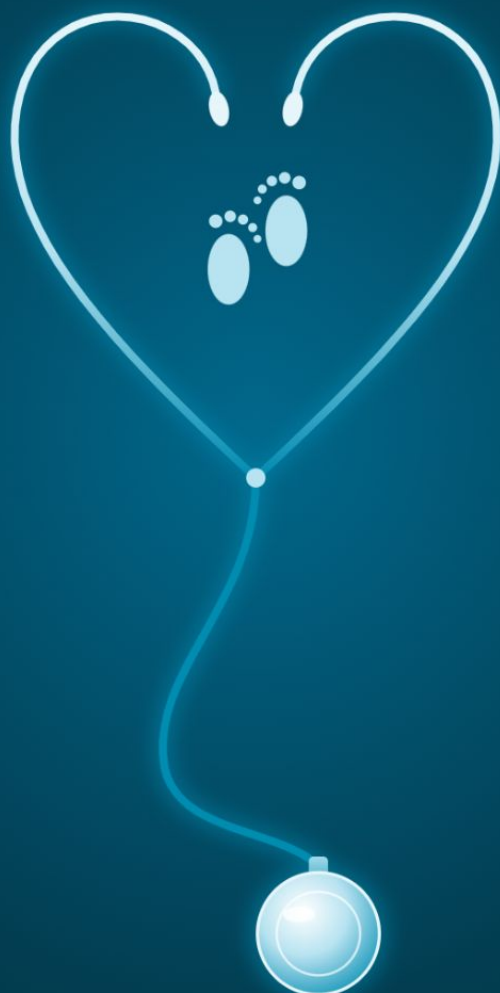




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Volume 1 & Volume 2 | 39 Chapters | Pages 1 – 505

Contents at a Glance

VOLUME 1 Pages 1 – 256		VOLUME 2 Pages 257 – 505	
1. Growth, Development & Puberty	1 – 6	26. Pediatric Endocrinology (Part 1)	257 – 267
2. Pediatric Nutrition	7 – 17	27. Pediatric Endocrinology (Part 2)	268 – 280
3. Fluid, Electrolyte & Acid-Base Disorders	18 – 31	28. Cyanotic Heart Disease	281 – 294
4. Pediatric Behavioural, Psychiatric & Learning Disorders	32 – 42	29. Acyanotic Congenital Heart Disease	295 – 316
5. Infectious Diseases	43 – 55	30. Respiratory System	317 – 331
6. Pediatric Metabolic Disorders	56 – 78	31. Systemic Cardiology	332 – 365
7. Pediatric Genetics & Inborn Errors of Metabolism	79 – 104	32. Pediatric Gastroenterology	366 – 393
8. Perinatology	105 – 117	33. Pediatric Hepatology	394 – 417
9. General Neonatology	118 – 128	34. Pediatric Hematology	418 – 431
10. Systematic Neonatology	129 – 139	35. Pediatric Oncology	432 – 458
11. Neonatal Infectious Diseases & Antimicrobials	140 – 144	36. Pediatric Emergencies & Critical Care	459 – 473
12. Neonatal Hematological Disorders	145 – 149	37. Pediatric Dermatology	474 – 490
13. Neonatal Neurological Disorders	150 – 154	38. Pediatric Otology	491 – 498
14. Neonatal Respiratory Disorders	155 – 158	39. Pediatric Ophthalmology	499 – 505
15. Neonatal Cardiovascular Disorders	159 – 163		
16. Neonatal Renal Disorders	164 – 168		
17. Neonatal Endocrine Disorders	169 – 174		
18. Neonatal Gastrointestinal & Liver Disorders	175 – 180		
19. Miscellaneous Neonatology	181 – 191		
20. Rheumatology	192 – 202		
21. Immunology	203 – 210		
22. Pediatric Neurology	211 – 223		
23. Neuromuscular Disorders	224 – 230		
24. Pediatric Nephrology	231 – 242		
25. Pediatric Urology	243 – 256		

VOLUME 1 | Pages 1 – 256

CHAPTER 1 | Growth, Development & Puberty

Pages 1 – 6

1. Basics: Growth Axis & Scammon's Curves	1
• Scammon's curves, growth failure, weight / length / HC norms	1
2. Workup: Short Stature & Delayed Puberty	2
3. Comparison Tables	2
• Table 1: Short stature etiologies	2
• Table 2: Patterns of milestone abnormalities; Table 3: Tanner staging	3
4. Developmental Milestones	4
• Gross motor, fine motor, language, social; Goodenough Draw-A-Man	4
5. Puberty	5
• Girls, boys, Prader orchidometer	5
6. High-Yield Clinical Entities	5
• Enuresis (enuresis alarm)	5
7. Visual Anchors	6
• Scaphocephaly, Prader orchidometer	6

CHAPTER 2 | Pediatric Nutrition

Pages 7 – 17

1. Infant Feeding, Growth Standards & Requirements	7
• The big picture; breastfeeding vs breast milk jaundice	7
• Infant formulas & milks; complementary feeding	8
2. Severe Acute Malnutrition (SAM) & Refeeding Syndrome	9
• Marasmus vs kwashiorkor	9
• WHO 10-step management; refeeding syndrome	10
3. Fat-Soluble Vitamins (A, D, E, K)	11
• Vitamin D & rickets	11
• Working up rickets (Ca, Pi, PTH, 25-OH-D table)	12
• Vitamin K, vitamin E	13
4. Water-Soluble Vitamins	13
• Cobalamin (B12) & folate (B9); vitamin C: scurvy	14
5. Trace Minerals & Micronutrients	15
• Zinc, copper & selenium	15
6. Visual Anchors	16
• Vitamin A, rickets, pellagra	16
• Scurvy (radiographic), malnutrition & zinc	17

CHAPTER 3 | Fluid, Electrolyte & Acid-Base Disorders

Pages 18 – 31

1. Fluid Compartments & Osmolality	18
• Total body water through childhood; ICF vs ECF	18
• Osmolality & tonicity, osmolal gap; workup: low sodium	19

2. Hypernatremia & Hyponatremia	20
• Why the brain suffers; hypernatremia causes	20
• Hyponatremia by volume status	21
• Table 1: Primary hyponatremic etiologies (SIADH vs salt wasting)	22
3. Potassium, Magnesium & Phosphate	22
• Hyperkalemia	23
• Hypokalemia, magnesium, refeeding hypophosphatemia	24
• Table 2: Potassium & divalent cation test distractors	25
4. Acid-Base	25
• Metabolic acidosis (anion gap), metabolic alkalosis (urine chloride)	26
• Table 3: Primary acid-base disturbances & expected compensation	27
5. Maintenance, Deficit & Replacement Fluids	27
• Maintenance: Holliday-Segar (100-50-20)	27
• Dehydration grading	28
• Deficit & ongoing losses; replacement fluids	29
6. Visual Anchors	30
• Hyperkalemia ECG progression	30
• Hypokalemia ECG, hypernatremic brain injury, formulas to know cold	31

CHAPTER 4 | Pediatric Behavioural, Psychiatric & Learning Disorders

Pages 32 – 42

1. Basics: Circuits & Sorting the Presentation	32
2. Workup: Suspected ADHD	33
• Comparison table: learning & communication spectrum	33
3. Neurodevelopmental Disorders	34
• ADHD, autism spectrum disorder (ASD)	34
• Structural malformations & global delay	35
4. Learning & Communication Disorders	35
• Dyslexia	35
• Dyscalculia, dysgraphia, speech / fluency disorders	36
5. Disruptive & Conduct Disorders	37
• ODD, conduct disorder, DMDD, IED	37
6. Mood, Anxiety, Somatic & Eating Disorders	38
• MDD, separation anxiety, OCD, PANDAS, somatic & eating disorders	38
7. Paroxysmal Events, Motor Disorders & Psychoses	39
• Breath-holding spells	39
• Tics, stereotypy, catatonia	40
8. Visual Anchors	41
• Dyslexia fMRI, corpus callosum agenesis, ADHD MRI	41
• Bulimia, NSSI	42

CHAPTER 5 | Infectious Diseases

Pages 43 – 55

1. Fever Evaluation & Febrile Infant	43
• Febrile infant (0–60 days)	43
• Inflammatory marker cutoffs; febrile neutropenia	44

2. Exanthems, Soft Tissue & Toxin-Mediated Infections	45
• Toxin-mediated & systemic rashes	45
• Viral exanthems	46
3. Invasive Bacterial Disease, Meningitis & Sepsis	48
• Acute bacterial meningitis	48
• Meningococemia & asplenia	49
4. Congenital & Perinatal Infections (TORCH)	50
5. Deep Neck, Mycobacterial & Vector-Borne	51
• Deep neck space infections (RPA vs PTA), TB & NTM, vector-borne & zoonotic	51
• Post-COVID MIS-C (PIMS)	52
6. Visual Anchors	53
• Skin & mucosa	53
• Lab & smear	54
• Radiology & late signs	55

CHAPTER 6 | Pediatric Metabolic Disorders

Pages 56 – 78

1. Approach & Emergency Evaluation	56
• Three functional groups, bedside sorting	56
• Workup (in order); urine odours	57
2. Amino Acidopathies & Organic Acidemias	58
• PKU & BH4 deficiency	58
• Tyrosinemias, alkaptonuria & Hartnup	59
• MSUD; organic acidemias; biotin defects	60
• Table 1: Amino acid & organic acid disorders	61
3. Urea Cycle Disorders & Hyperammonemia	62
• Specific defects (NAGS, CPS1, OTC, ASS, ASL, HHH); acute hyperammonemia	63
4. Carbohydrate Disorders	64
• Hepatic GSDs	64
• Muscle GSDs; galactose & fructose disorders	65
• Table 2: Carbohydrate & glycogen disorders	67
5. Fatty Acid Oxidation & Mitochondrial Disorders	68
• MCAD deficiency	68
• Long-chain defects & MADD; PDHC deficiency, Leigh, MELAS, MERRF	69
6. Lysosomal & Peroxisomal Disorders	70
• Sphingolipidoses: Gaucher, Tay-Sachs, Fabry	70
• MPS & mucopolipidoses	71
• Peroxisomal disorders: RCDP, X-ALD, Refsum	72
• Table 3: Lysosomal & peroxisomal storage diseases	73
7. Purine & Pyrimidine Disorders	74
• Lesch-Nyhan syndrome, hereditary orotic aciduria	74
8. Visual Anchors	75
• Gaucher cells, cherry-red spot, Erlenmeyer flask	75
• Krabbe, Pompe	76
• Wolman, Refsum, Lesch-Nyhan	77
• PMM2-CDG, Zellweger	78

CHAPTER 7 | Pediatric Genetics & Inborn Errors of Metabolism

Pages 79 – 104

1. Cytogenetics & Chromosome Errors	79
• Karyotyping; chromosomal disorders	79
• Translocations & other structural changes	80
2. Inheritance Patterns	81
• Autosomal dominant	81
• Autosomal recessive, X-linked	82
• Y-linked, pseudoautosomal; triplet repeat disorders & anticipation	83
• Imprinting & UPD; digenic & mitochondrial	84
3. Genetic Testing	85
• When & ethics; which test	85
• Table 1: Testing technologies	86
4. Dysmorphology & Chromosomal Syndromes	87
• Four kinds of defect; Down syndrome, Edwards	87
• Patau, trisomy 8 mosaicism	88
• Turner, Klinefelter	89
• Table 2: High-yield microdeletion profiles	90
• Chromosome breakage syndromes	91
5. Inborn Errors of Metabolism: Screening & Emergencies	91
• Newborn screening, the sick neonate, urine odours	92
6. Amino Acid, Organic Acid & Urea Cycle Disorders	93
• PKU, tyrosinemia, alkaptonuria, MSUD	93
• Propionic & methylmalonic acidemia; urea cycle disorders	94
• Table 3: Differential diagnosis of urea cycle defects	95
7. Lysosomal, Peroxisomal & Glycogen Storage Disorders	96
• Sphingolipidoses	96
• Mucopolysaccharidoses	97
• Peroxisomal disorders; glycogen storage diseases	98
• Table 4: Major glycogen storage diseases	99
8. Visual Anchors	100
• Cytogenetics; faces & clinical gestalt	100
• Angelman, Prader-Willi, cri du chat	101
• Wolf-Hirschhorn, Rubinstein-Taybi, Gaucher biopsy	102
• Pedigrees	103

