:** attractions, behaviors, fantasies, and emotional attachments toward men, women, or both

- **Sexual behavior:** any sensual activity to pleasure oneself or another person sexually

- **Gender variant:** any gender identity or role that varies from what is typically associated with one's sex assigned at birth

- **Transgender people:** a diverse group of individuals who cross or transcend culturally defined categories of gender

  A. **Transsexuals:** people who typically live in the cross-gender role and seek hormonal and/or surgical interventions to modify primary or secondary sex characteristics

  B. **Cross-dressers/Transvestites:** people who wear clothing and adopt behaviors associated with the other sex for emotional or sexual gratification and may spend part of the time in the cross-gender role

- **Sexual orientation:** the degree of attraction to the people of a particular sex

- **Childhood gender incongruity/dysphoria:** a distinct phenom--enon in which an individual's gender identity differs from phenotypic sex and assigned gender at birth

- **Violence:** the intentional use of physical force or power, threatened or actual, against oneself, another person, or against a group or community that either results in or has a high likelihood of resulting in injury, death, psychologic harm, maldevelopment or deprivation

- **Oppositional defiant disorder:** recurrent pattern of negativistic, defiant, disobedient, and hostile behavior toward authority figures that has a significant adverse effect on functioning (e.g., social, academic, occupational)

- **Conduct disorder:** repetitive and persistent pattern of behavior that violates the basic rights of others or major age-appropriate societal norms or rules

- **Legal label juvenile delinquency:** offenses that are illegal because of age; illegal acts

- **Huffing:** the practice of inhaling fumes accomplished using a paper bag containing a chemical-soaked cloth, spraying aerosols directly into the nose/ mouth, or using a balloon, plastic bag, or soda can filled with fumes

- **Chronic fatigue syndrome:** a complex, diverse, and debilitating illness characterized by chronic or intermittent fatigue accompanied by selected symptoms for >3 mths (young children) or >6 mths duration (adolescents or adults)

# 12 .Immunology

- **Chemotaxis:** the direct migration of cells into sites of infection, involving a complex series of events

- **Phagocytosis:** a process of particle ingestion

- **Leukopenia:** an abnormally low number of white blood cells in the circulating blood secondary to a paucity of lymphocytes, granulocytes or both

- **Neutropenia:** a decrease in the absolute number of circulating segmented neutrophils and bands in the peripheral blood

  A. **Chronic neutropenia:** lasts >3 mths and arises from reduced production, increased destruction or excessive splenic sequestration of neutrophils

- **Agranulocytosis:** absolute neutrophil count <200/cumm

- **Lymphopenia:** absolute lymphocyte count <3000/cumm for infants, and <100/cumm for older children

- **Leukocytosis:** an elevation in the total leukocyte count >2 SD above the mean count for a particular age

- **Leukemoid reaction:** a WBC count >50,000/cumm

- **Basophilia:** an absolute basophil count >120 cells/cumm

- **Immunization:** the process of inducing immunity against a specific disease

- **Vaccines:** whole or parts of microorganisms administered to prevent an infectious disease

- **Toxoid:** a modified bacterial toxin that is made nontoxic but is still able to induce an active immune response against the toxin

- **Autoimmune lymphoproliferative/Canale-Smith syndrome:** a disorder of abnormal lymphocyte apoptosis leading to polyclonal populations of T cells (double-negative T cells), which express CD3 and / antigen receptors but do not have CD4 or CD8 coreceptors (CD3 + T cell receptor /+ CD4 CD8)

- **Hyper-IgM syndrome:** a condition genetically heterogeneous and characterized by normal or elevated serum IgM levels associated with low or absent IgG, IgA, and IgE serum levels, indicating a defect in the class-switch recombination process

- **T-cell activation defects:** defects characterized by the presence of normal or elevated numbers of blood T cells that appear phenotypically normal but fail to proliferate or produce cytokines normally in response to stimulation with mitogens, antigens, or other signals delivered to the T cell receptors (TCR), owing to defective signal transduction from the TCR to intracellular metabolic pathways

- **Wiskott-Aldrich syndrome:** an X-linked recessive syndrome, characterized by atopic dermatitis, thrombocytopenic purpura with normal-appearing megakaryocytes but small defective platelets, and undue susceptibility to infection

- **Ataxia-telangiectasia:** a complex syndrome with immunologic, neurologic, endocrinologic, hepatic, and cutaneous abnormalities

- **Chronic granulomatous disease:** a disease characterized by neutrophils and monocytes capable of normal chemotaxis,

ingestion, and degranulation, but unable to kill catalase positive microorganisms because of a defect in the generation of microbicidal oxygen metabolites

- **Hematopoietic stem cell transplantation:**

  A. **Primary graft failure:** failure to achieve a neutrophil count of 200/cumm by 21 days posttransplantation

  B. **Secondary graft failure:** loss of peripheral blood counts following initial transient engraftment of donor cells

- **Allergy/Atopy:** altered state of reactivity to common environmental and food antigens that do not cause clinical reactions in unaffected people

- **Allergic rhinitis:** an inflammatory disorder of the nasal mucosa marked by nasal congestion, rhinorrhea, and itching, often accompanied by sneezing and conjunctival inflammation

- **Asthma:** a chronic inflammatory condition of the lung airways resulting in episodic airflow obstruction
- **Cold urticaria:** a condition characterized by the development of localized pruritus, erythema, and urticaria/angioedema

after exposure to a cold stimulus

- **Cholinergic urticaria:** a condition characterized by the onset of small punctate pruritic wheals surrounded by a prominent erythematous flare associated with exercise, hot showers, and sweating

- **Dermatographism/Urticaria factitia:** the ability to write on skin

- **Urticaria:** transient, pruritic, erythematous, raised wheals, with flat tops and edema that may become tense and painful

- **Angioedema:** pruritis involving the deeper subcutaneous tissues in locations such as the eyelids, lips, tongue, genitals, dorsum of the hands or feet, or wall of the gastrointestinal tract

- **Anaphylaxis:** a serious allergic reaction that is rapid in onset and may cause death

- **Atopic dermatitis:** a form of eczema that generally

begins in early infancy and is characterized by pruritus, a chronically relapsing course, and association with asthma and allergic rhinitis

- **Systemic lupus erythematosus:** a chronic autoimmune disease characterized by multisystem inflammation and the presence of circulating autoantibodies directed against self-antigens

- **Heliotrope rash:** a blue-violet discoloration of the eyelids that may be associated with periorbital edema

- **Gottron papules:** bright pink or pale, shiny, thickened or atrophic plaques over the proximal interphalangeal joints and distal interphalangeal joints and occasionally on the knees, elbows, small joints of the toes, and ankle malleoli

- **Scleredema:** a transient, self-limited disease of both children and adults that has sudden onset after a febrile illness (especially streptococcal infections) and is characterized by patchy sclerodermatous lesions on the neck and shoulders and extending to the face, trunk, and arms

- **Chilblains:** a condition with episodic color changes and the

development of nodules related to severe cold exposure and spasm-induced vessel and tissue damage

- **Sjögren syndrome:** a chronic, inflammatory, autoimmune disease characterized by progressive lymphocytic and plasma cell infiltration of the exocrine glands, especially salivary and lacrimal, with potential for systemic manifestations

- **Hereditary periodic fever syndromes:** a group of monogenic diseases that present with recurrent bouts of fever and associated pleural and/or peritoneal inflammation, arthritis, and various types of skin rash

- **Amyloidosis:** a group of diseases characterized by extracellular deposition of insoluble, fibrous amyloid proteins in various body tissues

- **Hereditary autoinflammatory diseases:** a group of illnesses that are characterized by attacks of seemingly unprovoked recurrent inflammation without significant levels of either autoantibodies or antigen-specific Tbcells, which are typically found in patients with autoimmune diseases

- **Vasculitis:** an inflammation of blood vessels

- **Wegener's granulomatosis:** a granulomatous inflammation involving the respiratory tract and necrotising vasculitis affecting small- to medium- sized vessels

- **Microscopic polyangiitis/polyarteritis:** a nongranulomatous, multisystem, pauci-immune vasculitis without upper airway involvement

- **Polyarteritis nodosa:** a necrotising vasculitis of medium- and/or small-sized arteries

- **Henoch-Schönlein purpura:** vasculitis characterized by leukocytoclastic vasculitis and immunoglobulin (Ig) A deposition in the small vessels in the skin, joints, gastrointestinal tract, and kidney

- **Relapsing polychondritis:** a condition characterized by episodic chondritis causing cartilage destruction and deformation of the ears (sparing the earlobes), nose, larynx and tracheobronchial tree

- **Sweet syndrome/Acute febrile neutrophilic dermatosis:** a condition characterized by fever, elevated neutrophil count, and raised, tender erythematous plaques and nodules over the face, extremities, and trunk

## 13. Infectious diseases

- **Prebiotic:** nondigestible food components that beneficially affect the host by selectively stimulating the growth and/or activity of 1 or a limited number of bacteria in the colon and thereby improving host health

- **Probiotics:** viable organisms that have health benefits following administration

- **Clean wounds:** uninfected operative wounds where no inflammation is noted at the operative site, and respiratory, alimentary, and genitourinary tracts and the oropharynx are not entered

- **Clean-contaminated wounds:** operative wounds in which the respiratory, alimentary, or genitourinary tract is entered under controlled conditions and that do not have unusual bacterial contamination preoperatively

- **Contaminated wounds:** open, fresh, and accidental wounds; major breaks in otherwise sterile operative technique; gross spillage from the gastrointestinal tract; penetrating trauma occurring <4 hr earlier; and incisions in which acute

nonpurulent inflammation is encountered

- **Dirty and infected wounds:** penetrating traumatic wounds >4 hr prior to surgery, those with retained devitalized tissue, and those in which clinical infection is apparent or in which the viscera have been perforated

- **Travel medicine:** specialty which provide specialized guidance on the infectious and noninfectious risks based on age, itinerary, duration, season, purpose of travel, and underlying traveler characteristics (health and vaccination status)

- **Fever:** a rectal temperature ≥38°C (100.4°F)

- **Hyperpyrexia:** a rectal temperature ≥40°C (104°F)

- **Fever without a focus:** a rectal temperature ≥38°C (100.4°F) as the sole presenting feature

  A. **Fever without localising signs:** fever of acute onset, with duration of <1 wk and without localizing sign

  B. **Fever of unknown origin:** fever documented by a healthcare provider and for which the cause could not be

identified after 3 wk of evaluation as an outpatient or after 1 wk of evaluation in the hospital

- **Catheter associated infections**

  A. **Exit-site infection:** infection localized to the exit site, without significant tracking along the tunnel, often with purulent discharge

  B. **Tunnel-tract infection:** infection in the subcutaneous tissues tracking along a tunneled catheter, which may also include serous or serosanguineous discharge from a draining sinus along the path

  C. **Pocket infection:** suppurative infection of a subcutaneous pocket containing a totally implanted device

- **Nosocomial/Hospital acquired infection:** infection acquired during hospital care which are not present or incubating at admission

- **Toxic shock syndrome:** an acute and potentially severe illness characterized by fever, hypotension, erythematous rash with subsequent desquamation on the hands and feet, and multisystem involvement, including vomiting, diarrhea, myalgias, nonfocal neurologic abnormalities, conjunctival hyperemia, and strawberry tongue

- **Facultative anaerobes:** organisms being able to survive in the presence of oxygen but growing better in reduced oxygen tensions

- **Obligate anaerobes:** organisms which cannot survive any exposure to oxygen

- **Vincent angina/Acute necrotizing ulcerative gingivitis/ Trench mouth:** an acute, fulminating, mixed anaerobic bacterial-spirochetal infection of the gingival margin and floor of the mouth

- **Ludwig angina:** an acute, life-threatening cellulitis of dental origin of the sublingual and submandibular spaces

- **Cerebral malaria:** the presence of coma in a child with *P. falciparum* parasitemia and an absence of other reasons for coma

# 14. The Digestive System

- **Dysphagia:** difficulty in swallowing

- **Odynophagia:** painful swallowing

- **Globus:** the sensation of something stuck in the throat without a clear etiology

- **Swallowing:** a complex process that starts in the mouth with mastication and lubrication of food that is formed into a bolus

- **Regurgitation:** the effortless movement of stomach contents into the esophagus and mouth

- **Gastroesophageal reflux:** the physiologic retrograde passage of fluid from the stomach to the esophagus

  A. **Gastroesophageal reflux disease:** the pathologic condition wherein such retrograde flow into the esophagus causes medical complications

- **Anorexia:** prolonged lack of appetite

- **Vomiting:** a highly coordinated reflex process that may be preceded by increased salivation and begins with involuntary retching followed by violent descent of the diaphragm and constriction of the abdominal muscles with relaxation of the gastric cardia actively force gastric contents back up the esophagus

- **Open bite:** a condition when the posterior mandibular and maxillary teeth make contact with each other, but the anterior teeth are still apart

- **Closed/Deep bite:** a condition when mandibular anterior teeth occlude inside the maxillary anterior teeth in an overclosed position

- **Cheilitis:** the dryness of the lips followed by scaling and cracking and accompanied by a characteristic burning sensation

- **Ankyloglossia/Tongue-tie:** a condition characterized by an abnormally short lingual frenum that can hinder the tongue movement but rarely interferes with feeding or speech

- **Geographic tongue/Migratory glossitis:** a benign and asymptomatic lesion characterized by ≥1 smooth, bright red patches, often showing a yellow, gray, or white membranous margin on the dorsum of an otherwise normally roughened tongue

- **Fissured/Scrotal tongue:** a malformation manifested clinically by numerous small furrows or grooves on the dorsal surface

- **Santmyer swallow:** a reflex characterised by facial stimulation by a puff of air inducing swallowing and esophageal peristalsis in healthy young infants

- **Dysphagia lusoria:** the dysphagia produced by a developmental vascular anomaly, which is often an aberrant right subclavian artery or right-sided or double aortic arch

- **Achalasia:** a primary esophageal motor disorder of unknown etiology characterized by loss of lower esophageal sphincter relaxation and loss of esophageal peristalsis, both contributing to a functional obstruction of the distal esophagus

- **Eosinophilic esophagitis:** a chronic esophageal disorder characterized by infiltration of the esophageal epithelium by eosinophils, typically in a density >15/hpf

- **Pill esophagitis:** acute injury to esophagus produced by contact with a damaging agent

- **Bezoar:** an accumulation of exogenous matter in the stomach or intestine

- **Peptic ulcers:** deep mucosal lesions that disrupt the muscularis mucosa of the gastric or duodenal wall

- **Stress ulceration:** ulceration in GIT occurring within 24 hr of onset of a critical illness in which physiologic stress is present

- **Diarrhea:** excessive loss of fluid and electrolyte in the stool

  A. **Acute diarrhea:** sudden onset of excessively loose stools of >10 mL/kg/day in infants and >200 g/day in older children, which lasts <14 days

  B. **Chronic/Persistent diarrhea:** stool volume >10 ml/kg/day in toddlers/infants and >200 g/day in older children that

lasts for ≥14 days

C. **Secretory diarrhea:** loose stools occurring when the intestinal epithelial cell solute transport system is in an active state of secretion

D. **Osmotic diarrhea:** diarrhea occurring after ingestion of a poorly absorbed solute

- **Dysentery:** the syndrome of bloody diarrhea with fever, abdominal cramps, rectal pain, and mucoid stools

- **Malrotation of gut:** incomplete rotation of the intestine during fetal development and involves the intestinal nonrotation or incomplete rotation around the superior mesenteric artery

- **Chronic intestinal pseudoobstruction:** a group of disorders characterized as a motility disorder with a primary defect of impaired peristalsis; symptoms are consistent with intestinal obstruction in the absence of mechanical obstruction

- **Constipation:** a delay or difficulty in defecation present for ≥2 wk and significant enough to cause distress to the patient

- **Encopresis:** voluntary or involuntary passage of feces into inappropriate places at least once a mth for 3 consecutive mths once a chronologic or developmental age of 4 yrs has been reached

- **Nonretentive fecal incontinence:** no evidence of fecal retention (impaction), 1 episode per wk in the previous 2 mths in a child at a developmental age >4 yr, defecation in places inappropriate to the social context and no evidence of anatomic, inflammatory, metabolic, endocrine, or neoplastic process that could explain the symptoms

- **Ileus:** the failure of intestinal peristalsis caused by loss of coordinated gut motility without evidence of mechanical obstruction

- **Intussusception:** condition when a portion of the alimentary tract is telescoped into an adjacent segment

- **Gluten sensitivity:** enteric (abdominal pain, bloating, diarrhea) and systemic (headache, fatigue, muscle aches, rash) symptoms after ingesting wheat in the absence of enteropathy or HLA risk factors and autoantibodies

- **Gastroenteritis:** infections of the gastrointestinal tract caused by bacterial, viral, or parasitic pathogens

- **Recurrent abdominal pain in children:** a condition with ≥3 episodes of pain over ≥3 mths that interfered with function

- **Chronic abdominal pain:** long-lasting intermittent or constant abdominal pain that is functional or organic (disease based)

- **Functional abdominal pain:** abdominal pain without demonstrable evidence of pathologic condition, such as anatomic metabolic, infectious, inflammatory or neoplastic disorder

- **Abdominal migraine:** functional abdominal pain with features of migraine (paroxysmal abdominal pain associated with anorexia, nausea, vomiting or pallor as well as maternal history of migraine headaches)
- **Functional abdominal pain syndrome:** functional abdominal pain without the characteristics of dyspepsia, irritable bowel syndrome, or abdominal migraine

- **Anal fissure:** a laceration of the anal mucocutaneous

junction

- **Rectal mucosal prolapse:** the exteriorization of the rectal mucosa through the anus

- **Cirrhosis:** the presence of bands of fibrous tissue that link central and portal areas and form parenchymal nodules

- **Jaundice/Icterus:** yellow discoloration of the sclera, skin, and mucous membranes

- **Pruritus:** intense generalized itching

- **Hepatorenal syndrome:** functional renal failure in patients with end-stage liver disease

- **Neonatal cholestasis:** prolonged elevation of the serum levels of conjugated bilirubin beyond the 1st 14 days of life

- **Cholestasis:** elevated serum conjugated bilirubin levels, resulting from abnormal bile flow at the canalicular and cellular level as a result of hepatocyte damage and

inflammatory mediators

- **Choledochal cysts:** congenital dilatations of the common bile duct that can cause progressive biliary obstruction and biliary cirrhosis

- **Acute hydrops of gall bladder:** acute noncalculous, noninflammatory distention of the gallbladder in the absence of calculi, bacterial infection, or congenital anomalies of the biliary system

- **Portal hypertension:** an elevation of portal pressure >10-12 mm Hg

- **Ascites:** the pathologic accumulation of fluid within the peritoneal cavity

  A. **Chylous ascites:** peritoneal fluid containing lymphatic drainage with a characteristic milky appearance that is rich in triglycerides

- **Peritonitis:** inflammation of the peritoneal lining of the abdominal cavity resulting from infectious, autoimmune, neoplastic, and chemical processes

A. **Primary peritonitis:** bacterial infection of the peritoneal cavity without a demonstrable intraabdominal source

B. **Secondary peritonitis:** peritonitis resulting from entry of enteric bacteria into the peritoneal cavity through a necrotic defect in the wall of the intestines or other viscus as a result of obstruction or infarction or after rupture of an intraabdominal visceral abscess

- **Hernia:** a protrusion of whole or a part of a viscus through the wall that contains it

- **Guarding:** an involuntary reflex contraction of the muscles of the abdominal wall overlying an inflamed viscus and peritoneum

  A. **Rigidity:** generalised guarding

- **Rebound tenderness:** sudden pain experienced when palpating slowly and deeply over a viscus and then releasing the palpating hand

- **Linea alba:** a pale line marking the midline of the abdomen formed by fusion of the aponeurosis of the rectus abdominis

- **Linea nigra:** a black line in the midline of the abdomen below the umbilicus, seen in pregnancy

- **Subcostal line:** a horizontal line drawn across at the level of the lowest point of the 10th rib

- **Transpyloric plane:** an imaginary plane at the level of the lower border of L1 vertebra and the midpoint of the line between the upper end of the xiphisternum and the umbilicus

- **Transtubercular plane:** an imaginary plane at the level of the transverse line joining the tubercles marking the highest point on the iliac crest

# 15. Respiratory System

- **Tidal volume:** the amount of air moved in and out of the lungs during each breath

- **Inspiratory capacity:** the amount of air inspired by maximum inspiratory effort after tidal expiration

- **Expiratory reserve volume:** the amount of air exhaled by maximum expiratory effort after tidal expiration

- **Residual volume:** the volume of gas remaining in the lungs after maximum expiration

- **Vital capacity:** the amount of air moved in and out of the lungs with maximum inspiration and expiration

- **Total lung capacity:** the volume of gas occupying the lungs after maximum inhalation

- **Functional residual capacity:** the amount of air left in the

lungs after tidal expiration

- **Elastance:** the property of a substance to oppose deformation or stretching. It is calculated as a change in pressure (P) divided by change in volume (V)

- **Elastic recoil:** a property of a substance that enables it to return to its original state after it is no longer subjected to pressure

- **Resistance:** the amount of pressure required to generate flow of gas

- **Grunting:** a respiratory sound produced by expiration against a partially closed glottis in an attempt to maintain positive airway pressure during expiration for as long as possible

- **Tachypnea:** increased rate of breathing

- **Eupnea:** normal rate of breathing

- **Closing capacity:** the lung volume at which the dependent airways start to close

- **Dead space ventilation:** air movement in areas that are poorly perfused

- **Crackles/Rales:** high-pitched, interrupted sounds found during inspiration and more rarely during early expiration, which denote opening of previously closed air spaces

- **Wheezes:** musical, continuous sounds usually caused by the development of turbulent flow in narrow airways

- **Sudden infant death syndrome:** the sudden, unexpected death of an infant that is unexplained by a thorough postmortem examination, which includes a complete autopsy, investigation of the scene of death, and review of the medical history