CHAPTER 8 | Perinatology

Pages 105 – 117

1. Basics: Fetal Response to Placental Insufficiency	105
2. Workup: Reduced Fetal Movements	106
3. Comparison Tables	107
• Table 1: Antepartum surveillance; Table 2: FGR vs SGA	107
• Table 3: Monochorionic twin complications; Table 4: RBC alloimmunization	108
• Table 5: In utero fetal surgical interventions	109
4. Antepartum Surveillance	110
5. Intrapartum Monitoring & PPRM	110
• CST, BPP, decelerations, pathological CTG	110

6. FGR & Preeclampsia	111
7. Monochorionic Twins	112
• TTTS, TRAP, chorionicity	112
• Quintero staging, Solomon technique	113
8. Fetal Anemia, Alloimmunization & Hydrops	114
• HDFN, Kleihauer-Betke	114
9. Prenatal Screening & Fetal Surgery	115
• First-trimester screening, MSAFP, MOMS, severe CDH, EXIT	115
10. Visual Anchors	116
• Lambda / twin-peak sign	116

CHAPTER 9 | General Neonatology

Pages 118 – 128

1. Basics: Transition, Resuscitation & Exam	118
2. Workup: Respiratory Distress	119
3. Comparison Tables	119
• Table 1: Apgar score; Table 2: Benign oral & facial lesions	119
• Table 3: Neonatal respiratory disorders	120
• Table 4: Sarnat staging of HIE; Table 5: TORCH differentiators	121
4. Respiratory Disorders	122
• RDS, TTN	122
• PPHN	123
• CDH & EXIT	124
5. Cardiovascular & Hematologic	124
• Hyperbilirubinemia & kernicterus	125
• Vitamin K deficiency bleeding (HDN)	126
6. GI & Metabolic Emergencies	126
• NEC	126
• Infant of diabetic mother & hypoglycemia	127
7. HIE	127
• Therapeutic hypothermia, MRI / MRS	127
8. Visual Anchors	128
• RDS, CDH, CMV	128

CHAPTER 10 | Systematic Neonatology

Pages 129 – 139

1. Basics: Transition, Oxygen & Temperature	129
2. Workup: Delivery Room Resuscitation	130
3. Comparison Tables	131
• Table 1: Neonatal respiratory pathology	131
• Table 2: GI & surgical pathologies; Table 3: Sarnat staging	132
• Table 4: Neonatal neurological insults	133
4. Respiratory	133
• RDS, TTN, MAS, PPHN	133

5. Cardiovascular	134
• Apnea of prematurity, PDA, cyanosis workup	134
• TGA, TOF, arrhythmias	135
6. GI, Surgical & Metabolic	135
• NEC, meconium ileus, IHPS, IDM	135
7. Neurology	136
• HIE, IVH, PVL, myelomeningocele, NAS / Nows	136
8. Jaundice, Hematology & Infection	137
9. Visual Anchors	138
• Neonatal tetanus, transient neonatal MG, RDS, CDH	138
• TGA, IVH, NEC	139

CHAPTER 11 | Neonatal Infectious Diseases & Antimicrobials

Pages 140 – 144

1. Basics & Workup	140
• EOS vs LOS, TORCH triad, CMV, syphilis	140
2. Comparison Tables	141
• Table 1: EOS vs LOS	141
• Table 2: Congenital infection matrix	142
3. Antimicrobials	143
4. Visual Anchors	144

CHAPTER 12 | Neonatal Hematological Disorders

Pages 145 – 149

1. Basics & Workup: Bleeding / Thrombocytopenia	145
• Developmental hemostasis, VKDB, polycythemia	145
2. Comparison Tables	146
• Table 1: Neonatal bleeding disorders	146
• Table 2: Blood component transfusion thresholds	147
3. Polycythemia & Test Traps	148
4. Visual Anchors	149
• Blueberry muffin	149

CHAPTER 13 | Neonatal Neurological Disorders

Pages 150 – 154

1. Basics & Workup: Neonatal Seizures	150
• Term HIE, preterm PVL	150
2. Comparison Tables	151
• Table 1: HIE staging (Sarnat vs Levene)	151
• Table 2: Preterm brain lesions	152
3. IVH Surveillance & Hydrocephalus; Hypothermia & Seizure Traps	153
4. Visual Anchors	154

CHAPTER 14 | Neonatal Respiratory Disorders

Pages 155 – 158

1. Basics & Workup	155
-------------------------------	------------

2. Comparison Table: Major Neonatal Respiratory Conditions	156
3. Management	157
• RDS, TTN, MAS, CDH, PPHN	157
4. Visual Anchors	158

CHAPTER 15 Neonatal Cardiovascular Disorders	Pages 159 – 163
1. Basics & Workup: Cyanosis & CCHD Screen	159
2. Comparison Tables	160
• Table 1: Critical congenital heart diseases	160
• Table 2: Pharmacotherapy & inotropes	161
3. Test Traps	162
4. Visual Anchors	163

CHAPTER 16 Neonatal Renal Disorders	Pages 164 – 168
1. Basics & Workup: Neonatal AKI	164
• PUV / LUTO, RVT, renovascular HTN	164
• Antenatal hydronephrosis	165
2. Comparison Tables	165
• Table 1: Prerenal azotemia vs intrinsic AKI	165
• Table 2: Neonatal renal & urologic pathologies	166
3. Management	167
4. Visual Anchors	168
• Keyhole sign, bunch of grapes	168

CHAPTER 17 Neonatal Endocrine Disorders	Pages 169 – 174
1. Basics & Workup: Hypoglycemia	169
• Congenital hypothyroidism, CAH, hypocalcemia	169
• DSD	170
2. Comparison Tables	171
• Table 1: CAH enzymatic defects	171
• Table 2: Neonatal calcium & bone disorders	172
3. Test Traps	173
4. Visual Anchors	174

CHAPTER 18 Neonatal Gastrointestinal & Liver Disorders	Pages 175 – 180
1. Basics	175
• EA / TEF, CDH, malrotation, duodenal atresia, NEC, Hirschsprung, biliary atresia	175
2. Workup: Neonatal Obstruction / Vomiting; Cholestasis	176
3. Comparison Tables	177
• Table 1: Neonatal bowel obstructions	177
• Table 2: Gastroschisis vs omphalocele	178

4. Test Traps	179
5. Visual Anchors	180
CHAPTER 19 Miscellaneous Neonatology	Pages 181 – 191
1. Basics: Developmental Disruption vs Benign Artifact	181
• Workup: seizures after delivery	181
2. Comparison Tables	182
• Table 1: Neonatal scalp & head swellings; Table 2: Fetal thoracic lesions	182
• Table 3: Fetal GU & abdominal defect interventions	183
3. Dermatologic, Morphologic & GU Anomalies	184
• Prune-belly, Beckwith-Wiedemann	184
• Single umbilical artery, TAR syndrome	185
4. Hypoglycemia, IEM & Teratogens	186
5. Trauma, Head Swellings & Hearing	187
6. Fetal Interventions	188
• SCT, CPAM	188
• LUTO / PUV, renal anhydramnios, amniotic band syndrome	189
7. Visual Anchors	190
CHAPTER 20 Rheumatology	Pages 192 – 202
1. Basics: Immune Dysregulation & Hallmark Syndromes	192
2. Workup	193
3. Comparison Tables	194
• Table 1: JIA subtypes	194
• Table 2: Pediatric vasculitic syndromes	195
4. Juvenile Idiopathic Arthritis	196
5. Connective Tissue Diseases	197
• SLE, neonatal lupus, JDM	197
• Morphea, systemic sclerosis, Raynaud	198
6. Vasculitis	199
• HSP, Kawasaki, PAN	199
• Takayasu, GPA	200
7. Drugs	201
8. Visual Anchors	201
CHAPTER 21 Immunology	Pages 203 – 210
1. Basics: The Infection Tells You the Arm	203
• Workup	203

2. Comparison Tables	204
• Table 1: Primary antibody deficiencies	204
• Table 2: Combined & syndromic immunodeficiencies	205
• Table 3: Phagocytic disorders	206
• Table 4: Complement & immune dysregulation disorders	207
3. Antibody Deficiencies; T-Cell & Combined Defects	208
• XLA, CVID, hyper-IgM, IgA deficiency, SCID, WAS, DiGeorge	208
4. Phagocyte & Innate Defects; Complement & Immune Dysregulation	209
• LAD-1, CGD, HAE, ALPS, HLH-2004	209
5. Visual Anchors	210

CHAPTER 22 | Pediatric Neurology

Pages 211 – 223

1. Neurodevelopment & Congenital Malformations	211
• Neural tube defects & occult spinal dysraphism	211
• Skull deformities & craniosynostosis	212
• Syndromic craniosynostosis; brain death	213
2. Epilepsies & Seizure Syndromes	214
• Neonatal & infantile epileptic encephalopathies, Dravet	214
• Childhood epilepsy syndromes; metabolic epilepsies	215
• Status epilepticus	216
3. Neurocutaneous Syndromes	216
• NF1 & NF2, tuberous sclerosis, Sturge-Weber	216
4. Neurodegenerative, Leukodystrophies & Neurometabolic	217
• Leukodystrophies, Wilson disease, Aicardi-Gouttières	217
5. Neuro-inflammatory, Infectious & Demyelinating	218
• Autoimmune encephalitis, CNS infections	219
6. Paroxysmal & Movement Disorders	219
7. Visual Anchors	220
• EEG patterns	220
• Neurocutaneous & neurometabolic imaging	221

CHAPTER 23 | Neuromuscular Disorders

Pages 224 – 230

1. Basics: Localizing the Lower Motor Unit	224
• Workup	224
2. Comparison Table: Anatomic Localization of Weakness	225
3. Spinal Muscular Atrophy	226
• SMN1 / SMN2, types, nusinersen	226
4. Muscular Dystrophies	227
• DMD / BMD, EDMD	227
5. Myopathies, Channelopathies & Metabolic	228
• Congenital myopathies, periodic paralysis, myotonias	228
6. Neuropathies & NMJ	229
• CMT1A, GBS, myasthenia gravis	229

7. Visual Anchors 230

CHAPTER 24 | Pediatric Nephrology

Pages 231 – 242

1. Glomerular Diseases & Hematuria	231
• APSGN, IgA nephropathy vs HSP, RPGN	231
• Alport vs TBMD; nephrotic syndromes (MCNS, FSGS, membranous, MPGN)	232
• Congenital nephrotic syndromes; lupus nephritis	233
2. Tubular & Metabolic Disorders	233
• Proximal RTA & Fanconi syndrome	233
• Distal & type IV RTA; Bartter, Gitelman, pseudohypoaldosteronism	234
3. Tubulointerstitial, Vascular & Pigment Nephropathies	235
• ATIN, nephronophthisis, ADTKD, sickle cell nephropathy	235
• Contrast nephropathy, RVT, pigment-induced AKI	236
4. Cystic Kidney Diseases, CAKUT & Stone Disease	237
• ARPKD, ADPKD, syndromic cystic diseases, Potter sequence	237
• Hemorrhagic cystitis, idiopathic hypercalciuria	238
5. AKI, CKD & Pediatric Renal Transplant	238
• AKI: KDIGO staging, prerenal vs ATN	238
• CKD: etiology, formulas, CKD-MBD & growth	239
• Hypertension, dialysis, transplant	240
6. Visual Anchors	241

CHAPTER 25 | Pediatric Urology

Pages 243 – 256

1. CAKUT	243
• Embryology, renal agenesis & syndromic associations	243
• Müllerian & mesonephric malformations	244
• Multicystic dysplastic kidney (MCDK)	245
• Renal fusion & ectopia	246
2. Obstructive Uropathies	246
• Diagnosis, MAG-3 renogram	246
• Ureterocele & ectopic ureter	247
3. VUR & Reflux Nephropathy	248
• IRS grading (VCUG), management	248
4. Lower Tract & Voiding Dysfunction	249
• Neurogenic bladder, functional disorders	249
5. Penis & Scrotum	251
• Acute scrotum	251
• Penile anomalies, hypospadias	252
6. GU Trauma	252
• Renal & urethral trauma	252
7. Urolithiasis	253
• Cystinuria, calcium oxalate, struvite	253
8. Visual Anchors	254