- **Arhinia:** congenital absence of the nose

- **Nasal polyps:** benign pedunculated tumors formed from edematous, usually chronically inflamed nasal mucosa

- **Common cold:** an acute viral infection of the upper respiratory tract in which the symptoms of rhinorrhea and nasal obstruction are prominent

- **Rhinitis medicamentosa:** an apparent rebound effect that causes the sensation of nasal obstruction when the topical adrenergic drug is discontinued

- **Pharyngitis:** inflammation of the pharynx, including erythema, edema, exudates, or an enanthem (ulcers, vesicles)

- **Cough:** a reflex response of the lower respiratory tract to stimulation of irritant or cough receptors in the airways' mucosa

- **Stridor:** a harsh, high-pitched respiratory sound, which is usually inspiratory but can be biphasic and is produced by turbulent airflow

- **Laryngitis/Laryngotracheitis/Laryngotracheobronchitis:** infla--mmation involving the vocal cords and structures inferior to the cords

- **Supraglottitis:** inflammation of the structures superior to the cords (i.e., arytenoids, aryepiglottic folds ["false cords"], epiglottis)

- **Croup:** a heterogeneous group of mainly acute and infectious processes that are characterized by a barklike or brassy cough and may be associated with hoarseness, inspiratory stridor, and respiratory distress

- **Laryngocele:** an abnormal air-filled dilation of the laryngeal saccule that arises vertically between the false vocal cord, the base of the epiglottic and the inner surface of the thyroid cartilage

- **Protracted/Persistent bacterial bronchitis:** a chronic (>3 wk) wet cough, characterized by bacterial counts of 10^4 colony-forming units/mL or greater from bronchoalveolar lavage and resolution of cough within 2 wk of treatment with antimicrobial therapy

- **Plastic bronchitis:** a condition characterized by recurrent episodes of airway obstruction secondary to the formation of large proteinaceous branching casts that take on the shape of and obstruct the tracheobronchial tree

- **Pulmonary emphysema:** distention of air spaces with irreversible disruption of the alveolar septa, involving a part or all of a lung

- **Overinflation:** excessive distention of alveoli with or without alveolar rupture and is often reversible

- **Bronchiolitis obliterans:** a chronic obstructive lung disease of the bronchioles and smaller airways, resulting from an insult to the lower respiratory tract leading to fibrosis of the small airways

- **Follicular bronchitis:** a lymphoproliferative lung disorder characterized by the presence of lymphoid follicles alongside the airways (bronchi or bronchioles) and infiltration of the walls of bronchi and bronchioles

- **Pulmonary agenesis:** the complete absence of a lung

- **Pulmonary aplasia:** the absence of a bronchial stump or carina

- **Bronchobiliary fistula:** a fistulous connection between the

right middle lobe bronchus and the left hepatic ductal system

- **Hypereosinophilic syndrome:** a group of disorders that are characterized by the persistent overproduction of eosinophils accompanied by eosinophil infiltration in multiple organs with end-organ damage from mediator release

- **Hypereosinophilia:** an absolute eosinophil number in the blood ≥1500/cumm eosinophils on 2 separate occasions

- **Pneumonia:** inflammation of the lung parenchyma

- **Recurrent pneumonia:** ≥2 episodes in a single yr or ≥3 episodes ever, with radiographic clearing between occurrences

- **Bronchiectasis:** a condition characterized by irreversible abnormal dilation and anatomic distortion of the bronchial tree and represents a common end stage of a many nonspecific and unrelated antecedent events

- **Atelectasis:** the incomplete expansion or complete collapse of airbearing tissue, resulting from obstruction of air intake into the alveolar sacs

- **Pleurisy:** an inflammation of the pleura

- **Serofibrinous pleurisy:** a fibrinous exudate on the pleural surface and an exudative effusion of serous fluid into the pleural cavity

- **Empyema:** an accumulation of pus in the pleural space

- **Pneumothorax:** the accumulation of extrapulmonary air within the chest, most commonly from leakage of air from within the lung

- **Pneumomediastinum:** presence of air or gas in the mediastinum

- **Hemothorax:** an accumulation of blood in the pleural cavity

- **Chylothorax:** a pleural collection of fluid formed by the

escape of chyle from the thoracic duct or lymphatics into the thoracic cavity

- **Drowning:** death by suffocation after submersion in a liquid medium

  A. **Near drowning:** a patient recovers, at least temporarily from the drowning episode

  B. **Drowning victims:** patients who are initially resuscitated after submersion but who die within 24 hrs

  C. **Secondary drowning:** patients who have recovered completely from a submersion injury and are asymptomatic for a period of time, but who die from respiratory failure secondary to the episode

- **Pectus excavatum/Funnel chest:** midline narrowing of the thoracic cavity

- **Acute lung injury:** a syndrome of diffuse pulmonary inflammation and increased capillary permeability that manifests in acute refractory hypoxemia and lung infiltrates
- **Aerosol:** a biphasic system containing a gaseous phase and a particulate phase

- **Particle:** a body with a defined solid or liquid boundary

bordering its gaseous environment

- **Chest physiotherapy:** a spectrum of physical and mechanical interventions aimed at interacting therapeutically with acute and chronic respiratory disorders

- **Respiratory failure:** the impaired ability of the respiratory system to maintain adequate oxygen and carbon dioxide homeostasis

- **Pulmonary edema:** the accumulation of abnormal amounts of fluids in the extravascular spaces of the lung

- **Acute respiratory distress syndrome:** a type of respiratory failure caused by acute lung inflammation with pulmonary edema resulting from increased permeability

- **Vocal resonance:** sounds heard over various parts of the chest during the act of speech

# 16. The Cardiovascular System

- **Precordium:** the area of the anterior chest wall beneath which the heart lies

- **Cardiac impulse:** the area over which the impact due to cardiac systole is seen

- **Apex beat:** the lowest and most lateral point at which the cardiac impulse can be palpated

- **Arterial pulse:** the abrupt expansion of an artery resulting from the sudden ejection of blood into the aorta and its transmission throughout the arterial system

- **Murmur:** sound produced by turbulent flow within the heart and great vessels, and indicating valve disease

- **Dyspnea:** the subjective sensation of shortness of breath

- **Palpitation:** the subjective sensation of one's own heartbeats

- **Thrills:** the palpable equivalent of murmurs and correlate with the area of maximal auscultatory intensity of the murmur

- **Arrhythmia:** a disturbance in heart rate or rhythm

- **Sinus bradycardia:** a sinus rate <90 beats/min in neonates and <60 beats/min in older children

- **Chaotic or multifocal atrial tachycardia**: atrial tachycardia with ≥3 ectopic P waves, frequent blocked P waves, and varying P-R intervals of conducted beats

- **Accelerated junctional ectopic tachycardia:** an automatic (non-reentry) arrhythmia in which the junctional rate exceeds that of the sinus node and AV dissociation results

- **Atrial flutter/Intraatrial reentrant tachycardia:** an atrial tachycardia characterized by atrial activity at a rate of 250-300 beats/min in children and adolescents, and 400-600 beats/min in neonates

- **Dilated cardiomyopathy:** cardiomyopathy characterized predominantly by left ventricular dilation and decreased left ventricular systolic function

- **Hypertrophic cardiomyopathy:**cardiomyopathy characterized predominantly by increased ventricular myocardial wall thickness, normal or increased systolic function, and often, diastolic (relaxation) abnormalities

- **Restrictive cardiomyopathy:** cardiomyopathy characterized predominantly by nearly normal ventricular chamber size and wall thickness with preserved systolic function, but dramatically impaired diastolic function leading to elevated filling pressures and atrial enlargement

- **Heart failure:** a condition when the heart cannot deliver adequate cardiac output to meet the metabolic needs of the body

- **Hypertension:** average systolic blood pressure and/or diastolic BP that is >95th percentile for age, sex, and height on 3 occasions

- **Prehypertension:** average SBP or diastolic BP that

are >90th percentile but <95th percentile

- **White coat hypertension:** BP in a child >95th percentile in a medical setting but normal BP outside of the office

- **Palpitation:** a subjective feeling of rapid heartbeats

- **Clubbing:** a condition characterized by widening and thickening of the ends of the fingers and toes as well as by convex fingernails and loss of angle between the nail and nail bed

- **Aneurysm:** a dilatation of a localised segment of arterial system

- **Varicose vein:** a dilated and tortuous vein

- **Pulseless cardiac arrest:** the documented cessation of cardiac mechanical activity, determined by the absence of a palpable central pulse, unresponsiveness, and apnea

- **Arterial blood pressure:** the force exerted by the blood against any unit area of the arterial vessel wall

# 17. Diseases of the Blood

- **Hematopoiesis:** the process by which the cellular elements of blood are formed

- **Hemoglobin switching:** sequential expression of different globin genes in RBC precursors

- **Anemia:** a reduction of the hemoglobin concentration or red blood cell volume below the range of values occurring in healthy persons

- **Hemolysis:** the premature destruction of red blood cells (a shortened RBC life span)

- **Priapism:** an unwanted painful erection of the penis

- **Thalassemia:** a group of genetic disorders of globin chain production in which there is an imbalance between the alpha-globin and beta-globin chain production

- **Pancytopenia:** a reduction below normal values of all 3

peripheral blood lineages: leukocytes, platelets, and erythrocytes

- **Inherited/Constitutional pancytopenia:** a decrease in marrow production of the 3 major hematopoietic lineages that occurs on an inherited basis, resulting in anemia, neutropenia, and thrombocytopenia

- **Aplastic anemia:** a condition in which ≥2 blood cell components have become seriously compromised

- **Hemostasis:** the active process that clots blood in areas of blood vessel injury yet simultaneously limits the clot size only to the areas of injury

- **Hypersplenism:** increased splenic function (sequestration or destruction of circulating cells)

- **Lymphangiectasia:** dilation of the lymphatics

- **Lymphangioma circumscriptum:** the presence of many small, superficial lymphangiomas

- **Lymphangiomatosis:** the presence of multiple or disseminated malformations

- **Lymphangioleiomyomatosis:** a condition characterized by proliferation of lymphatic endothelial cells and smooth muscle cells in the lungs, leading to airway and lymphatic obstruction, cyst formation, pneumothorax, and respiratory failure

- **Lymphedema:** a localized swelling caused by impaired lymphatic flow

- **Lymphangitis:** an inflammation of the lymphatics that drain an area of infection

- **Enlarged lymph nodes:** lymph nodes of size >1.5 cm for cervical and axillary, >1 cm for inguinal region

- **Generalized lymphadenopathy:** enlargement of >2 noncontiguous node regions

- **Porphyrias:** acquired or inborn disorders due to abnormalities of specific enzyme mutations in the heme

biosynthetic pathway

# 18. Nephrology

- **Hematuria:** the presence of ≥5 RBC/microlitre of a fresh uncentrifuged midstream urine specimen or >3 RBC/hpf in the centrifuged sediment from 10 ml of freshly voided midstream urine

  A. **Gross hematuria:** >50 RBC/hpf

- **Urethrorrhagia:** urethral bleeding in the absence of urine

- **Benign familial hematuria:** isolated hematuria in multiple family members without renal dysfunction