VOLUME 2 | Pages 257 – 505

CHAPTER 26 | Pediatric Endocrinology (Part 1)

Pages 257 – 267

1. Hypopituitarism & Growth Disorders	257
• GH axis physiology (GHRH, somatostatin, ghrelin, JAK2/STAT5b, IGF-1)	257
• Clonidine–arginine GH stimulation curve	257
• Factors stimulating vs inhibiting GH secretion (table)	257
• Congenital & genetic hypopituitarism: PROP1, POU1F1 (PIT1), HESX1, GH1	258
• Clinical picture: neonatal signs, solitary median maxillary central incisor	258
• Diagnosis flowchart: bone age, IGF-1/IGFBP3, GH stimulation, MRI	258
• Management: rhGH, Mecasermin, MPHD replacement order	259
• Precocious puberty: GnRH agonists (Leuprolide)	259
2. McCune-Albright Syndrome (MAS)	259
• GNAS1 mutation, triad, diagnosis, polyostotic fibrous dysplasia X-ray	259
3. Van Wyk-Grumbach Syndrome	260
• Profound hypothyroidism, TSH–FSH receptor cross-reaction	260
4. Delayed Puberty	260
• Constitutional growth delay, Kallmann syndrome, Turner syndrome (45,X)	260
5. Thyroid Disorders	261
• Fetal development & dysgenesis: FOXE-1, NKX2.5, PAX-8	261
• Dyshormonogenesis: Pendred syndrome (SLC26A4)	261
• Congenital hypothyroidism & newborn screening flowchart	261
• Acquired: Hashimoto, Graves, consumptive hypothyroidism	262
• Familial dysalbuminemic hyperthyroxinemia	262
6. Adrenal Disorders & CAH	262
• Addison disease, APS-1 (APECED), Allgrove (3A) syndrome	262
• X-linked adrenoleukodystrophy (ABCD1, VLCFA)	262
• Congenital adrenal hyperplasia: enzyme table (21-OH, 11β-OH, 17α-OH, 3β-HSD)	263
• CAH management: acute crisis & chronic therapy	263
7. Calcium & Parathyroid	264
• PTH action & calcium homeostasis diagram	264
• DiGeorge syndrome (22q11.2, CATCH22)	264
• Pseudohypoparathyroidism 1a, AHO & PPHP	265
• Neonatal hypocalcemia: early vs late	265
• Familial hypocalciuric hypercalcemia (FHH)	265
• Jansen's metaphyseal chondrodysplasia	265
• Williams syndrome (7q11.23)	266
8. Visual Anchors	267
• Septo-optic dysplasia, Laron, Jansen's, Beckwith-Wiedemann, X-linked ALD	267

CHAPTER 27 | Pediatric Endocrinology (Part 2)

Pages 268 – 280

1. Water Balance & Posterior Pituitary	268
• AVP basics, central vs nephrogenic DI, Wolfram syndrome (WFS1)	268
• DI + adipsia: holoprosencephaly, optic nerve hypoplasia, Niikawa-Kuroki	268
• Polyuria workup flowchart: copeptin, water deprivation, DDAVP	268
• Water deprivation test curves	269
• Central vs nephrogenic DI (table)	269
• Post-neurosurgical triphasic response	270
• Runner's hyponatremia	270
• Hyponatremia: SIAD vs cerebral salt wasting	271
2. Diabetes & Glucose Regulation	271
• T1DM basics: HLA associations, Stage 2 T1DM, Teplizumab	271
• DKA management flowchart	271
• Two-bag system & DKA cerebral edema	272
• MODY: GCK vs HNF1A	273
• Monogenic & neonatal diabetes (KCNJ11/ABCC8), Wolman disease	273
3. Hypercortisolism & Adrenal Neoplasia	273
• Clinical picture: ACTH-dependent vs ACTH-independent	273
• ACTH localization pathway & BIPSS	274
• Management: TSS, preop Ketoconazole/Metyrapone, Nelson syndrome	275
• Carney complex (PRKAR1A)	275
4. Pheochromocytoma & Paragangliomas	276
• Biochemical workup: plasma & urinary metanephrines	276
• Preop management: alpha-blockade before beta-blockers	276
• Imaging: MRI "light bulb" sign	276
• Functional imaging: 123I-MIBG, 68Ga-DOTATATE PET-CT	277
• Hereditary syndromes: VHL, MEN2A/B, SDHB	277
5. APS & MEN	277
• APS-1, APS-2, IPEX (table)	277
• MEN1, MEN2A, MEN2B (table)	277
• MEN2B M918T: prophylactic thyroidectomy	278
• ROHHAD syndrome	278
6. Extra High-Yield Pearls	278
• GH & growth: radiation-induced GHD, skipping stimulation testing, Mecasermin, Sotos	278
• Thyroid & adrenal: endemic cretinism, perchlorate discharge test, GRA	279
• TBG deficiency	279
• Prolactinoma: the hook effect	279
7. Visual Anchors	280
• Pheochromocytoma MRI, pheo localization PET-CT, Wolman disease	280

CHAPTER 28 | Cyanotic Heart Disease

Pages 281 – 294

1. Pathophysiology	281
• Cyanosis: central, acrocyanosis, differential	281
• What happens at birth: fetal circulation & ductal closure	281
• The main split: oligemia vs plethora	282

2. Diagnosis	282
• Workup, step by step: pulse ox, hyperoxia test, CXR, ECG, echo	283
• Table 1: Physiological & ECG classification of cyanotic CHD	284
• Table 2: High-yield exam differentiators across cyanotic lesions	285
• Table 3: Interpretation of the hyperoxia test	287
3. Lesions: Oligemic	288
• Tetralogy of Fallot (TOF), Pentalogy of Fallot	288
• Tricuspid atresia	289
• Ebstein anomaly	290
4. Lesions: Plethoric / Mixing	290
• d-TGA	290
• TAPVR / TAPVC	291
• Persistent truncus arteriosus	291
• Hypoplastic left heart syndrome (HLHS), Norwood → Glenn → Fontan	291
5. Complications of Unrepaired TOF	293
• Cerebral abscess, cerebral venous thrombosis / stroke	293
6. CXR Silhouettes	293
• Boot-shaped heart (TOF), egg-on-a-string (d-TGA)	293
• Snowman / figure-of-8 (TAPVR), box-shaped heart (Ebstein)	294

CHAPTER 29 | Acyanotic Congenital Heart Disease

Pages 295 – 316

1. The Basics	295
• What makes it acyanotic; why shunts show up at 4–6 weeks	295
2. Left-to-Right Shunts (Volume Overload)	297
• Ventricular septal defect (VSD): basics, size decides the picture	297
• VSD: large VSD clinical signs, workup & management	298
• Atrial septal defect (ASD): basics & clinical signs	300
• ASD: workup & management	301
• Patent ductus arteriosus (PDA): basics & clinical signs	302
• PDA: workup & management	303
• Atrioventricular septal defect (AVSD / endocardial cushion defect)	303
• AVSD: clinical signs, workup & management	304
3. Obstructive Lesions (Pressure Overload)	305
• Coarctation of the aorta (CoA)	305
• Pulmonary stenosis (PS)	307
• Congenital aortic stenosis (AS)	308
4. Special Scenario: ALCAPA (Bland-White-Garland Syndrome)	310
5. Comparison Tables	311
• Table 1: Left-to-right shunt lesions	311
• Table 2: Obstructive acyanotic lesions	312
• Table 3: Genetic syndromes & associated acyanotic defects	313
6. Visual Anchors	314
• Chest X-ray	314
• Echo	316

CHAPTER 30 | Respiratory System

Pages 317 – 331

1. Basics: Airway Mechanics & Lung Function

317

- Extrathoracic vs intrathoracic narrowing, spirometry, DLCO

317

2. Workup

318

- Step-by-step: imaging, sweat chloride / nasal NO, bronchoscopy, biopsy

318

3. Comparison Tables

319

- Table 1: Congenital airway & parenchymal malformations
- Table 2: Acute inflammatory upper airway obstruction
- Table 3: Suppurative & obstructive lower airway disorders
- Table 4: Cystic fibrosis vs primary ciliary dyskinesia
- Table 5: Children's diffuse & interstitial lung diseases

319

320

321

321

322

4. Congenital Airway & Lung Anomalies

323

- Laryngomalacia, subglottic stenosis & hemangioma, vocal cord paralysis
- Vascular ring, congenital lobar emphysema, CPAM, sequestration

323

323

5. Upper Airway Obstruction

324

- Viral & spasmodic croup, epiglottitis, bacterial tracheitis, foreign body

324

6. Lower Airway & Suppurative Disease

325

- Bronchiolitis, CAP, empyema, protracted bacterial bronchitis, bronchiectasis

325

7. Cystic Fibrosis & PCD

326

8. chILD & Diffuse Lung Disease

327

- Surfactant disorders (SFTPB, ABCA3, NKX2-1), NEHI, PAP
- Acute eosinophilic pneumonia, ABPA, IPH / DAH

327

327

9. Pleural & Emergency Disorders

328

- Pneumothorax, pneumomediastinum, BRUE, CDH

328

10. Visual Anchors

329

- Steeple sign, thumb sign, shaggy tracheal membranes
- Foreign body hyperlucency, signet ring (bronchiectasis), soap-bubble (meconium ileus)
- Ground-glass (NEHI), crazy paving (PAP), sail sign (pneumomediastinum)

329

330

331

CHAPTER 31 | Systemic Cardiology

Pages 332 – 365

1. Heart Development & Birth Transition

332

- How the heart forms; heart fields
- Teratogens & epigenetics, looping, aortic arch derivatives
- Fetal heart & transition; Table 1: Embryologic lineages & arch derivatives

332

334

335

2. Pediatric Cardiac Assessment

336

- Echo, ECG, preparticipation screening (AHA 14-element checklist)
- Cath hemodynamics

336

337

3. Acyanotic CHD

338

- Table 2: Acyanotic shunts & continuous murmur differentiators
- Williams syndrome & supraventricular AS

339

340

4. Cyanotic CHD	341
• TOF, TOF with pulmonary atresia, MAPCAs	341
• Pulmonary atresia with intact septum (PA-IVS), tricuspid atresia	342
• Ebstein anomaly, d-TGA	343
• Obstructed TAPVR, HLHS	344
• Pentalogy of Cantrell	345
• Table 3: High-yield cyanotic congenital heart lesions	346
5. Heart Failure, Cardiomyopathies, Pericardium & Tumors	347
• Pulmonary arterial hypertension (PAH), HCM	347
• DCM & LV noncompaction, restrictive cardiomyopathy	348
• Takotsubo, acute myocarditis, pericardial disease	349
• Cardiac tumors; heart failure staging & assessment	350
• Nutrition & HF drugs	351
• Mechanical support & transplant	352
6. Rhythm Disorders	353
• Antiarrhythmics	353
• Specific rhythms, channelopathies	354
• Table 4: Heritable arrhythmia syndromes	355
7. Infective Endocarditis & Rheumatic Heart Disease	356
• Modified Duke criteria	356
• Prophylaxis, regimens, surgery in active IE	357
• Rheumatic carditis echo criteria, eponymous murmurs	358
8. Systemic Hypertension	359
• BP categories (AAP), primary vs secondary, monogenic HTN	359
• Vascular causes, hypertensive emergency	360
• Table 5: Syndromic associations in systemic cardiology	362
9. Visual Anchors	363
• Imaging	363
• Echo	365

CHAPTER 32 | Pediatric Gastroenterology

Pages 366 – 393

1. Esophagus & Foregut Anomalies	366
• The basics; esophageal atresia / TEF	366
• EA / TEF management; achalasia; eosinophilic esophagitis (EoE)	367
• Workup (in order): EA types, contrast esophagogram	368
• EGD + biopsy, high-resolution manometry	369
• Table 1: Esophageal pathology	370
2. Stomach & Pylorus	371
• Infantile hypertrophic pyloric stenosis (IHPS): why it happens, clinical picture, diagnosis	371
• IHPS management; gastric duplication & peptic ulcer	372
• Table 2: Gastric & pyloric pathology	373
3. Malrotation, Atresias & Mechanical Obstruction	373
• Malrotation & midgut volvulus	373
• Duodenal atresia, jejunoileal atresia; intussusception	374
• Intussusception management; Meckel's diverticulum	375
• Workup: acute neonatal / infant obstruction; meconium ileus	376
• Table 3: Intestinal obstructive anomalies	377

4. Motility: Hirschsprung & Pseudo-Obstruction	378
• Hirschsprung disease (HD): why it happens, clinical picture, diagnosis	378
• Pediatric intestinal pseudo-obstruction (PIPO)	379
• Table 4: Pediatric constipation & motility disorders	380
5. Inflammatory Bowel Disease	380
• Very-early-onset IBD (VEO-IBD); classification	381
• Management; Table 5: Crohn's disease vs ulcerative colitis	382
6. Malabsorption & Congenital Diarrheas	383
• Celiac disease: why it happens, diagnosis, nonresponsive / refractory	383
• Congenital diarrheas (CODEs)	384
• Table 6: Congenital & malabsorptive diarrheas	385
7. Gastroenteritis & Functional GI Disorders	386
• Infections: Giardia, Cryptosporidium, Shigella, Yersinia; probiotics	386
• Disorders of gut-brain interaction (Rome IV); functional (toddler's) diarrhea	386
• Cyclic vomiting & abdominal migraine; Table 7: Functional GI disorders (Rome IV)	387
8. Neonatal Cholestasis, Liver & Polyps	388
• Neonatal cholestasis, biliary atresia, GALD	388
• Polyposis syndromes: Peutz-Jeghers, juvenile polyposis	389
• Table 8: Neonatal cholestasis & liver failure	389
9. Visual Anchors	390
• Radiology	390
• Target / doughnut and coiled-spring signs (intussusception)	391
• String, shoulder, double-tract signs (pyloric stenosis)	392
• Pathology: tufting enteropathy, microvillus inclusion disease, Peutz-Jeghers polyp	393

CHAPTER 33 | Pediatric Hepatology

Pages 394 – 417

1. Neonatal Cholestasis & Biliary Malformations	394
• Why bile stops flowing; biliary atresia (EHBA)	394
• Anatomical types; Kasai hepatoportoenterostomy (KPE)	395
• Choledochal malformations	396
• Workup (in order): serum MMP7, succinylacetone	397
• Table 1: Extrahepatic & structural biliary disorders	398
2. Intrahepatic & Familial Cholestasis	399
• Alagille syndrome (ALGS); PFIC: the GGT split	399
• Low-GGT PFIC, high-GGT PFIC	400
• GALD / neonatal hemochromatosis; Table 2: Intrahepatic & metabolic cholestatic disorders	401
3. Metabolic & Inherited Liver Disease	402
• Wilson disease	402
• Alpha-1 antitrypsin deficiency; tyrosinemia type I & hereditary fructose intolerance	403
• Inherited hyperbilirubinemias (Crigler-Najjar, Gilbert, Dubin-Johnson, Rotor)	403
• Table 3: Inherited metabolic & bilirubin disorders	404
4. Acute Liver Failure, Viral Hepatitis & DILI	405
• Pediatric acute liver failure (PALF); King's College criteria	405
• Ischemic hepatitis ("shock liver")	405
• Hepatitis A; drug-induced liver injury (DILI)	406
• Table 4: Acute hepatic insults & prognostic profiles	406

5. Autoimmune Hepatitis, NAFLD & SOS	407
• Autoimmune hepatitis (AIH)	407
• NAFLD / MASLD; sinusoidal obstruction syndrome (SOS / VOD)	408
• Table 5: Autoimmune & vascular liver diseases	409
6. Portal Hypertension & Ascites	410
• The basics, causes (prehepatic, intrahepatic, posthepatic), variceal bleeding	410
• Ascites	411
• Table 6: Portal hypertension & ascites manifestations	412
7. Liver & Biliary Tumors	413
• Hepatoblastoma, HCC, mesenchymal hamartoma, PSC	413
• Table 7: Pediatric hepatic & biliary lesions	414
8. Visual Anchors	415
• Triangular cord sign, PAS-positive globules (A1AT), black liver (Dubin-Johnson)	415
• Beading (PSC), K-F ring & azure lunulae (Wilson), Meso-Rex bypass	416
• Alagille (posterior embryotoxon, butterfly vertebrae), SOS / VOD	417

CHAPTER 34 | Pediatric Hematology

Pages 418 – 431

1. Basics: Hematopoiesis & Globin Switching	418
• Lineage drivers; marrow failure syndromes; acquired aplastic anemia	418
2. Workup: Anemia	419
• Reticulocyte count & MCV-based algorithm	419
3. Comparison Table	420
• High-yield diagnostic differentiators (HS, G6PD, LCH, HLH and more)	420
4. Red Cell Production & Marrow Failure	422
• DBA, Pearson, Fanconi anemia, dyskeratosis congenita	422
• Shwachman-Diamond, CAMT, severe aplastic anemia, parvovirus B19, anemia of chronic disease	423
5. Hemoglobinopathies & Thalassemias	424
• HPLC / electrophoresis, sickle cell disease, β -thal major, β -thal intermedia, α -thal	424
6. Hemolytic Anemias (DAT Negative)	425
• HS, hereditary hydrocytosis, SE Asian ovalocytosis, G6PD, PK deficiency	425
• PNH, PCH, cold agglutinin disease, drug hemolysis	425
7. Nutritional, Megaloblastic & Sideroblastic Anemias	425
• Hereditary orotic aciduria, X-linked sideroblastic anemia (ALAS2)	425
8. Hemostasis & Thrombosis	426
• Platelet function disorders: Glanzmann, Bernard-Soulier, MYH9	426
• Mixing study approach, hemophilia (emicizumab), factor X deficiency	427
• Neonatal purpura fulminans, TTP, warfarin embryopathy, dabigatran	427
9. Leukemia, Lymphoma & Histiocytosis	428
• B-ALL, T-ALL, AML, lymphomas	428
• ALCL, LCH, HLH (HLH-2004)	429
10. Transfusion	430
11. Visual Anchors	431
• Howell-Jolly bodies, Birbeck granules (EM)	431

CHAPTER 35 | Pediatric Oncology

Pages 432 – 458

1. Epidemiology, Genetics & Predisposition 432

- How childhood cancer differs; how tumors arise 432
- Predisposition syndromes: Li-Fraumeni, WAGR, Denys-Drash, NF1 433
- Retinoblastoma (RB1), Down syndrome; workup (in order) 434
- Table 1: Genetic & translocation markers in pediatric cancers 435

2. Leukemias 436

- The basics; acute lymphoblastic leukemia (ALL), precursor B-ALL 436
- Precursor T-ALL; treatment phases 437
- Targeted therapy (blinatumomab, CAR-T, inotuzumab); AML & APL 438
- CML & JMML 439
- Table 2: Pediatric leukemia subtypes 440

3. Lymphomas 441

- The basics; Hodgkin lymphoma, Lugano staging 441
- Non-Hodgkin lymphoma: Burkitt, lymphoblastic, ALCL 442
- Table 3: Pediatric non-Hodgkin lymphoma subtypes 443

4. CNS Tumors 443

- The basics; location by age 443
- Astrocytic tumors: pilocytic astrocytoma, high-grade gliomas, DIPG 444
- Embryonal tumors: medulloblastoma, molecular subgroups 445
- Ependymoma, choroid plexus carcinoma, craniopharyngioma 446
- Table 4: Major pediatric brain tumors 447

5. Neuroblastoma & Renal Tumors 448

- Neuroblastoma: classic picture, sites, metastatic & paraneoplastic signs 448
- Neuroblastoma: diagnosis, staging imaging, high-risk protocol 449
- Wilms tumor (nephroblastoma): COG staging, histology, treatment 450
- Non-Wilms renal tumors; Table 5: Abdominal solid tumors of childhood 451

6. Bone & Soft Tissue Sarcomas 452

- Osteosarcoma 452
- Ewing sarcoma, rhabdomyosarcoma (RMS) 453
- Table 6: Pediatric bone & soft tissue sarcomas 454

7. Liver Tumors, LCH & Oncologic Emergencies 454

- Liver tumors: hepatoblastoma, HCC 454
- Langerhans cell histiocytosis (LCH), tumor lysis syndrome (TLS) 455
- Hyperleukocytosis, SVC / superior mediastinal syndrome, febrile neutropenia 456

8. Visual Anchors 456

- Starry-sky (Burkitt) 456
- Onion-skinning + CD99 (Ewing) 457
- Floating teeth + Birbeck granules (LCH), sarcoma botryoides (embryonal RMS) 458

CHAPTER 36 | Pediatric Emergencies & Critical Care

Pages 459 – 473

1. Triage, Resuscitation & Airway 459

- Why kids' airways are different; triage algorithms 459
- Airway assessment 460
- Advanced airway & RSI; RSI drugs 461

2. Cardiorespiratory Emergencies & Arrhythmias	462
• BLS standards; arrhythmias; bradycardia with pulse; epinephrine	462
• SVT algorithm (vagal maneuvers, adenosine)	463
• Post-arrest care; markers of good CPR	464
3. Shock & Hemodynamic Instability	464
• Classification; sepsis & septic shock, 1-hour bundle	465
• Vasoactives	466
4. Acute Respiratory Failure & Mechanical Ventilation	466
• Sudden deterioration: DOPE	467
5. Trauma & Neurocritical Care	467
• Chest trauma differentiators (tension pneumothorax, tamponade, hemothorax)	467
• TBI & raised ICP; brain death	468
6. Environmental & Toxicological Emergencies	469
• Drowning, burns, toxidromes & poisonings	469
7. Visual Anchors	470
• Airway	470
• Ventilator waveforms	471
• Neuroimaging	472
• Environmental	473

CHAPTER 37 | Pediatric Dermatology

Pages 474 – 490

1. Neonatal & Transient Skin Disorders	474
• Erythema toxicum neonatorum (ETN), transient neonatal pustular melanosis (TNPM)	474
• Congenital dermal melanocytosis; workup: neonatal pustules / vesicles	475
• Table 1: Transient neonatal cutaneous eruptions	476
2. Genodermatoses & Pigmentary Disorders	477
• Waardenburg vs piebaldism; Peutz-Jeghers syndrome	477
• Café-au-lait macules & neurocutaneous syndromes (NF1, McCune-Albright, Legius)	478
• Albinism & lysosomal transport syndromes (Hermansky-Pudlak, Chédiak-Higashi)	478
• Table 2: Congenital hypopigmentation & phakomatoses	479
3. Blistering Disorders & Severe Drug Reactions	479
• Why skin blisters	479
• Epidermolysis bullosa (EBS, JEB, DEB); SJS / TEN	480
• Staphylococcal scalded skin syndrome (SSSS); workup: blisters & sloughing	481
• Table 3: Severe blistering & sloughing disorders	482
4. Papulosquamous & Inflammatory Rashes	483
• Psoriasis, pityriasis rosea, diaper dermatitis	483
• Table 4: Common pediatric inflammatory dermatoses	484
5. Ichthyoses	484
• Ichthyosis vulgaris	484
• X-linked, epidermolytic, harlequin ichthyosis & Netherton	485
• Table 5: Primary inherited ichthyotic disorders	486
6. Vascular Anomalies & Photosensitivity	487
• Infantile hemangioma, PHACES, HHT; xeroderma pigmentosum, Cockayne	487
• Table 6: Photosensitive DNA repair & metabolic disorders	488

7. Visual Anchors	489
• Pityriasis rosea, bamboo hair (Netherton), ETN vs TNPM	489
• Psoriasis, SJS / TEN, SSSS, Waardenburg	490