- **Hemorrhagic cystits:** the presence of sustained hematuria and lower urinary tract symptoms (e.g., dysuria, frequency, urgency) in the absence of other bleeding conditions such as vaginal bleeding, a generalized bleeding condition, or a bacterial urinary tract infection

- **Nephritic syndrome:** a clinical syndrome defined by the association of hematuria, proteinuria, and often hypertension and renal failure

- **Abnormal proteinuria:** excretion of 4-40 mg/m2/hr of proteins in urine

- **Nephrotic-range proteinuria:** excretion of >40 mg/m2/hr of proteins in urine or >3.5 g/24 hr or a urine protein/ creatinine ratio >2

- **Microalbuminuria:** the presence of albumin in the urine above the normal level but below the detectable range of conventional urine dipstick methods

- **Nephrotic syndrome:** a clinical syndrome characterized by edema, massive proteinuria (>40 mg/m2/hr) or a urine protein/creatinine ratio >2, and hypoalbuminemia (<2.5 g/dl)

  A. **Response:** the attainment of remission within the initial 4 wk of corticosteroid therapy

  B. **Remission:** consists of a urine protein/creatinine ratio of <0.2 or <1+ protein on urine dipstick (for 3 consecutive days) or urine proteins (<4 mg/m2/hr) in association with resolution of edema and normalization of serum albumin (>3.5 g/dl)

  C. **Relapse:** a recurrence of massive proteinuria (>40 mg/m2/hr), urine protein/creatinine ratio of >2 or 3+ protein on urine dipstick testing (for 3 consecutive days),

most often in association with edema

D. **Steroid sensitive:** patients enter remission in response to corticosteroid treatment alone

E. **Steroid resistance:** the failure to achieve remission after 8 wk of corticosteroid therapy

F. **Steroid dependent:** patients respond to initial corticosteroid treatment by entering complete remission but develop a relapse either while still receiving steroids or within 2 wks of discontinuation of treatment following a steroid taper

G. **Frequent relapsing:** relapses occurring ≥4 times in a 12 mths period

- **IgA nephropathy:** a glomerular disease characterised by the presence of IgA deposits prevalent over other classes of immunoglobulins

- **Rapildly progressive glomerulonephritis:** a clinical syndrome characterized by a rapid loss of renal function (>50% decrease in GFR) over days to wks

- **Crescents:** the presence of ≥2 layers of cells in Bowman's space

- **Asymptomatic bacteriuria:** a condition in which there is a positive urine culture without any manifestations of infection

- **Ureterocele:** a cystic dilation of the terminal ureter

- **Ectopic ureter:** ureter that drains outside the bladder

- **Nocturnal enuresis:** the occurrence of involuntary voiding at night after 5 yr, the age when volitional control of micturition is expected

  A. **Primary enuresis:** nocturnal urinary control never achieved

  B. **Secondary enuresis:** the child was dry at night for at least a few mths and then enuresis developed

- **Varicocele:** a congenital condition in which there is abnormal dilation of the pampiniform plexus in the scrotum

- **Spermatocele:** a cystic lesion that contains sperm and is attached to the upper pole of the sexually mature testis

- **Hydrocele:** an accumulation of fluid in the tunica vaginalis

- **Functional urinary incontinence:** the involuntary loss of urine as a result of a failure of control of the bladder-sphincteric unit

- **Stress incontinence:** the involuntary leakage of urine occurring when the intravesical pressure exceeds the bladder outlet or urethral resistance in the absence of measurable detrusor contraction

- **Giggle incontinence:** involuntary and typically unpredictable wetting during or immediately after giggling or laughing

- **Postvoiding dribbling:** the involuntary leakage of urine immediately after voiding has finished in a toilet trained child

- **Dysfunctional voiding:** incomplete relaxation or involuntary intermittent contractions of the pelvic floor muscle during voiding in neurologically intact children
- **Staccato voiding:** interrupted voiding that is caused by periodic bursts of pelvic floor muscle activities during

voiding

- **Fractionated voiding:** several small, discontinuous voids that results from poor and unsustained detrusor contractions characterized by infrequent and incomplete emptying

- **Vesicoureteral reflux:** the retrograde flow of urine from the bladder to the kidneys

- **Obstructive uropathy:** a restriction of urine flow which, if left uncorrected, will lead to progressive renal deterioration or hamper normal renal development

- **Ureteropelvic junction obstruction:** an impedance in urine flow from the renal pelvis to the proximal ureter

- **Oxalosis:** extrarenal deposition of calcium oxalate

- **Urolithiasis:** calculi formed in the kidney that may be found anywhere in the urinary tract as well as primary bladder stones

- **Nephrocalcinosis:** calcium deposition within the renal tissue

# 19. Gynecologic Problems of Childhood

- **Clitoromegaly in newborn:** clitoral width >6 mm
- **Amastia:** complete absence of the breast
- **Polymastia:** presence of supernumerary breast tissue
- **Polythelia:** presence of accessory nipples
- **Primary amenorrhoea:** absence of menses within 4 yrs of onset of puberty
- **Secondary amenorrhoea:** absence of menses for the length of 3 previous cycles in a postmenarchal patient
- **Dysmenorrhea:** painful uterine cramps that precede and accompany menses

  A. **Primary dysmenorrhea:** characterized by the absence of any specific pelvic pathologic condition

  B. **Secondary dysmenorrhea:** due to underlying pathology such as anatomic abnormality, or infection such as pelvic inflammatory disease

- **Endometriosis:** a condition in which implants of endometrial tissue are found outside the uterus, most commonly near the fallopian tubes and ovaries

- **Rape:** penetration of any genital, oral, or anal orifice by a part of the assailant's body or any object

  A. **Acquaintance rape:** by a person known to the victim

  B. **Date rape:** by a person dating the victim (is often drug facilitated)

  C. **Male rape:** same-sex rape of male teens by other males

  D. **Gang rape:** group of young men rape a solitary female victim

  E. **Statutory rape:** sexual activity between an adult and an adolescent under the age of legal consent, as defined by individual state law

  F. **Stranger rape:** by a stranger to the victim

- **Hirsutism:** abnormally increased terminal (mature, heavy, dark) hair growth in areas of the body where hair growth is normally androgen dependent

- **Hydrocolpos:** an accumulation of mucus or

nonsanguineous fluid in the vagina

- **Hemihematometra:** atretic segment of vagina with menstrual fluid accumulation

- **Hydrosalpinx:** an accumulation of serous fluid in the fallopian tube, often an end result of pyosalpinx

- **Didelphic uterus:** 2 cervices, each associated with 1 uterine horn

- **Bicornuate uterus:** 1 cervix associated with 2 uterine horns

- **Unicornuate uterus:** result of failure of 1 Müllerian duct to descend

# 20. The Endocrine System

- **Hypothyroidism:** the deficient production of thyroid hormone, either from a defect in the gland itself (primary hypothyroidism) or a result of reduced thyroid-stimulating hormone (TSH) stimulation (central or hypopituitary hypothyroidism)

- **Goiter/Thyromegaly:** an enlargement of the thyroid gland

- **Hyperthyroidism:** excessive secretion of thyroid hormone

- **Gynecomastia:** the proliferation of mammary glandular tissue in the male

- **Primary ovarian insufficiency/Hypergonadotropic hypogona--dism/Premature ovarian failure:** a condition characterized by the arrest of normal ovarian function before the age of 40 yr

- **Disorder of sex development:** a condition in which development of chromosomal, gonadal, or anatomical sex is atypical

- **Bilateral anorchia/Vanishing testes syndrome:** a condition when testes are absent, but the male phenotype is complete

- **Hypopituitarism:** underproduction of growth hormone alone or in combination with deficiencies of other pituitary hormones

- **Precocious puberty:** the onset of secondary sexual characteristics before the age of 8 yr in girls and 9 yr in boys

- **Central precocious puberty:** the onset of breast development before the age of 8 yr in girls and by the onset of testicular development (volume 4 mL) before the age of 9 yr in boys, as a result of the early activation of the hypothalamic-pituitary-gonadal axis

- **Premature adrenarche:** the appearance of sexual hair before the age of 8 yr in girls or 9 yr in boys without other evidence of maturation

# 21. The Nervous System

- **Consciousness:** the spontaneously occurring state of awareness of self and environment

- **Clouding of consciousness:** the minimal reduction of wakefulness or awareness wherein the main difficulty is attention or vigilance

- **Confusion:** the state of impaired ability to think and reason clearly at a developmentally and intellectually appropriate level

- **Obtundation:** mild to moderate alertness reduction with decreased interest in the environment and slower than normal reactivity to stimulation

- **Stupor:** a state of unresponsiveness with little or no spontaneous movement resembling deep sleep from which the patient can only be aroused by vigorous and repeated stimulation

- **Coma:** a state of deep, unarousable, sustained pathologic unconsciousness with the eyes closed the results from dysfunction of the ascending reticular activating system in the brainstem or in both cerebral hemispheres

- **Vegetative state:** a condition of complete unawareness of the self and the environment accompanied by sleep-wake cycles with either complete or partial preservation of hypothalamic or brainstem autonomic functions

- **Minimally conscious state:** patients who were in coma or a vegetative state and who are beginning to demonstrate minimal signs of awareness

- **Locked-in syndrome:** a condition in which patients retain consciousness and cognition but are unable to move or communicate because of severe paralysis

- **Akinetic mutism:** a condition consisting of pathologically slowed or nearly absent bodily movement accompanied by a similar loss of speech

- **Sedation:** a medically induced state that is on a continuum between the fully alert, awake state and general anesthesia

A. **Deep sedation:** a state of unarousability to voice and is accompanied by suppression of reflex responses

B. **Conscious sedation:** a condition in which a patient is sleepy, comfortable, and cooperative but maintains airway -protective and ventilatory reflexes

- **Concussion:** a traumatically induced transient disturbance of brain function that involves a complex pathophysiologic process which may be caused either by a direct blow to the head, face, neck, or elsewhere on the body with an "impulsive" force transmitted to the head, whether these are linear or rotational forces

- **Lucid interval:** a period of clinical improvement after an initial loss of consciousness, followed by deterioration within mins to hrs

- **Cerebral reperfusion injury:** a complex series of interactions between the brain parenchyma and microcirculation that results in detrimental effects that negate some of the benefits of reperfusion

- **Atrophy of muscle:** decreased muscle bulk

- **Hypertrophy of muscle:** increased muscle bulk

- **Pseudohypertrophy of muscle:** muscle tissue that has been replaced by fat and connective tissue, giving it a bulky appearance with a paradoxical reduction in strength

- **Paresis:** feeble power of contraction of a muscle group due to weakness of the muscle

- **Paralysis:** loss of power of a muscle group resulting in absence of contraction of those muscle

- **Muscle tone:** an unconscious, continuous, partial contraction of muscle, creating resistance to passive movement of a joint

  A. **Passive tone:** range of motion around a joint

  B. **Active tone:** physiologic resistance to movement

- **Spasticity:** a condition characterized by an initial resistance to passive movement

- **Rigidity:** a condition characterized by resistance to passive

movement that is equal in the flexors and extensors regardless of the velocity of movement. It is felt throughout the passive movement

- **Opisthotonos:** severe hyperextension of the spine caused by hypertonia of the paraspinal muscles

- **Hypotonia:** abnormally diminished tone

- **Fasciculations:** small, involuntary muscle contractions that result from the spontaneous discharge of a single motor unit

- **Ataxia:** a disturbance in the smooth performance of voluntary motor acts

- **Abasia:** unsteadiness of stance

- **Asynergia:** decomposition of complex movements into isolated , successive parts

- **Dysmetria:** errors in judging distance

- **Rebound:** inability to inhibit a muscular action

- **Dysdiadochokinesia:** impaired performance of rapid alternating movements

- **Titubation:** a bobbing of the head predominantly in the anteroposterior dimension

- **Electroencephalogram (EEG):** a continuous recording of electrical activity between reference electrodes placed on the scalp

- **Lissencephaly/Agyria:** a disorder that is characterized by the absence of cerebral convolutions and a poorly formed sylvian fissure

- **Schizencephaly:** the presence of unilateral or bilateral clefts within the cerebral hemispheres owing to an abnormality of morphogenesis

- **Polymicrogyria:** a condition characterized by an augmentation of small convolutions separated by shallow enlarged sulci

- **Porencephaly:** the presence of cysts or cavities within the brain that result from developmental defects or acquired lesions, including infarction of tissue

- **Colpocephaly:** an abnormal enlargement of the occipital horns of the ventricular system

- **Holoprosencephaly:** a developmental disorder of the brain that results from defective formation of the prosencephalon and inadequate induction of forebrain structures

- **Microcephaly:** a head circumference that measures >3 SD below the mean for age and sex

- **Hydrocephalus:** a diverse group of conditions that result from impaired circulation and/or absorption of CSF or, in rare circumstances, from increased production of CSF by a choroid plexus papilloma

- **Megalencephaly:** an anatomic disorder of brain growth with brain weight/volume ratio >98th percentile for age (or 2 SD above the mean)

- **Macrocephaly:** an occipitofrontal circumference >98th percentile

- **Hydranencephaly:** condition when cerebral hemispheres are absent or represented by membranous sacs with remnants of frontal, temporal, or occipital cortex dispersed over the membrane

- **Craniosynostosis:** premature closure of the cranial sutures

  A. **Primary craniosynostosis:** due to closure of ≥1 sutures owing to abnormalities of skull development

  B. **Secondary craniosynostosis:** due to failure of brain growth and expansion

- **Scaphocephaly:** premature closure of the sagittal suture producing a long and narrow skull

- **Frontal plagiocephaly:** premature fusion of a coronal and sphenofrontal suture characterized by unilateral flattening of the forehead, elevation of the ipsilateral orbit and eyebrow, and a prominent ear on the corresponding side

- **Occipital plagiocephaly:** flattening of the occiput

- **Trigonocephaly:** premature fusion of the metopic suture characterised by a keel-shaped forehead and hypotelorism

- **Turricephaly:** a cone-shaped head from premature fusion of the coronal, and often sphenofrontal and frontoethmoidal sutures

- **Plagiocephaly:** a condition in which head shape is asymmetric in the sagittal or coronal plane that can result from asymmetry in suture closure or from asymmetry of brain growth

- **Deformational/Positional plagiocephaly:** the development of cranial flattening and asymmetry in the infant as a result of extrinsic molding forces placed on the skull, such as consistently sleeping on the same area of the head

- **Brachycephaly:** a condition in which head shape is shortened from front to back along the sagittal plane; the back of the skull and face are flatter than normal

- **Scaphocephaly/Dolicocephaly:** a condition in which the head is elongated from front to back in the sagittal plane

- **Muscular torticollis:** a continuous tightening of muscles in the neck preventing passive rotation

- **Tummy time:** infant's awake time spent lying on their stomach

- **Seizure:** a transient occurrence of signs and/or symptoms resulting from abnormal excessive or synchronous neuronal activity in the brain

  A. **Tonic seizures:** characterized by increased tone or rigidity

  B. **Atonic seizures:** characterized by flaccidity or lack of movement during a convulsion

  C. **Clonic seizures:** rhythmic fast muscle contractions and slightly longer relaxations

  D. **Myoclonus:** a "shock-like" contraction of a muscle of <50 msec that is often repeated

  E. **Astatic seizures:** a very momentary loss of tone with a sudden fall

- **Epilepsy:** a disorder of the brain characterized by an enduring predisposition to generate seizures and by the neurobiologic, cognitive, psychologic, and social consequences of this condition

- **Epileptic syndrome:** a disorder that manifests ≥1 specific seizure types and has a specific age of onset and a specific prognosis

- **Epileptic encephalopathy:** an epilepsy syndrome in which there is a severe EEG abnormality which is thought to result in cognitive and other impairments in the patient

- **Genetic epilepsy:** epilepsy syndrome which is the direct result of a known or presumed genetic defect(s) in which the genetic defect is not causative of a brain structural or metabolic disorder other than the epilepsy

- **Febrile seizures:** seizures that occur between the age of 6 and 60 mths with a temperature ≥38°C (100.4°F), that are not the result of central nervous system infection or any metabolic imbalance, and that occur in the absence of a history of prior afebrile seizures

- **Vaccine encephalopathy:** seizures and psychomotor regression occurring after vaccination and presumed to be caused by it

- **Aura:** sensory experience reported by the patient; not observed externally

- **Head drop:** loss of tone or myoclonus in only the neck muscles resulting in a milder seizure

- **Epileptic spasms/Axial spasms:** flexion or extension of truncal and extremity musculature that is sustained for 1 -2 sec, shorter than what is seen in tonic seizures, which last >2 sec

- **Epilepsia partialis continua:** focal motor clonic and/or myoclonic seizures that persist for days, mths, or even longer

- **Absence seizures:** generalized seizures consisting of staring, unresponsiveness, and eye flutter lasting usually for few secs

  A. **Typical absences:** associated with 3 Hz

spike–and–slow-wave discharges

B. **Atypical absences:** associated with 1-2 Hz spike–and–slow-wave discharges, and with head atonia and myoclonus during the seizures

C. **Juvenile absences:** associated with 4-5 Hz spike-and-slow waves and occur in juvenile myoclonic epilepsy

- **Automatisms:** automatic semipurposeful movements of the mouth (oral, alimentary such as chewing) or of the extremities (manual, such as manipulating the sheets; leg automatisms such as shuffling, walking)

- **Epileptogenesis:** the mechanism through which the brain, or part of it, turns epileptic

- **Kindling:** an animal model for human focal epilepsy in which repeated electrical stimulation of selected areas of the brain with a low-intensity current initially causes no apparent changes but with repeated stimulation results in epilepsy

- **Spasms:** sudden generalized jerks lasting 1-2 sec that are usually associated with a single, very brief, generalized discharge

- **Jitteriness:** rapid motor activities, such as a tremor or shake, that can be ended by flexion or holding the limb

- **Status epilepticus:** a condition resulting either from failure of the mechanism responsible for seizure termination or from the initiation of mechanisms which lead to abnormally prolonged seizures (after time point t1). It is a condition that can have long term consequences (after time pont t2), including neuronal death, neuronal injury, and alteration of neuronal networks, depending on the type and duration of seizures

  t1 (treatment started):

  tonic clonic= 5 min;

  focal with impaired consciousness= 10 min;

  absence= 15 min.

  t2 (consequences expected):

  tonic clonic= 30 min;

  focal with impaired consciousness >60 min

  absence= unknown

  A. **Prodromal/Incipient/Impending status epilepticus:** first 5 min

  B. **Early status epilepticus:** 5-10 min

C. **Established status epilepticus:** > 30 min

D. **Refractory status epilepticus:** persistent seizures despite 2-3 appropriate doses of antiepileptics

- **Anoxic seizures:** generalised convulsions due to syncope causing drop attacks

- **Shuddering attacks:** rapid tremor of the head, shoulder, and trunk, lasting a few secs, often associated with eating, and recurring many times a day

- **Hyperekplexia/Stiff baby syndrome:** a disorder with neonatal onset of life-threatening episodes of tonic stiffening that precipitate apnea and convulsive hypoxic seizures

- **Motor tics:** movements that are under partial control, and are associated with an urge to do them and with a subsequent relief

- **Stereotypies:** repetitive movements that are more complex than tics and do not change and wax and wane like tics (e.g., head banging, head rolling, body rocking, and hand clapping),

usually occur in neurologically impaired children

- **Mannerism:** a pattern of socially acceptable, situational behavior that is seen in particular situations such as gesturing when talking

- **Fussiness:** a state of irritability that is not easily explained by a cause, such as tiredness, hunger, teething or pain from an injury

- **Migraine:** recurrent headache characterized by episodic attacks that may be moderate to severe in intensity, focal in location on the head, have a throbbing quality, and may be associated with nausea, vomiting, light sensitivity, and sound sensitivity

- **Status migrainosus:** migraine lasting for >72 hrs

- **Movement disorders:** abnormal or excessive involuntary movements that may result in abnormalities in posture, tone, balance, or fine motor control

- **Tremor:** oscillating, rhythmic movements about a fixed

point, axis, or plane

A. **Rest tremor:** maximal when the affected body part is inactive and supported against gravity

B. **Postural tremor:** notable when the patient sustains a position against gravity

C. **Action tremor:** seen with performance of a voluntary activity and can be subclassied into simple kinetic tremor, which occurs with limb movement, and intention tremor, which occurs as the patient's limb approaches a target

D. **Essential tremor:** characterized by a slowly progressive, bilateral, 4-9 Hz postural tremor that involves the upper extremities and occurs in the absence of other known causes of tremor

E. **Holmes/Midbrain/Rubral tremor:** characterized by a slow frequency, high amplitude tremor that is present at rest and with intention

- **Dystonia:** intermittent and sustained involuntary muscles contractions that produce abnormal postures and movements of different parts of the body, often with a twisting quality

- **Chorea:** involuntary, continual, irregular movements or

movement fragments with variable rate and direction that occur unpredictably and randomly

- **Ballism:** involuntary, high amplitude, flinging movements typically occurring proximally

- **Athetosis:** slow, writhing, continuous, involuntary movements

- **Posture:** position of the different body parts adopted in relation to one another and in relation to surrounding

- **Decubitus:** position adopted during lying down

- **Gait:** position adopted while walking

- **Encephalopathy:** a generalized disorder of cerebral function that may be acute or chronic, progressive, or static

- **Cerebral palsy:** a group of permanent disorders of movement and posture causing activity limitation, that

are attributed to nonprogressive disturbances in the in the developing fetal or infant brain

- **Spastic diplegia:** bilateral spasticity of the legs that is greater than in the arms

- **Spastic quadriplegia:** marked motor impairment of all extremities

- **Acquired demyelinating disorders of the central nervous system:** conditions resulting from neurologic dysfunction caused by immune-mediated attacks on white matter insulating the brain, optic nerves and spinal cord

- **Multiple sclerosis:** a chronic demyelinating disorder of the brain, spinal cord, and optic nerves characterized by a relapsing–remitting course of neurologic events without encephalopathy separated in time and space