CHAPTER 38 | Pediatric Otology Pages 491 – 498

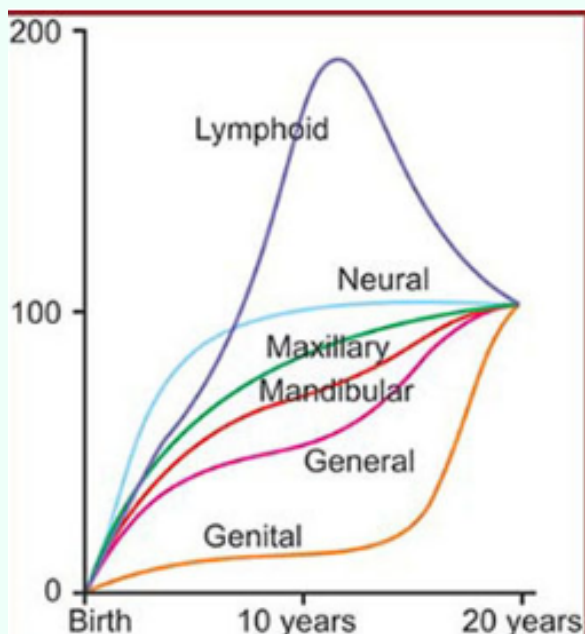
1. Basics, Hearing Loss Grades & Milestones	491
• Conductive vs SNHL; SNHL grades; auditory milestones	491
2. Workup	492
• Auditory neuropathy spectrum disorder (ANSD)	492
• Tympanogram types (A, B, C)	493
3. Comparison Tables	493
• Table 1: Syndromic causes of congenital SNHL	493
• Table 2: Acute external & middle ear inflammatory lesions	494
4. Otitis Media & Its Sequelae	495
• Acute otitis media (AOM) management; tympanosclerosis, acquired cholesteatoma	495
5. Inner Ear, Vestibular & Emergencies	496
• Vestibular neuritis, acoustic trauma, cochlear implant	496
• Button battery in the ear canal	497
6. Visual Anchors	497
• Audiograms, tympanograms, quick recall	497
• Temporal bone rhabdomyosarcoma, Waardenburg, Pendred	498

CHAPTER 39 | Pediatric Ophthalmology Pages 499 – 505

1. Basics, Refraction & Amblyopia	499
• Amblyopia, hyperopia, myopia, acuity norms	499
2. Workup	499
3. Comparison Tables	500
• Table 1: Anisocoria & pupil reactivity differentiators	500
• Table 2: Major causes of pediatric leukocoria	501
4. Strabismus, Pupils & Iris	502
• Pseudoesotropia, infantile & accommodative esotropia, Duane, aniridia (PAX6)	502
5. Glaucoma, Lens, Retina & ROP	502
• Congenital glaucoma, corneal opacities (STUMPED), keratoconus, cataract, ectopia lentis	502
• JIA uveitis, ROP screening	502
• Retinoblastoma (RB1), retinitis pigmentosa	503
6. Optic Nerve, Orbit & Trauma	504
• Septo-optic dysplasia, LHON, preseptal vs orbital cellulitis, white-eyed blowout	504
7. Visual Anchors	504
• Double ring sign	504
• Morning glory disc, medial fluid along lamina papyracea	505

1. Basics: Growth Axis & Scammon's Curves

- Growth = genes + **GH-IGF-1 axis** + thyroid hormone + sex steroids
- **Scammon's curves:**
 - **Neural:** ~80% adult size by 2 yrs, 100% by 6-7 yrs
 - **Lymphoid:** peaks ~200% at 10-12 yrs, then involutes (sex hormones)
 - **Genital:** quiet till the pubertal spurt (HPG axis)
 - **General body:** biphasic: fast infancy, steady childhood, adolescent spurt
- **Growth failure:** velocity < 2 SD or downward crossing of > 2 percentile lines; sort it out with **velocity vs bone age vs target height**
- **Weight:** 3.4 kg at birth; loses up to 10% (12% C-section), regained by 2 wks; ×2 at 5 mo, ×3 at 1 yr, ×4 at 2 yrs, ×5 at 3 yrs
- **Length:** 50 cm; +50% at 1 yr (~75), ×2 at 4 yrs (100), ×3 at 13 yrs (~150)
- **HC:** 35 cm; 2 cm/mo (0-3 mo), 1 cm/mo (3-6), 0.5 cm/mo (6-12) → +12 cm, ~47 cm at 1 yr
- **U/L ratio** (lower segment = pubic symphysis to floor): 1.7 birth, 1.3 at 3 yrs, 1.0 at 7-10, 0.9 adult
 - **High:** short-limbed dwarfism (**achondroplasia, Turner**) | **Low:** hypogonadism, Marfan



2. Workup: Short Stature & Delayed Puberty

[1. Stadiometer height (Frankfurt plane) → 1-year velocity (cm/yr)]



[2. Mid-parental height (± 8.5 cm)

Boys: $[(\text{Mother} + 13) + \text{Father}] / 2$ | Girls: $[(\text{Father} - 13) + \text{Mother}] / 2$]



[3. Bone age: left hand / wrist X-ray]



[4. If velocity ↓ or height < -2 SD off target: CBC, ESR / CRP, CMP, TSH, FT4, tTG-IgA, IGF-1, IGFBP-3, urinalysis, karyotype in ALL girls]

• **Delayed puberty:** boys **no testes ≥ 4 mL by 14**; girls **no thelarche by 13** or **no menarche by 15**

◦ Check velocity, **smell (Kallmann)**, dysmorphism, systemic illness → bone age + **LH, FSH, testosterone / estradiol, TSH, FT4, prolactin**

◦ **Low / normal LH, FSH (hypogonadotropic): CDGP (most common)**, hypothyroidism, chronic illness, **Kallmann**

◦ **High LH, FSH (hypergonadotropic):** gonadal failure: **Turner (45,X), Klinefelter (47,XXY)**

3. Comparison Tables

Table 1: Short Stature Etiologies

Feature	Familial Short Stature (FSS)	Constitutional Delay of Growth & Puberty (CDGP)	Growth Hormone Deficiency (GHD)
Growth Velocity	Normal (5-6 cm/yr)	Normal (5-6 cm/yr)	Severely Decreased (< 4 cm/yr)
Bone Age	Equals Chronological Age	Delayed (= Height Age)	Severely Delayed ($< \text{Height Age}$)
Parental Height	Short (consistent with target)	Normal ("late bloomers" history)	Normal
Body Habitus / Weight	Proportionate, normal BMI	Thin/slender, normal BMI	Truncal obesity, cherubic face, preserved weight
Final Adult Height	Short (within genetic target)	Normal (reaches normal adult target)	Reduced if untreated

Table 2: Patterns of Milestone Abnormalities

Pattern	Definition	Classic Clinical Scenario
Developmental Delay	Slow rate of achievement across domains (DQ < 70).	Global Developmental Delay, Down Syndrome.
Developmental Dissociation	Lag in ONE specific stream with normal progress in others.	Isolated speech delay at 12-19 months with normal motor skills.
Developmental Deviation	Milestone achievement occurs out of normal sequence.	Standing/walking before sitting in diplegic Cerebral Palsy.
Developmental Regression	Loss of previously acquired milestones.	Rett syndrome, Neurodegenerative disorders, Autism (25%).

Table 3: Sexual Maturity Rating (Tanner Staging)

Tanner Stage	Female Breasts	Female & Male Pubic Hair	Male Genitalia
Stage I	Prepubertal; elevation of papilla only	None	Prepubertal (Testes < 4 mL)
Stage II	Breast bud stage; subareolar tissue, areola enlarges	Sparse, straight, lightly pigmented along labia/penile base	Testes 4-6 mL; scrotal thinning/reddening
Stage III	Enlargement of breast/areola without contour separation	Darker, coarser, curlier; spreads over pubis	Testes 6-12 mL; length of penis increases
Stage IV	Areola and papilla form a secondary mound above breast	Adult-type hair; abundant, does not involve thighs	Testes 12-20 mL; penis increases in length and girth
Stage V	Mature contour; areola recedes to general breast contour	Adult distribution; extends to medial thighs	Testes > 20 mL (Adult size: 20-25 mL)

 Heads up:

Short kid, normal velocity, delayed bone age = CDGP, not FSS (FSS bone age = chronological age). **Malnutrition:** weight falls **more** than height (↓ BMI). **GHD / endocrine:** short but weight **preserved or high**.

WHO 10-step management Stabilization phase (days 1-7)

Treat hypoglycemia / hypothermia



F75 starter feed
(100 kcal/kg/day)



ReSoMal (low sodium)
for dehydration



Broad-spectrum antibiotics
(Empiric)



Vitamin A on day 1

Rehabilitation phase (weeks 2-6)



F100 / RUTF catch-up growth
(150-220 kcal/kg/day)



Iron supplementation
(3 mg/kg/day)
Only once appetite returns



Heads up:

Never give iron in the stabilization phase of SAM. It promotes bacterial proliferation and free radical toxicity. Wait for the rehab phase and a returning appetite.

Refeeding syndrome

- Carbs reintroduced → **rapid insulin surge** → drives **phosphate, potassium, magnesium** into cells (for ATP production and protein synthesis)
- **Hypophosphatemia** = pathognomonic finding → **diaphragm failure, cardiac collapse, seizures**



Heads up:

In refeeding syndrome, the electrolyte to watch is phosphate. Hypophosphatemia is the pathognomonic finding.

3. Fat-soluble vitamins (A, D, E, K)

Vitamin A (retinol)

- Needed for vision (retinal), gene expression, immune function
- Deficiency: most prevalent micronutrient deficiency globally (33% of children <5). Leads to xerophthalmia



- Toxicity: pseudotumor cerebri (raised ICP). Classic X-ray sign: mid-shaft long bone wavy cortical hyperostosis



Vitamin D & rickets

- Deficiency impairs **intestinal Ca and phosphorus absorption** → **unmineralized osteoid** at the growth plate



- **Nutritional rickets:** low/normal Ca, low Pi, **high PTH**, elevated **ALP**

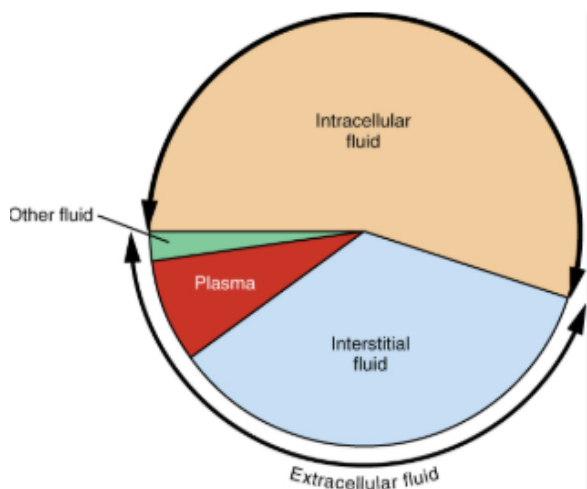
1. Fluid Compartments & Osmolality

Total body water (TBW) through childhood

- **Fetus:** ~75-80%
- **Term newborn:** ~75% of birth weight (higher in preterms)
- **By 1 year:** ~60%, stays there until puberty
- **After puberty:** girls drop to ~50% (more fat); boys stay ~60% (muscle)

ICF vs ECF

- **Newborns have more ECF than ICF** (ECF ~40% vs ICF ~35% of body weight)
- Postnatal diuresis shrinks ECF; then cell growth expands ICF
- **By 1 year, the ICF:ECF ratio reaches adult proportions:**
 - **ECF 20-25%** of body weight (plasma water 5%, interstitial 15%)
 - **ICF 30-40%** (nearly double the ECF)
- **ECF:** cations **Na⁺** and **Ca²⁺**; anions **Cl⁻** and **bicarbonate**
- **ICF:** cations **K⁺** and **Mg²⁺**; anions proteins, organic anions, phosphate



Heads up:

Major ECF cations: Na⁺ (~140 mEq/L) and Ca²⁺ (~2.5 mEq/L). Major ICF cations: K⁺ (~140 mEq/L) and Mg²⁺ (~7 mEq/L). And the ICF:ECF ratio hits adult proportions by 1 year.