- **Neuromyelitis opica/Devic disease:** a demyelinating disorder characterized by monophasic or polyphasic episodes of optic neuritis and/or transverse myelitis

- **Acute disseminated encephalomyelitis:** an initial inflammatory, demyelinating event with multifocal neurologic deficits, typically accompanied by encephalopathy

- **Idiopathic intracranial hypertension/Pseudotumor cerebri:** a clinical syndrome that mimics brain tumors and is characterized by increased intracranial pressure ≥280 mm Hg in sedated or obese children; ≥250 mm Hg in nonobese, nonsedated children with a normal cerebrospinal fluid cell count and protein content and normal to slightly decreased ventricular size, and normal ventricular anatomy and position documented by MRI

- **Diastematomyelia:** a form of occult dysraphism in which the spinal cord is divided into 2 halves

- **Syringomyelia:** a cystic distention of the spinal cord caused by obstruction of the flow of spinal fluid from within the spinal cord to its point of absorption

- **Syringobulbia:** a slowly progressive cavity formation within the medulla oblongata, associated with gliosis

- **Hydromyelia:** a congenital deformity resulting from distension of the central canal of spinal cord, which in turn compresses the surrounding cord structures, including the anterior horn cells

- **Transverse myelitis:** a condition characterized by rapid development of both motor and sensory deficits at any level of the spinal cord with evidence of spinal cord inflammation by an MRI-documented enhancing lesion, or CSF pleocytosis (>10 cells), or increased immunoglobulin G index

- **Neuromuscular disease:** disorders of the motor unit and excludes influences on muscular function from the brain, such as spasticity

- **Benign congenital hypotonia:** nonprogressive hypotonia of unknown origin

- **Muscular dystrophy:** a condition which is a primary myopathy, has a genetic basis, the course is progressive, and degeneration and death of muscle fibers occur at some stage

- **Inflammatory myopathies:** a heterogenous group of

disorders characterised pathologically by inflammation in skeletal muscle with resulting muscle fibre damage and subsequent clinical weakness

- **Myotonia:** a very slow relaxation of muscle after contraction, regardless of whether that contraction was voluntary or was induced by a stretch reflex or electrical stimulation

- **Myotonia congenita/Thomsen disease:** a channelopathy characterized by weakness and generalized muscular hypertrophy

- **Paramyotonia:** a temperature-related myotonia that is aggravated by cold and alleviated by warm external temperatures

- **Myasthenia gravis:** a chronic autoimmune disease of neuromuscular blockade, characterized clinically by rapid fatigability of striated muscle, particularly extraocular and palpebral muscles and those of swallowing

- **Myasthenic crisis:** an acute or subacute severe increase in weakness in patients with myasthenia gravis, usually

precipitated by an intercurrent infection, surgery, or even emotional stress

- **Bell palsy:** an acute unilateral peripheral facial nerve palsy that is not associated with other cranial neuropathies or brainstem dysfunction

- **Paraesthesia:** altered sensations felt in the form of pins and needles, tingling and numbness etc.

- **Hyperaesthesia:** hypersensitive skin to normal stimuli

- **Hypoaesthesia:** decreased feeling of sensation

- **Anaesthesia:** total loss of sensation of the affected part

- **Pain:** a complex sensation triggered by actual or potential tissue damage and influenced by cognitive, behavioral, emotional, social, and cultural factors

  A. **Somatic pain:** pain resulting from injury to or inflammation of tissues (skin, muscle, tendons, bone, joints, fascia, vasculature, etc.)

B. **Visceral pain:** pain resulting from injury to or inflammation of viscera

C. **Neuropathic pain:** pain resulting from injury to, inflammation of, or dysfunction of the peripheral or central nervous systems

D. **Chronic pain:** recurrent or persistent pain lasting longer than the normal tissue healing time, approximately 3-6 mths

- **Tenderness:** pain which occurs in response to a stimulus given by somebody

- **Root pain:** spontaneous severe pain due to disease process involving the root fibres

- **Referred pain:** a pain arising from a visceral organ projected to a definite site on the surface of the body, away from anatomical situation of the organ

- **Complex regional pain syndrome:** a condition characterized by ongoing burning limb pain that is subsequent to an injury, immobilization, or another noxious event affecting

the extremity

- **Dermatome:** the area of skin innervated by a single spinal segment

- **Allodynia:** a heightened pain response to normally non-noxious stimuli

- **Hyperalgesia:** exaggerated pain reactivity to noxious stimuli

- **Guillain-Barré syndrome:** an acute demyelinating disease of the peripheral nervous system characterized clinically by acute flaccid paralysis

- **Breath holding:** voluntary holding of breath by a child in prolonged inspiration

- **Bacterial meningitis:** an inflammation of the leptomeninges triggered by bacteria in the subarachnoid space

- **Meningismus:** tense neck and back muscles as a reflex to

avoid painful extension of inflamed meninges

- **Stroke:** the sudden occlusion or rupture of cerebral arteries or veins resulting in focal cerebral damage and clinical neurologic deficits

- **Bland infarct:** infarct without any visible hemorrhage

- **Muscle twitch:** a transient contraction of the muscle fibres of a motor unit by a single action potential of the neuron

- **Topagnosis:** the ability to localize the area of contact of a tactile stimulus

- **Stereognosis:** recognition of familiar objects by touch

- **Graphesthesia:** the ability to recognize numbers, letters, or other readily identifiable symbols traced on the skin

- **Two-point discrimination:** the ability to distinguish between closely approximated stimulation at two points

- **Ageusia:** a complete loss of gustatory function

- **Dysgeusia:** distortion in the perceived qualities of a taste stimulus

- **Phantogeusia:** the experience of a taste sensation in the apparent absence of a gustatory stimulus

- **Anosmia:** the complete absence of olfactory functioning

- **Specific anosmia:** a deficit in perception of only a specific odorous compound

- **Hyperosmia:** an increased sensitivity to smell

- **Dysosmia/Parosmia:** distortions in the perceived qualities of the odor stimulus in the presence of an odor

- **Phantosmia:** perception of an odor when there is no odor present

- **Mental retardation:** a disability characterised by a significant limitation both in intellectual functioning and in adaptive behaviour as expressed in conceptual, social, practical, and adaptive skills

- **Aphasia:** disorder of speech due to injury to higher centres

  A. **Sensory aphasia:** inability to understand the spoken words or written words

  B. **Motor aphasia:** inability to express thoughts by writing or speaking

- **Agraphia:** selective loss to writing ability, spontaneous or dictation

# 22. Disorders of the Eye

- **Refraction:** the focusing power of the eye

- **Tonometry:** the method of assessing intraocular pressure

- **Emmetropia:** the state in which parallel rays of light come to focus on the retina with the eye at rest (nonaccommodating)

- **Hyperopia/Far sightedness:** parallel rays of light come to focus posterior to the retina with the eye in a neutral state

- **Myopia/Near sightedness:** parallel rays of light come to focus anterior to the retina with the eye in a neutral state

- **Astigmatism:** the refractive powers of the various meridians of the eye differ

- **Anisometropia:** refractive state of one eye is significantly

different from the refractive state of the other eye

- **Amblyopia:** decrease in visual acuity, unilateral or bilateral, that occurs in visually immature children as a result of a lack of a clear image projecting onto the retina

  A. **Strabismic amblyopia:** the unformed retinal image may occur secondary to a deviated eye

  B. **Anisometropic amblyopia:** an unequal need for vision correction between the eyes

  C. **Ametropic amblyopia:** a high refractive error in both eyes

  D. **Deprivation amblyopia:** a media opacity within the visual axis

- **Amaurosis:** partial or total loss of vision

- **Nyctalopia/Night blindness:** vision that is defective in reduced illumination

- **Coloboma of iris:** developmental defect may present as a defect in a sector of the iris, a hole in the substance of the iris, or a notch in the pupillary margin

- **Microcoria/Congenital miosis:** a small pupil that does not react to light or accommodation and that dilates poorly, if at all, with medication

- **Congenital mydriasis:** the pupils appear dilated, do not constrict significantly to light or near gaze, and respond minimally to miotic agents

- **Dyscoria:** abnormal shape of the pupil

- **Corectopia:** abnormal pupillary position

- **Anisocoria:** inequality of the pupils

- **Tonic pupil:** typically a large pupil that reacts poorly to light (the reaction may be very slow or essentially nil), reacts poorly and slowly to accommodation, and redilates in a slow, tonic manner

- **Heterochromia iridum:** the 2 irides are of different color

- **Heterochromia iridis:** a portion of an iris differs in color

from the remainder

- **Strabismus/Squint:** misalignment of the eye

- **Orthophoria:** the ideal condition of exact ocular balance. It implies that the oculomotor apparatus is in perfect equilibrium so that the eyes remain coordinated and aligned in all positions of gaze and at all distances

- **Heterophoria:** a latent tendency for the eyes to deviate which is normally controlled by fusional mechanisms that provide binocular vision or avoid diplopia

- **Heterotropia:** a misalignment of the eyes that is constant. It occurs because of an inability of the fusional mechanism to control the deviation

- **Accommodative esotropia:** convergent deviation of the eyes associated with activation of the accommodative (focusing) reflex

- **Nystagmus:** rhythmic oscillations of 1 or both eyes

A. **Latent nystagmus:** conjugate jerk nystagmus toward viewing eye

B. **Manifest latent nystagmus:** fast jerk to viewing eye

C. **Periodic alternating:** cycles of horizontal or horizontal-rotary that change direction

D. **Seesaw nystagmus:** one eye rises and intorts as other eye falls and extorts

E. **Nystagmus retractorius:** eyes jerk back into orbit or toward each other

F. **Gaze-evoked nystagmus:** jerk nystagmus in direction of gaze

G. **Gaze-paretic nystagmus:** eyes jerk back to maintain eccentric gaze

H. **Downbeat nystagmus:** fast phase beating downward

I. **Upbeat nystagmus:** fast phase beating upward

J. **Vestibular nystagmus:** horizontal-torsional or horizontal jerks

K. **Asymmetric or monocular nystagmus:** pendular vertical nystagmus

L. **Spasmus nutans:** fine, rapid, pendular nystagmus

- **Opsoclonus:** multidirectional conjugate movements of varying rate and amplitude

- **Ocular dysmetria:** overshoot of eyes on rapid fixation

- **Ocular flutter:** horizontal oscillations with forward gaze and sometimes with blinking

- **Ocular bobbing:** downward jerk from primary gaze, remains for a few sec, then drifts back

- **Ocular myoclonus:** rhythmic to-and-fro pendular oscillations of the eyes, with synchronous nonocular muscle movement

- **Ptosis:** drooping of upper eyelid below the normal level

- **Epicanthal folds:** vertical or oblique folds of skin extending on either side of the bridge of the nose from the brow or lid area, covering the inner canthal region

- **Lagophthalmos:** a condition in which complete closure of the lids over the globe is difficult or impossible

- **Ectropion:** eversion of the lid margin

- **Entropion:** inversion of the lid margin

- **Epiblepharon:** a roll of skin beneath the lower eyelid lashes causing the lashes to be directed vertically and to touch the cornea

- **Trichiasis:** inward turning of the eyelashes

- **Blepharospasm:** spastic or repetitive closure of the eyelids

- **Blepharitis:** inflammation of the lid margins characterized by erythema and crusting or scaling

- **Internal hordeolum:** acute bacterial infection of the Meibomian glands

- **External hordeolum/Stye:** acute bacterial infection of the Zeis/ Moll glands

- **Chalazion:** granulomatous inflammation of a Meibomian gland characterized by a firm, nontender nodule in the

upper or lower lid

- **Coloboma of the eyelid:** cleft-like deformity may vary from a small indentation or notch of the free margin of the lid to a large defect involving almost the entire lid

- **Dacryocystocele/Mucocele:** an unusual presentation of a nonpatent nasolacrimal sac that is obstructed both proximally and distally

- **Alacrima/Dry eye:** a wide spectrum of disorders with reduced or absent tear secretion

- **Pinguecula:** a yellowish-white, slightly elevated mass on the bulbar conjunctiva, usually in the interpalpebral region

- **Pterygium:** a fleshy triangular conjunctival lesion that may encroach on the cornea

- **Symblepharon:** a cicatricial adhesion between the conjunctiva of the lid and the globe

- **Megalocornia:** nonprogressive symmetric condition characterized by an enlarged cornea (>12 mm in diameter) and an anterior segment in which there is no evidence of previous or concurrent ocular hypertension

- **Microcornea/Anterior microphthalmia:** an abnormally small cornea in an otherwise relatively normal eye

- **Keratoconus:** progressive thinning and bulging of the central cornea, which becomes cone shaped

- **Peters anomaly:** a central corneal opacity (leukoma) that is present at birth

- **Hypopyon:** pus in the anterior chamber

- **Phlyctenules:** small, yellowish, slightly elevated lesions usually located at the corneal limbus; they may encroach on the cornea and extend centrally

- **Interstitial keratitis:** nonulcerative inflammation of the corneal stroma

- **Cataract:** any opacity of the lens

- **Microspherophakia:** a small, round lens

- **Anterior lenticonus:** bilateral condition in which the anterior capsule of the lens thins, allowing the lens to bulge forward centrally

- **Posterior lenticonus:** a condition characterized by a circumscribed round or oval bulge of the posterior lens capsule and cortex, involving the central region of the lens

- **Uveitis:** inflammation of the uveal tract (the inner vascular coat of the eye, consisting of the iris, ciliary body, and choroid)

- **Choroiditis:** inflammation of the posterior portion of the uveal tract

- **Panophthalmitis:** inflammation involving all parts of the eye

- **Sympathetic ophthalmia:** an inflammatory response that affects the uninjured eye after a perforating injury

- **Retinitis pigmentosa:** progressive retinal degeneration characterized by pigmentary changes, arteriolar attenuation, usually some degree of optic atrophy, and progressive impairment of visual function

- **Retinal detachment:** a separation of the outer layers of the retina from the underlying retinal pigment epithelium

- **Hypoplasia of the optic nerve:** a nonprogressive condition characterized by a subnormal number of optic nerve axons with normal mesodermal elements and glial supporting tissue

- **Morning glory disc anomaly:** congenital malformation of the optic nerve characterized by an enlarged, excavated, funnel-shaped disc with an elevated rim, resembling a morning glory flower

- **Tilted disc:** congenital anomaly in which the vertical axis of the optic disc is directed obliquely, so that the upper temporal portion of the nerve head is more prominent

and anterior to the lower nasal portion of the disc

- **Papilledema:** swelling of the nerve head secondary to increased intracranial pressure

- **Optic neuritis:** inflammation or demyelinization of the optic nerve with attendant impairment of function

- **Optic atrophy:** degeneration of optic nerve axons, with attendant loss of function

- **Glaucoma:** damage to the optic nerve with visual field loss that is caused by or related to elevated pressure within the eye

  A. **Primary glaucoma:** the cause is an isolated anomaly of the drainage apparatus of the eye (trabecular meshwork)

  B. **Secondary glaucoma:** other ocular or systemic abnormalities are associated

- **Hypertelorism:** wide separation of the eyes or an increased interorbital distance

- **Hypotelorism:** narrowness of the interorbital distance

- **Exophthalmos/Proptosis:** protrusion of the eye

- **Enophthalmos:** posterior displacement or sinking of the eye back into the orbit

- **Dacryoadenitis:** inflammation of the lacrimal gland

- **Dacryocystitis:** infection of the lacrimal sac

- **Periorbital/Preseptal cellulitis:** inflammation of the lids and periorbital tissues without signs of true orbital involvement (such as proptosis or limitation of eye movement)

- **Orbital cellulitis:** condition involving inflammation of the tissues of the orbit, with proptosis, limitation of movement of the eye, edema of the conjunctiva (chemosis), and inflammation and swelling of the eyelids with potentially decreased visual acuity

- **Hyphema:** the presence of blood in the anterior chamber of the eye

- **Open globe:** penetrating, perforating, or blunt injury resulting in compromise of the cornea or sclera of the eye

- **Diplopia:** perception of two images when only one object is present

# 23. The Ear

- **Dizziness:** a sensation of altered orientation in space

- **Vertigo:** a type of dizziness associated with any illusion or sensation of motion

- **Congenital cholesteatoma:** a nonneoplastic, destructive, cystic lesion that usually appears as a white, round, cyst -like structure medial to an intact tympanic membrane

- **Conductive hearing loss:** hearing loss resulting from pathologic conditions in the external ear canal, tympanic membrane, or middle ear

- **Sensorineural hearing loss:** hearing loss due to damage or malfunction of cochlea or the auditory nerve

- **Exostoses:** benign hyperplasia of the perichondrium and underlying bone

# 24. The Skin

- **Macule:** an alteration in skin color but cannot be felt, <1 cm size
- **Patch:** an alteration in skin color but cannot be felt, >1 cm size
- **Papules:** palpable solid lesions <1 cm
- **Plaques:** palpable lesions >1 cm in size and have a flat surface
- **Nodules:** palpable lesions >1 cm with a rounded surface
- **Tumour/Neoplasm:** a growth of new cells which proliferate independent of the need of the body
- **Vesicles:** raised, fluid-filled lesions <1 cm in diameter
- **Bullae:** raised, fluid-filled lesions >1 cm in diameter

- **Pustules:** lesions containing purulent material

- **Wheals:** flat-topped, palpable lesions of variable size, duration, and configuration that represent dermal collections of edema fluid

- **Cysts:** circumscribed, thick-walled lesions covered by a normal epidermis and contain fluid or semisolid material

- **Scales:** compressed layers of stratum corneum cells that are retained on the skin surface

- **Purpura:** lesions on skin as the result of bleeding into the skin and have a red-purple color; they may be flat or palpable

- **Petechiae:** small purpura <2-3 mm

- **Erosions:** focal loss of the epidermis which heals without scarring

- **Ulcer:** a break in the continuity of the covering epithelium

(skin or mucous membrane), extending into the dermis and tend to heal with scarring

- **Excoriations:** ulcerated lesions inflicted by scratching, often linear or angular in configuration

- **Fissures:** lesions caused by splitting or cracking

- **Crusts:** lesions consisting of matted, retained accumulations of blood, serum, pus, and epithelial debris on the surface of a weeping lesion

- **Scars:** end-stage lesions that can be thin, depressed, and atrophic; raised and hypertrophic; or flat and pliable

- **Lichenification:** a thickening of skin with accentuation of normal skin lines that is caused by chronic irritation (rubbing, scratching) or inflammation

- **Sebaceous hyperplasia:** min, profuse, yellow-white papules that are frequently found on the forehead, nose, upper lip, and cheeks of a term infant; representing hyperplastic sebaceous glands

- **Milia:** superficial epidermal inclusion cysts that contain laminated keratinized material

- **Sucking blisters:** solitary or scattered superficial bullae present at birth on the upper limbs of infants at birth are presumably induced by vigorous sucking on the affected part in utero

- **Cutis marmorata/Livido reticularis:** an evanescent, lacy, reticulated red and/or blue cutaneous vascular pattern appearing over most of the body surface, when a newborn infant is exposed to low environmental temperatures

- **Nevus simplex:** a small, pale pink, ill-defined, vascular macule that occurs most commonly on the glabella, eyelids, upper lip, and nuchal area

- **Dermal melanocytosis/Mongolian spots:** blue or slate-gray macular lesions, with variably defined margin

- **Erythema toxicum:** benign, self-limited, evanescent eruption

- **Eosinophilic pustular folliculitis:** recurrent crops of pruritic, coalescing, follicular papulopustules on the face, trunk, and extremities

- **Skin dimples:** cutaneous depressions over bony prominences and in the acral area, at times associated with pits and creases

- **Aplasia cutis congenita/Congenital absence of skin:** developmental absence of skin usually noted on the scalp as multiple or solitary (70%), noninflammatory, well -demarcated, oval or circular 1-2 cm ulcers

- **Ectodermal dysplasia:** a heterogeneous group of disorders characterized by a constellation of findings involving defects of ≥2 of the following: teeth, skin, and appendageal structures including hair, nails, and eccrine and sebaceous glands

- **Angiokeratomas:** lesions characterized by ectasia of superficial lymphatic vessels and capillaries with hyperkeratosis of the overlying epidermis

- **Spider angioma/Vascular spider/Nevus araneus:** lesion

with a central feeder artery with many dilated radiating vessels and a surrounding erythematous flush, varying from a few millimeters to several centimeters in diameter

- **Nevus skin lesions:** lesions characterized histopathologica--lly by collections of well-differentiated cell types normally found in the skin

- **Melanocytic nevus:** a benign cluster of melanocytic nevus cells that arises as a result of alteration and proliferation of melanocytes at the epidermal–dermal junction

- **Ephelides:** are light or dark brown, round, oval or irregularly shaped, well-demarcated, macules usually <3 mm in diameter that occur in sun-exposed areas such as the face, upper back, arms, and hand

- **Lentigines:** are small (<3 cm), round, dark brown macules that can appear anywhere on the body with an early age of onset

- **Café-au-lait spots:** uniformly hyperpigmented, sharply

demarcated macular lesions, the hues of which vary with the normal degree of pigmentation of the individual: they are tan or light brown in white individuals and may be dark brown in black children

- **Congenital oculocutaneous albinism:** partial or complete failure of melanin production in the skin, hair, and eyes despite the presence of normal number, structure, and distribution of melanocytes

- **Piebaldism:** condition characterized by sharply demarcated amelanotic patches that occur most frequently on the forehead, anterior scalp (producing a white forelock), ventral trunk, elbows, and knees

- **Vitiligo:** macular depigmentation associated with the destruction of melanocytes

- **Eczematous skin disorders:** a broad group of cutaneous eruptions characterized by erythema, edema, and pruritus

- **Diaper dermatitis:** any rash in the diaper region

- **Photosensitivity:** a qualitatively or quantitatively abnormal cutaneous reaction to sunlight or artificial light because of UV radiation

- **Psoriasis:** an inflammatory autoimmune-related disease characterized by inflammation and keratinocyte proliferation

- **Keratosis pilaris:** a common papular eruption resulting from keratin plugging of hair follicles