Hyperkalemia

- **Pseudohyperkalemia:** hemolysis from a traumatic draw, long tourniquet, fist clenching; **thrombocytosis** (each **100,000/ μ L rise adds 0.15 mEq/L**) and **leukocytosis** (**> 200,000/ μ L**) release K^+ during clotting. Confirm with an **immediate atraumatic plasma sample**
- **Shifts:** acidosis (mineral acids), insulin deficiency (DKA), rhabdomyolysis, tumor lysis, exercise, digitalis / fluoride toxicity, β -blockers, **succinylcholine**, **hyperkalemic periodic paralysis (SCN4A)**
- **Poor excretion:** CKD, primary adrenal insufficiency (**21-hydroxylase CAH, Addison**), hyporeninemic hypoaldosteronism, **pseudohypoaldosteronism type 1 or 2 (Gordon)**, drugs (**ACEi, ARBs, spironolactone, amiloride, trimethoprim, heparin, NSAIDs, calcineurin inhibitors**)



Heads up:

Order matters. 1. Stabilize the membrane: IV calcium gluconate 10% (100 mg/kg) or calcium chloride; works in 1-3 min, but doesn't lower K^+ . 2. Shift K^+ in (15-30 min): IV regular insulin 0.1 U/kg + dextrose, nebulized albuterol, IV $NaHCO_3$ (if acidotic). 3. Remove K^+ : furosemide, SPS (Kayexalate), or hemodialysis (fastest definitive). Patiromer and SZC are for chronic hyperkalemia, not acute.

Metabolic acidosis: the anion gap

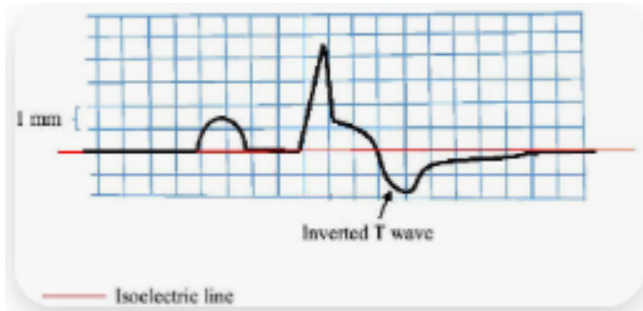
- **AG = Na - (Cl + HCO₃)**, normal **4-11 mEq/L**
- **Corrected AG = measured AG + 2.5 × (4.0 - albumin g/dL)**
- **High AG (HAGMA), unmeasured organic acids:**
 - **L-lactic** acidosis (shock, hypoxemia)
 - **D-lactic** acidosis (short bowel + bacterial overgrowth): encephalopathy, ataxia, high AG, **normal standard L-lactate**
 - **DKA** (ketoacids), **uremia** (phosphate / sulfate)
 - **Ingestions:** salicylates, **ethylene glycol (oxalate crystals)**, **methanol (formate)**
- **Normal AG (NAGMA / hyperchloremic), direct HCO₃ loss: diarrhea, RTA types 1, 2, 4, ureterosigmoidostomy, ammonium chloride**

Metabolic alkalosis: check urine chloride

- **Chloride-responsive (urine Cl < 15 mEq/L):** volume and Cl depletion keep the kidney holding HCO₃
 - **Vomiting / NG suction**, loop / thiazide diuretics (late), Cl-losing diarrhea, **cystic fibrosis**
 - Fixes quickly with **0.9% saline**
- **Chloride-resistant (urine Cl > 20 mEq/L):** mineralocorticoid excess or tubular salt wasting
 - **Hypertensive:** primary hyperaldosteronism, Cushing, **Liddle, 11β-HSD2 deficiency (licorice)**
 - **Normotensive: Bartter, Gitelman**

Hypokalemia ECG

- **ST depression, flat or inverted T**, prominent **U wave** after the T in **V2-V4**, giving an apparent long QT



Hypernatremic brain injury

- Brain water loss → brain shrinks away from the skull → **bridging veins stretch and tear** → subarachnoid / subdural / parenchymal hemorrhage, **high CSF protein**



Formulas to know cold

- **Effective osmolality:** $2 \times \text{Na} + \text{glucose} / 18$ (no urea)
- **Corrected Na:** $\text{measured Na} + 1.6 \times (\text{glucose} - 100) / 100$
- **Winter's:** $\text{expected PCO}_2 = 1.5 \times \text{HCO}_3 + 8 \pm 2$
- **Anion gap:** $\text{Na} - (\text{Cl} + \text{HCO}_3)$, normal 4-11
- **Albumin-corrected AG:** $\text{AG} + 2.5 \times (4.0 - \text{albumin})$
- **100-50-20:** 100 mL/kg (first 10 kg) + 50 mL/kg (11-20 kg) + 20 mL/kg (> 20 kg)
- **Diarrhea replacement:** D5 0.45% NS + 30 mEq/L NaHCO₃ + 20 mEq/L KCl

Gastric replacem



- **Rumination:** effortless regurgitation, re-chewing, re-swallowing ≥ 1 month, no nausea or retching, **absent in sleep**
- **Pica:** non-food items (clay, dirt, paper) ≥ 1 month, > 2 yrs $\rightarrow$ check **iron and zinc**

💡 Heads up:

MDD = leading psychiatric condition behind pediatric suicide attempts. With SSRIs, warn about **behavioural activation** and watch for the **black box suicidal ideation warning**. **No antidepressant monotherapy in bipolar I** (manic switch).

💡 Heads up:

Somatic symptom disorder is NOT a diagnosis of exclusion: it needs positive criteria, and the patient isn't faking. **Anorexia binge / purge vs bulimia = BMI** (anorexia emaciated with lanugo, bradycardia, hypothermia; bulimia normal or above). Both purgers get **hypokalemic metabolic alkalosis**.

8. Paroxysmal Events, Motor Disorders & Psychoses

- **Breath-holding spells:** 0.1-4.6% of healthy toddlers, peak onset **6-18 months**
- **Cyanotic (commonest):** anger / frustration $\rightarrow$ brief cry $\rightarrow$ **expiratory apnea** $\rightarrow$ cyanosis $\rightarrow$ brief LOC
- **Pallid:** pain / fright $\rightarrow$ **reflex vagal inhibition** $\rightarrow$ severe bradycardia $\rightarrow$ pallor, syncope
- Recovers in seconds to minutes; normal exam and milestones $\rightarrow$ **check ferritin / iron** $\rightarrow$ **reassure + iron if deficient**

1. Fever evaluation & febrile infant

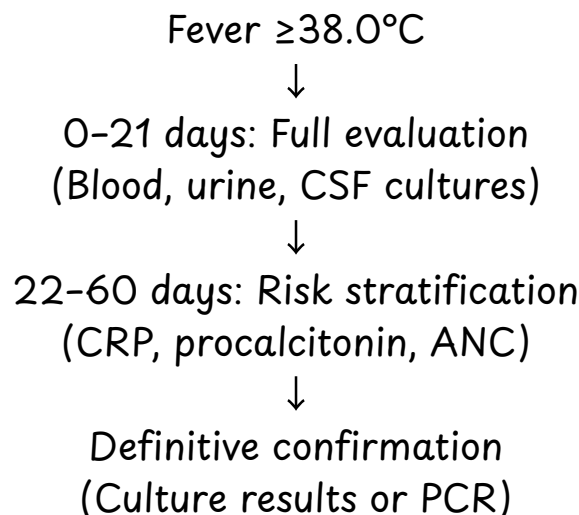
Why young infants are different

Immature immune systems → susceptible to invasive bacterial infections (IBI): bacteremia, meningitis

Often no clear source on exam, so you need a risk-stratified approach using inflammatory markers + age-based protocols

Goal: separate self-limiting viral illness from life-threatening bacteria: GBS, E. coli, Listeria monocytogenes

Febrile infant (0-60 days)



By age

- 0-21 days: full workup (including LP), hospitalization, empiric IV antibiotics (Ampicillin + Cefepime/Ceftazidime or Gentamicin)
- 22-28 days: risk stratification with markers. LP recommended if markers elevated or infant ill-appearing
- 29-60 days: can be managed as outpatient if low-risk (negative pyuria, normal inflammatory markers) and reliable follow-up is guaranteed

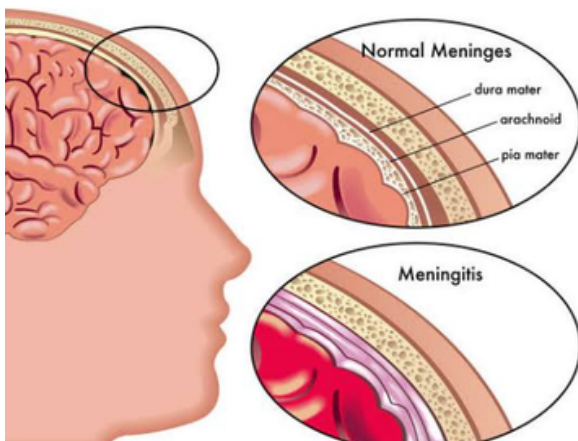
3. Invasive bacterial disease, meningitis & sepsis

Basics

- **Bacterial meningitis:** hematogenous spread to the **subarachnoid space** → meningeal inflammation + **cerebral edema**
- **Asplenia:** no splenic macrophages → impaired clearance of **encapsulated bacteria** → rapidly progressive, **fulminant sepsis**

Acute bacterial meningitis

Meningeal signs
(fever, nuchal rigidity,
Kernig / Brudzinski)



- **Neonates (<1 month):** GBS, *E. coli*, *Listeria*. Tx: **Ampicillin + Gentamicin**
- **Older children:** *S. pneumoniae*, *N. meningitidis*. Tx: **Ceftriaxone + Vancomycin**

💡 Heads up:

Suspected meningococemia with septic shock (hypotension, purpura)? Fluids come first. Resuscitation takes priority over the LP.

💡 Heads up:

Severe beta-lactam anaphylaxis? Empiric meningitis cover becomes Vancomycin + Chloramphenicol.

↓
Immediate IV fluid resuscitation
(If shock present)
↓
Empiric antibiotics
↓
Lumbar puncture
(If stable)
↓
CSF culture / PCR confirmation

💡 Heads up:

IEMs look exactly like **sepsis** (lethargy, hypothermia, apnea, poor perfusion). Start antibiotics, **but draw metabolic labs at the same time and don't delay stopping protein**. **Classic galactosemia predisposes to early E. coli sepsis**, which often comes before the metabolic diagnosis.

💡 Heads up:

Urea cycle disorders: hyperammonemia with **normal pH or respiratory alkalosis** (central hyperventilation). **Organic acidemias:** hyperammonemia with **high-AG metabolic acidosis**.