- **Acanthosis nigricans:** lesions characterized by symmetric, hyperpigmented, velvety, hyperkeratotic plaques with exaggerated skin lines in intertriginous areas

- **Keloid:** a sharply demarcated, benign, dense growth of connective tissue that forms in the dermis after trauma

- **Hyperhidrosis:** excessive sweating beyond what is physiologically necessary for temperature control

- **Miliaria:** retention of sweat in occluded eccrine sweat ducts

- **Hypertrichosis:** excessive hair growth at inappropriate locations

- **Hypotrichosis:** deficient hair growth

- **Trichotillomania:** visible hair loss attributable to pulling; mounting tension preceding hair pulling; gratification or release of tension after hair pulling; and absence of hair pulling attributable to hallucinations, delusions, or an inflammatory skin condition

- **Anonychia:** absence of the nail plate, usually a result of a congenital disorder or trauma

- **Koilonychia:** flattening and concavity of the nail plate with loss of normal contour, producing a spoon-shaped nail

- **Leukonychia:** a white opacity of the nail plate that may involve the entire plate or may be punctate or striate

- **Onycholysis:** separation of the nail plate from the distal nail bed

- **Beau lines:** transverse grooves in the nail plate that represent a temporary disruption of formation of the nail plate

- **Trachyonychia:** characterized by longitudinal ridging, pitting, fragility, thinning, distal notching, and opalescent discoloration of all the nails

- **Ingrown nail:** condition when the lateral edge of the nail, including spicules that have separated from the nail plate, penetrates the soft tissue of the lateral nail fold

- **Angular cheilitis/Perlèche:** inflammation and fissuring at the corners of the mouth, often with associated erosion, maceration, and crusting

- **Fordyce spots/granules:** asymptomatic, 1-3 mm, yellow white macules and papules on the vermilion lips and buccal mucosa

- **Epstein pearls/Gingival cysts of the newborn:** white, keratin-containing epidermal inclusion cysts on the palatal or alveolar mucosa

- **Geographic tongue/Benign migratory glossitis:** consists of single or multiple sharply demarcated, irregular, smooth red patches surrounded by an elevated yellowish white serpiginous border on the dorsum of the tongue

- **Cellulitis:** infection and inflammation of loose connective tissue, with limited involvement of the dermis and relative sparing of the epidermis

- **Necrotizing fasciitis:** a subcutaneous tissue infection that involves the deep layer of supercial fascia but may spare adjacent epidermis, deep fascia, and muscle

- **Blistering distal dactylitis:** a superficial blistering infection of the volar fat pad on the distal portion of the finger or thumb

- **Tinea corporis:** infection of the glabrous skin, excluding the palms, soles, and groin

- **Lump:** a vague mass of body tissue

- **Swelling:** any enlargement or protuberance in the body

- **Haemangioma:** a vascular malformation or hamartoma arising from a capillary, vein or artery

- **Abscess:** a collection of pus

- **Boil/Furuncle:** an infection of a hair follicle

- **Papilloma:** a simple overgrowth of all layers of the skin

- **Fibroma:** a tumour of the fibrous tissue

- **Lipoma:** an overactive cluster of fat cells producing a palpable swelling

- **Sebaceous cyst:** a cyst of the sebaceous gland due to blockage of the duct of this gland which opens mostly into the hair follicle

- **Callosity:** a raised thickened patch of hyperkeratosis commonly seen in areas of the body which undergo excessive wear and tear and repeated minor traumas

- **Sinus:** a blind track leading from the surface down to the tissues

- **Fistula:** a communicating track between two epithelial surfaces, commonly between a hollow viscus and the skin or between two hollow viscera

- **Bullous impetigo:** skin infection characterized by flaccid, transparent bullae usually <3 cm in diameter on previously untraumatized skin

- **Perianal dermatitis/cellulitis/streptococcal disease:** a distinct clinical entity characterized by well demarcated, perianal erythema associated with anal pruritus, painful defecation, and occasionally blood-streaked stools

# 25. Bone and Joint Disorders

- **Contractures:** a loss of mobility of a joint from congenital or acquired causes and are caused by periarticular soft-tissue fibrosis or involvement of muscles crossing the joint

- **Valgus:** bone deformity with apex towards midline

- **Varus:** bone deformity with apex away from midline

- **Range of motion of joints**

  A. **Abduction:** away from the midline

  B. **Adduction:** toward the midline

  C. **Flexion:** movement of bending from the starting position

  D. **Extension:** movement from bending to the starting position

  E. **Supination:** rotating the forearm to face the palm upward

  F. **Pronation:** rotating the forearm to face the palm downward

  G. **Inversion:** turning the hindfoot inward

  H. **Eversion:** turning the hindfoot outward

I. **Plantarflexion:** pointing the toes away from the body (toward the floor)

J. **Dorsiflexion:** pointing the toes toward the body (toward the ceiling)

K. **Internal rotation:** turning inward toward the axis of the body

L. **External rotation:** turning outward away from the axis of the body

- **Foot**

  A. **Forefoot:** toes and metatarsals

  B. **Midfoot:** cuneiforms, navicular, cuboid

  C. **Hindfoot:** talus and calcaneus

- **Hip subluxation:** partial contact between the femoral head and acetabulum

- **Hip dislocation:** a hip with no contact between the articulating surfaces of the hip

- **Legg-Calvé-Perthes disease:** a hip disorder that results from temporary interruption of the blood supply to the

proximal femoral epiphysis, leading to osteonecrosis and femoral head deformity

- **Slipped capital femoral epiphysis:** failure of the physis and displacement of the femoral head relative to the neck

- **Scoliosis:** a complex 3-dimensional spinal deformity that is defined in the coronal plane as a curve of ≥10°, measured by the Cobb method, on a posteroanterior radiograph of the spine

  A. **Congenital scoliosis:** a spinal deformity that results from abnormal development of the bony spinal column

- **Hyperkyphosis:** thoracic kyphosis in excess of the normal range of values (>50° measured from T3-T12)

- **Lordosis:** an increased anterior curvature of lumbar spine

- **Spondylolysis:** a defect in the pars interarticularis, the segment of bone connecting the superior and inferior articular facets in the vertebra

- **Spondylolisthesis:** a forward slippage of 1 vertebra on another

- **Spondylitis:** inflammation of the vertebrae

- **Syndactyly:** failure of the individual digits to separate during development

- **Brachydactyly:** a condition of having short digits

- **Camptodactyly:** permanent flexion of one or more fingers associated with missing inner phalangeal creases indicating lack of finger movement from before 8 wk of gestation

- **Clinodactyly:** a medial or lateral curving of the fingers; usually refers to incurving of the 5th finger

- **Postaxial polydactyly:** extra finger or toe present on the lateral side of the hand or foot

- **Preaxial polydactyly:** extra finger or toe present on the medial side of the hand or foot

- **Ectrodactyly:** a partial or total absence of the distal segments of a hand or foot, with the proximal segments of the limbs more or less normal

- **Arachnodactyly:** unusually long, spider-like digits

- **Brushfield spots:** speckled white rings (about 23) at a distance to the periphery of the iris of the eye

- **Hypoplastic nail:** an unusually small nail on a digit

- **Low-set ears:** this designation is made when the helix meets the cranium at a level below a horizontal plane that is an extension of a line through both inner canthi

- **Melia:** suffix meaning “limb” (e.g., amelia—missing limb; brachymelia—short limb)

- **Posterior parietal hair whorl:** a single whorl occurs to the right or left of midline and within 2 cm anterior to the posterior fontanel representing the focal point from which the posterior scalp skin was under growth tension during brain growth between the 10th and 16th wk of fetal

development

- **Prominent lateral palatine ridges:** relative overgrowth of the lateral palatine ridges secondary to a deficit of tongue thrust into the hard palate

- **Shawl scrotum:** the scrotal skin joins around the superior aspect of the penis and represents a mild deficit in full migration of the labial-scrotal folds

- **Short palpebral fissures:** decreased horizontal distance of the eyelid folds based on measurement from the inner to the outer canthus

- **Synophrys:** eyebrows that meet in the midline

- **Telecanthus:** lateral displacement of the inner canthi. The intercanthal distance (ICD) is increased, but the interpupillary distance (IPD) is normal

- **Widow's peak:** V-shaped midline, downward projection of the scalp hair in the frontal region representing an upper forehead intersection of the bilateral fields of periocular hair

growth suppression

- **Arthrogryposis multiplex congenita:** a heterogeneous group of muscular, neurologic, and connective tissue anomalies that present with ≥2 joint contractures at birth as well as muscle weakness

- **Sprain:** an injury to a ligament or joint capsule

- **Strain:** an injury to a muscle or tendon

- **Contusion:** a crush injury to any soft tissue

- **Overuse injuries:** injuries caused by repetitive microtrauma that exceeds the body's rate of repair

- **Felon/Pulp space infection:** a subcutaneous infection of the terminal segment of a digit

- **Dactylitis:** infection of the phalanges or metacarpals

- **Acheiria:** congenital absence of an entire hand

- **Acheiropodia:** congenital absence of both hands and feet

- **Ankylosis:** a stiffness of a joint

- **Rocker bottom feet:** feet with prominent heel and a loss of the normal concave longitudinal arch of the sole

- **Osteoporosis:** fragility of the skeletal system and a susceptibility to fractures of the long bones or vertebral compressions from mild or inconsequential trauma

- **Marfan syndrome:** an inherited, systemic, connective tissue disorder caused by mutations in the gene encoding the extracellular matrix protein fibrillin-1

- **Hyperphosphatasia:** excessive elevation of the bone isoenzyme of alkaline phosphatase in serum and significant growth failure

- **Arthritis:** intraarticular swelling or the presence of ≥2 of

the following signs: limitation in range of motion, tenderness or pain on motion, and warmth

A. **Oligoarthritis:** arthritis involving 4 joints within the 1st 6 mths of disease onset, and often only a single joint is involved

B. **Polyarthritis:** arthritis characterized by inflammation of 5 joints in both upper and lower extremities during 1st 6 mths of disease

C. **Psoriatic arthritis:** arthritis and psoriasis or arthritis and ≥2 of the following: (1) dactylitis, (2) nail pitting or onycholysis, or (3) psoriasis in a 1st-degree relative

- **Orthosis:** a device that is applied to the surface of the body to maintain alignment or position, to prevent or assist movement of the body part, or to provide support

- **Prosthesis:** a device that replaces a missing body part, such as an arm or a leg

- **Rheumatic disease:** chronic, multisystem diseases characterized by an unpredictable course with periods of exacerbation and remission

# 26. Environmental Health Hazards

- **Heat syncope:** fainting after prolonged exercise attributed to poor vasomotor tone and depleted intravascular volume, and it responds to fluids, cooling, and supine positioning

- **Heat edema:** mild edema of the hands and feet during initial exposure to heat which resolves with acclimatization

- **Heat tetany:** carpopedal tingling or spasms caused by heat-related hyperventilation which responds to moving to a cooler environment and decreasing respiratory rate (or rebreathing by breathing into a bag)

- **Heat exhaustion:** a moderate illness with core temperature 37.7-39.4°C (100-103°F)