2. Amino Acidopathies & Organic Acidemias

Phenylalanine / tyrosine pathway

[Phenylalanine]

| 1. PAH + BH₄ ← PKU



[Tyrosine]

| 2. TAT ← tyrosinemia II



[4-HPPA]

| 3. 4-HPPD ← tyrosinemia III / hawkinsinuria



[Homogentisic Acid]

| 4. HGD ← alkaptonuria



[Fumarylacetoacetate]

| 5. FAH ← tyrosinemia I



[Fumarate + Acetoacetate]

PKU & BH₄ deficiency

- **AR, phenylalanine hydroxylase (PAH)**; Phe can't become tyrosine, so it's shunted to **phenylpyruvate, phenyllactate and phenylacetate (phenylacetate gives the musty / mousy odour)**; **tyrosine becomes essential**
- Plasma Phe > **1,200 μmol/L** → impaired myelination and neurotransmitter synthesis
- **Untreated:** progressive ID / DD, **hypopigmentation** (fair hair, blue eyes, fair skin), **eczema-like rash**, microcephaly, spasticity, **musty odour**

Muscle GSDs

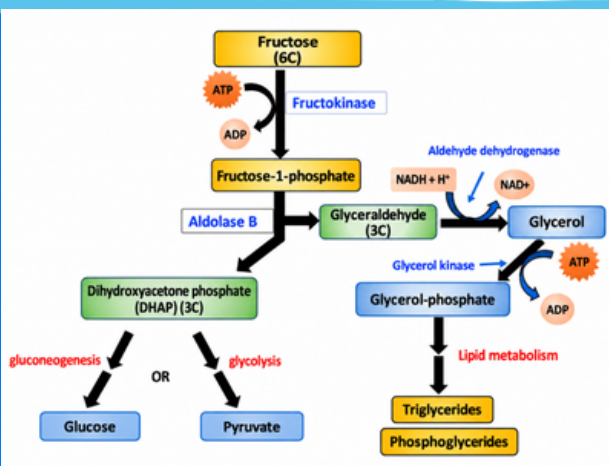
- **GSD II (Pompe, lysosomal acid α -1,4-glucosidase / acid maltase, AR):** glycogen in lysosomes of heart, skeletal and smooth muscle

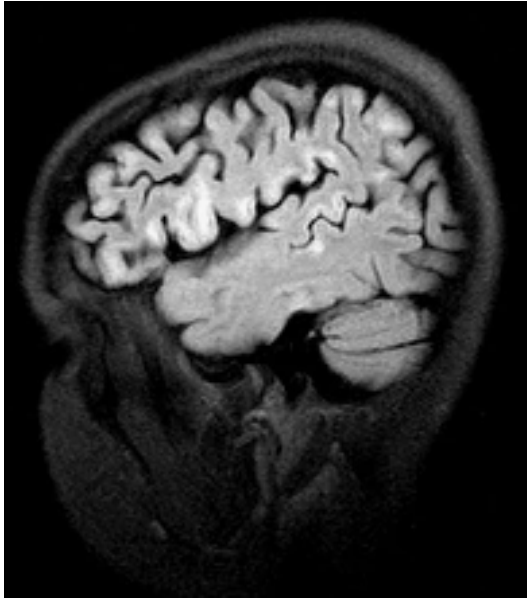


- **Infantile: floppy baby, massive cardiomegaly / HCM, macroglossia**, feeding difficulty, **high CK**; ECG **short PR + tall QRS**; death in year 1 untreated
- **Alglucosidase alfa ERT**
- **GSD V (McArdle, myophosphorylase PYGM, AR):** adolescents / young adults
 - Exercise intolerance, painful **cramps** with short intense exercise, **myoglobinuria** (cola urine) → AKI risk
 - **"Second wind"** after **10-15 min** (fatty acids and glucose mobilized)
 - CK chronically high; **lactate fails to rise on the ischemic forearm test**
- **GSD VII (Tarui, muscle PFK / PFKM):** like McArdle but **"out-of-wind"** (carb meals **worsen** exercise tolerance by blocking lipolysis) + **nonspherocytic hemolytic anemia** (RBCs have the M subunit)

Galactose & fructose

- **Lactose** → **galactose** → (galactokinase) → **Gal-1-P** → (GALT) → **glucose-1-P**
- **Sucrose** → **fructose** → (fructokinase) → **Fru-1-P** → (aldolase B) → **DHAP + glyceraldehyde**





Kearns-Sayre: onset < 20 years, PEO, pigmentary retinopathy, plus heart block or CSF protein > 100 mg/dL; single large mtDNA deletion

- **Alpers-Huttenlocher (POLG):** intractable epilepsy, regression, acute liver failure triggered by valproate

6. Lysosomal & Peroxisomal Disorders

Sphingolipidoses & lipid storage

- **Gaucher (GBA, AR):** most common LSD (Ashkenazi); glucocerebrosidase → glucosylceramide
 - **Type 1 (non-neuronopathic, 99%):** massive splenomegaly, hepatomegaly, anemia, thrombocytopenia, bone crises, Erlenmeyer flask femurs; no primary neuro involvement; Gaucher cells ("wrinkled / crumpled tissue paper", PAS+); imiglucerase ERT
 - **Type 2 (acute neuronopathic):** rapid infant neurodegeneration, opisthotonos, trismus, death < 2 years
 - **Type 3 (subacute neuronopathic):** juvenile progressive myoclonic epilepsy and ataxia
- **Niemann-Pick A & B (SMPD1):** acid sphingomyelinase → sphingomyelin
 - **Type A:** infant, massive HSM, neurodegeneration, cherry-red spot (50%), foamy histiocytes in marrow; death by 2-3 years
- **Tay-Sachs (HEXA):** GM2; onset 4-5 months: hyperacusis, motor regression, macrocephaly, cherry-red spot, no organomegaly
- **Sandhoff (HEXB):** hexosaminidase A and B; Tay-Sachs picture plus mild HSM, cardiac involvement, bone changes
- **Fabry (GLA, X-linked recessive):** α-galactosidase A → Gb3; young males with acroparesthesias, angiokeratomas (bathing-suit), hypohidrosis, cornea verticillata; later renal failure, HCM, stroke
- **Krabbe (GALC):** globoid cell leukodystrophy; extreme irritability, hypertonia, spasticity, optic atrophy, death < 3 years; multinucleated globoid cells in white matter
- **Metachromatic leukodystrophy (ARSA):** arylsulfatase A → sulfatides; central + peripheral demyelination, gait ataxia, spasticity, optic atrophy, neuropathy with absent reflexes

Table 3: Lysosomal & Peroxisomal Storage Diseases

Disease	Inheritance / Defective Gene	Stored / Accumulated Substrate	Key Pathognomonic / Differentiating Finding	Diagnostic Gold Standard
Gaucher Type 1	AR / GBA (Glucocerebrosidase)	Glucosylceramide	Gaucher cells ("wrinkled tissue paper") + Erlenmeyer flask	Leukocyte β -Glucosidase activity
Niemann-Pick A	AR / SMPD1 (Sphingomyelinase)	Sphingomyelin	Cherry-red spot + Hepatosplenomegaly + Foam cells	Leukocyte Sphingomyelinase assay
Tay-Sachs	AR / HEXA (β -Hexosaminidase A)	GM2 Gangliosides	Cherry-red spot + NO organomegaly + Hyperacusis	Leukocyte Hexosaminidase A assay
Fabry Disease	X-Linked / GLA (α -Galactosidase A)	Globotriaosylceramide (Gb3)	Acroparesthesias + Angiokeratomas + Renal/Cardiac	Leukocyte α -Galactosidase A assay
Krabbe Disease	AR / GALC (Galactocerebrosidase)	Galactosylceramide	Multinucleated Globoid cells in white matter	Leukocyte Galactocerebrosidase assay
MLD	AR / ARSA (Arylsulfatase A)	Cerebroside Sulfate	Demyelination + Metachromatic granules in urine	Leukocyte Arylsulfatase A assay
Hurler (MPS I-H)	AR / IDUA (α -L-Iduronidase)	Dermatan & Heparan Sulfate	Coarse facies + Dysostosis multiplex + CORNEAL CLOUDING	Leukocyte α -L-Iduronidase assay
Hunter (MPS II)	X-Linked / IDS (Iduronate Sulfatase)	Dermatan & Heparan Sulfate	Coarse facies + Dysostosis multiplex + CLEAR CORNEAS	Leukocyte Iduronate Sulfatase assay
Wolman Disease	AR / LIPA (Lysosomal Acid Lipase)	Cholesteryl Esters & Triglycerides	Diarrhea/Steatorrhea + BILATERAL ADRENAL CALCIFICATION	Leukocyte Acid Lipase assay
Zellweger (ZSD)	AR / PEX genes (Peroxisins)	Very Long-Chain Fatty Acids	Dysmorphic + Profound hypotonia + Renal cortical cysts	Elevated Plasma VLCFA C26:0 panel
X-Linked ALD	X-Linked / ABCD1 Transporter	Saturated VLCFAs (C26:0)	Parieto-occipital demyelination + Adrenal Insufficiency	Elevated Plasma VLCFA C26:0 panel
Refsum Disease	AR / PHYH (Phytanoyl-CoA Hydrox)	Phytanic Acid	Retinitis pigmentosa + Ataxia + Anosmia + Ichthyosis	Elevated Plasma Phytanic Acid

Pediatric Genetics & Inborn Errors of Metabolism

1. Cytogenetics & Chromosome Errors

The basics

- Chromosomal disorders: **1-2% of live births, 5% of stillbirths, 50% of first-trimester losses**
- Normal: **46 chromosomes** (22 autosome pairs + XX or XY)
- DNA replicates in **S phase** → sister chromatids joined at the centromere
- **Mitosis** (somatic) → identical diploid cells; **meiosis** (germline) → haploid gametes (23) via two divisions

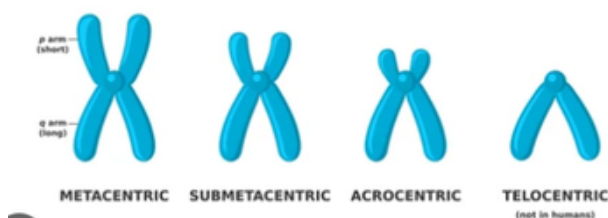
What goes wrong

- **Aneuploidy** (gain / loss of single chromosomes): mostly **meiotic nondisjunction**, mainly **maternal meiosis I**, linked to **maternal age ≥ 35**
 - Trisomy (21, 18, 13, 8, sex chromosomes), monosomy (45,X Turner)
- **Polyploidy**: whole extra haploid sets (**triploidy 69,XXX / XXY / XYY**)
 - Non-mosaic triploidy is **lethal early in the neonatal period**; often from **dispermy** or meiotic non-separation
 - **Extra paternal set** → **partial hydatidiform mole** (big placenta)
 - **Extra maternal set** → **severe embryonic growth restriction** with a small fibrotic placenta
- **Structural rearrangements** (breakage / repair errors): reciprocal and Robertsonian translocations, deletions, duplications, inversions, rings

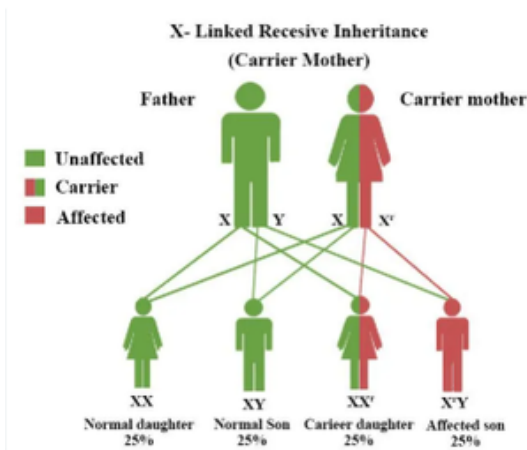
Karyotyping

- Culture nucleated cells (usually **peripheral blood T-lymphocytes**) → arrest in metaphase with **colchicine** (blocks spindle) → hypotonic solution → **Giemsa (G-banding)**
- Standard **550-600 bands**: dark **G-positive** = AT-rich, light **G-negative** = GC-rich
- **High-resolution prometaphase: 550-850 bands**
- **Centromere position**:
 - **Metacentric**: central, p and q nearly equal (e.g. **chromosome 1**)
 - **Submetacentric**: off-centre (e.g. **chromosome 11**)
 - **Acrocentric**: near one end (**13, 14, 15, 21, 22**); short arms are repetitive **rDNA stalks** and **satellites** with no essential unique genes

TYPES OF CHROMOSOMES



- **Y-linked (holandric):** males only; **100% father-to-son**, no daughters; genes for sex determination (**SRY**) and spermatogenesis
- **Pseudoautosomal:** X and Y share **PAR1 and PAR2** at their tips, which recombine in meiosis; genes there (e.g. **SHOX**) have two active copies in both sexes
 - **Heterozygous SHOX** → **Leri-Weill dyschondrosteosis** (short stature, forearm bowing, **Madelung deformity**); looks AD though it's on the sex chromosomes
 - **Homozygous SHOX** → **Langer mesomelic dysplasia**



Triplet repeat disorders & anticipation

- Unstable 3-bp repeats expand further in meiosis → **anticipation**: worse disease, earlier onset in each generation
- **Fragile X (FMR1, Xq27.3):** CGG in the **5' UTR**
 - **Normal 5-44; premutation 55-200 (FXTAS in adult males, FXPOI in females); full mutation > 200**
 - Full mutation → promoter **hypermethylation**, FMR1 silenced → intellectual disability, **macroorchidism**, long face, large ears, autistic features
 - **Premutation expands to full mutation only through the mother**
- **Myotonic dystrophy type 1 (DMPK, 19q13.3):** AD, CTG in the **3' UTR**
 - Strong anticipation, especially **maternal** → **congenital form** (neonatal hypotonia, respiratory distress, arthrogryposis)
 - Adult: **myotonia**, distal weakness, frontal balding, **cardiac conduction block**, **posterior subcapsular "Christmas tree" cataracts**, hypogonadism
- **Huntington:** CAG in an exon of **HTT**, **paternal** expansion bias
- **Friedreich ataxia:** GAA in an intron of **FXN**, **AR**
- Also the **spinocerebellar ataxias**

1. Basics

- GFR ~10-20 mL/min/1.73 m² at birth, doubles by 2 weeks; nephrogenesis continues to 34 wks in preterms; immature tubules lose Na, concentrate poorly
- AKI (KDIGO, within 7 days): SCr ↑ ≥ 0.3 mg/dL in 48 h, ≥ 50% rise, or UOP < 1 mL/kg/h over 24 h; prerenal 75-80% (volume loss, asphyxia, NSAIDs, ACE inhibitors) → sustained → ATN
- PUV / LUTO: oligohydramnios in the canalicular stage (16-28 wks) → pulmonary hypoplasia + Potter facies + palpable bladder (± urinary ascites, hydroureteronephrosis)



- RVT: most common non-catheter spontaneous thrombosis in newborns; IDM, asphyxia, polycythemia, dehydration, UVC; 70% unilateral, mostly left → gross hematuria + flank mass + severe thrombocytopenia
- Renovascular HTN: UAC thromboembolism, renal artery stenosis, compression; BP ≥ 95th centile for PMA + tachypnea, feeding intolerance, heart failure

2. Workup: Neonatal AKI

[1. Hydration status + abdomen / POCUS: distended bladder → 5 Fr feeding tube (no routine catheter)]



[2. SCr, urea, electrolytes, ABG, urinalysis + spot urine Na, Cr, FENa, RFI BEFORE fluid / diuretic]



[3. No CHF → NS 10-20 mL/kg over 30-60 min: UOP > 1 mL/kg/h = PRERENAL]



[4. Still oliguric → furosemide 1 mg/kg IV once: no response = INTRINSIC AKI]



[5. IWL (20 mL/kg/d term) + mL-for-mL losses, stop nephrotoxins, watch K, acidosis, overload]

Others

- **Treatable ataxias:**
 - **Cerebrotendinous xanthomatosis (CTX):** **Chenodeoxycholic acid**
 - **Friedreich ataxia:** **GAA triplet repeat**
- **Fabry disease:** **alpha-galactosidase A** deficiency. Triad: **acroparesthesia, cornea verticillata, angiokeratomas**
- **Aicardi-Gouttières (AGS):** **type I interferonopathy**. **JAK inhibitors (Baricitinib)** are the disease-modifying focus
- **Rett syndrome:** **MECP2**. **Trofinetide** (IGF-1 analog) is the only FDA-approved treatment

5. Neuro-inflammatory, infectious & demyelinating

- Demyelination is judged by **dissemination in space and time**
- Infections need monitoring for **long-term sequelae**

Demyelinating spectrum (ADS syndromes)

Disorder	Hallmark Feature	Diagnostic Marker
ADEM	Encephalopathy	"Fluffy" lesions (MRI)
MS	No encephalopathy	Oligoclonal Bands (CSF)
NMOSD	LETM	AQP4-IgG
MOGAD	Optic Neuritis	MOG-Ab

1. Basics: Localizing the Lower Motor Unit

- **Motor unit** = LMN (brainstem / **anterior horn**) + axon + **NMJ** + its muscle fibers. **UMN** lesions: **spasticity, hyperreflexia, Babinski**, bulk preserved initially
- **Passive tone** = range of motion; **active tone** = resistance to movement; **head lag** = **weakness**, not just low tone
- **Myopathy** → **proximal** (except **myotonic dystrophy, distal**); **neuropathy** → **distal** (except **5q-SMA, proximal**); **NMJ** → **fatigable** bulbar / ocular / facial
- **Reflexes** lost early in neuropathy / motor neuron disease, **preserved till late in myopathy**. **Tongue fasciculations** = LMN denervation
- **Pain** = **acuity** (dermatomyositis, rhabdo, acute GBS); dystrophies / SMA painless (except **Becker exercise cramps**)
- **Cryptorchidism** (weak gubernaculum) and **bell chest with thin radiolucent ribs: SMA 1, myotubular myopathy, neonatal DM1** (cryptorchidism also congenital myopathies)



2. Workup

- [1. CNS (encephalopathy, dysmorphism, hyperreflexia, clonus)
vs PNS (weak, areflexic, fasciculations, bell chest); examine mother]
▼
- [2. CK: < 1,000 SMA / congenital myopathy / neuropathy / NMJ
1,000–5,000 CMD, LGMD | 15,000–35,000+ DMD / BMD]
▼
- [3. Genetics first: SMN1 MLPA (~95%) | dystrophin del / dup | DMPK CTG | PMP22]
▼
- [4. EMG / NCS / RNS] → [5. NGS / exome / biopsy: Gomori rods | NADH-TR / SDH
cores | IHC dystrophin, sarcoglycan, merosin]



Heads up:

Low C3 that hasn't come back up after 8 weeks isn't APSGN anymore. APSGN normalises by 6-8 weeks, so think MPGN.

Congenital & syndromic nephrotic syndromes

Gene / syndrome	Key points
NPHS1	Nephrin, Finnish type. Elevated maternal AFP, enlarged placenta (>25% of birth weight)
NPHS2	Podocin
WT1	Denys-Drash
Galloway-Mowat	Microcephaly + hiatal hernia
Pierson syndrome	

Lupus nephritis

- **ISN/RPS Classes I-VI**
- Class III/IV induction: **IV cyclophosphamide** vs **MMF** (MMF to avoid gonadal toxicity)
- Class V = **membranous**
- Indication limits for **plasmapheresis**

Module 2: Tubular & metabolic disorders

Renal tubular acidosis

Proximal RTA (Type II) & Fanconi syndrome

- **Bicarbonate wasting** (Na^+ - 3HCO_3^- cotransporter defect)
- Fanconi triad: **glycosuria, phosphaturia, aminoaciduria**
- **Cystinosis** (CTNS)
- **Lowe syndrome** (OCRL1): cataracts, hypotonia, pRTA
- Drug etiologies

Management

- Observation: if differential function preserved and drainage satisfactory
- Dismembered pyeloplasty if:
 - Differential renal function $<40\%$
 - Poor drainage ($T_{1/2} >20$ min)
 - Progressive hydronephrosis

💡 Heads up:

If there's VUR, keep the bladder continuously drained during the MAG-3. Otherwise washout looks falsely prolonged and you'll call obstruction that isn't there.

Ureterocele & ectopic ureter

Ureterocele

- Cystic dilatation of the distal ureter
- IVP: "cobra head" or round, radiolucent filling defect near the ureteral orifice



Ectopic ureter

- Inserts **distal to the sphincter**

💡 Heads up:

Girl who voids normally but is always damp or leaking? Think ectopic ureter. Urine is bypassing the sphincter.

Penile anomalies

Hypospadias

- Ideal repair at 6-12 months



Heads up:

Don't circumcise a kid with hypospadias. The foreskin is needed for the repair.



Heads up:

Proximal hypospadias + cryptorchidism means DSD evaluation. Karyotype first.
Phimosis

- Pathologic (BXO): white/scarred
- Physiologic: normal in infants

Paraphimosis

- Emergency, trapped foreskin
- Granulated sugar (osmotic gradient) to reduce edema before reduction

VI. GU trauma



Heads up:

Hemodynamic stability decides the path, not just the injury grade.

Renal trauma grading

- Grade III: laceration >1 cm, no collecting system involvement
- Grade IV: urinary extravasation or collecting system involvement
- Grade V: shattered kidney or renal hilar avulsion

Urethral trauma

Blood at meatus + pelvic fracture



Retrograde urethrogram (RUG)



Membranous rupture:
Suprapubic tube first



Delayed repair at 3-6 months

3. Van Wyk-Grumbach syndrome

- **Profound hypothyroidism** → extremely high **TSH**
- **α -subunit homology** means TSH cross-reacts with **FSH receptors**

💡 Heads up:

Precocious puberty with a **delayed** bone age is the giveaway for Van Wyk-Grumbach. That's what sets it apart from the other causes.

Delayed puberty

- **Constitutional growth delay**: variant of normal. **Delayed bone age**, late pubertal spurt. Often a family history of "**late bloomers**"
- **Kallmann syndrome: hypogonadotropic hypogonadism + anosmia**
 - **KAL1** (X-linked) or **FGFR1** (autosomal dominant)
- **Turner syndrome (45,X): hypergonadotropic hypogonadism**
 - Webbed neck, short stature
 - **Bicuspid aortic valve** = most common cardiac defect



Acquired thyroid disorders

- **Hashimoto thyroiditis**: autoimmune destruction. **Most common cause of acquired goiter / hypothyroidism** in children
- **Graves disease**: **TSH-receptor stimulating antibodies** → hyperthyroidism
 - **Methimazole** preferred over **PTU** (less hepatotoxic)
 -
- **Consumptive hypothyroidism**: massive **Type 3 deiodinase** expression in **large hepatic hemangiomas**, inactivates T4/T3

💡 Heads up:

Go carefully with radioactive iodine in Graves. It can make Graves ophthalmopathy worse.

💡 Heads up:

Familial dysalbuminemic hyperthyroxinemia is a favourite MCQ distractor. High total T4 but normal free T4 and TSH. The patient is euthyroid and needs no treatment.

4. Adrenal disorders & CAH

Primary adrenal insufficiency (Addison disease)

Adrenal cortex destroyed → glucocorticoid + mineralocorticoid deficiency → high ACTH, hyperpigmentation

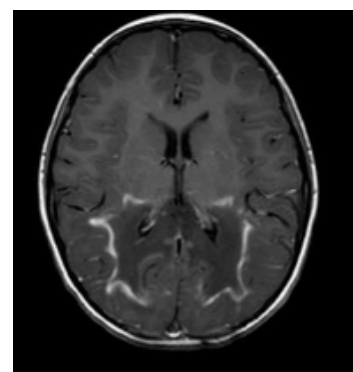
APS-1 (APECED): AIRE mutation. Triad of candidiasis, hypoparathyroidism, Addison

Allgrove (3A) syndrome: achalasia, alacrimia, adrenal insufficiency

X-linked adrenoleukodystrophy (ALD): ABCD1 mutation

Accumulation of VLCFA, especially hexacosanoic acid (C26:0)

MRI: symmetric white matter lesions in the splenium of the corpus callosum



Congenital adrenal hyperplasia (CAH)

- Enzyme blocks in the cortisol pathway push precursors toward **androgens** or **mineralocorticoid precursors**

CAH Type	Enzyme	BP	Genitals (Female)	Genitals (Male)	Other
21-Hydroxylase	CYP21A2	Low	Virilized	Normal	90% of cases; salt-wasting.
11 β -Hydroxylase	CYP11B1	High	Virilized	Normal	11-deoxycorticosterone excess.
17 α -Hydroxylase	CYP17A1	High	Normal	Ambiguous	Sexual infantilism.
3 β -HSD	HSD3B2	Low	Mildly Virilized	Ambiguous	Affects all pathways.

Management

- Acute crisis:** IV hydrocortisone (stress dosing) + aggressive fluid resuscitation (NS bolus)
- Chronic:** glucocorticoids (suppress ACTH / androgens) + **mineralocorticoids** (Fludrocortisone)

Opsoclonus-myoclonus-ataxia ("dancing eyes, dancing feet"): autoimmune; linked to **favorable, differentiated** tumors

- **Kerner-Morrison:** VIP secretion → intractable watery diarrhea + hypokalemia
- **Horner syndrome:** ptosis, miosis, anhidrosis from cervical / thoracic chain compression



💡 Heads up:

Stage 4S (INSS) / MS (INRG): localized primary in an infant < **18 months** with spread **only to liver, skin and / or minimal marrow (< 10%)**. **Cortical bone mets automatically exclude 4S / MS.** Prognosis is excellent (> **80-90%**) and it often **regresses spontaneously** without chemo.

Diagnosis

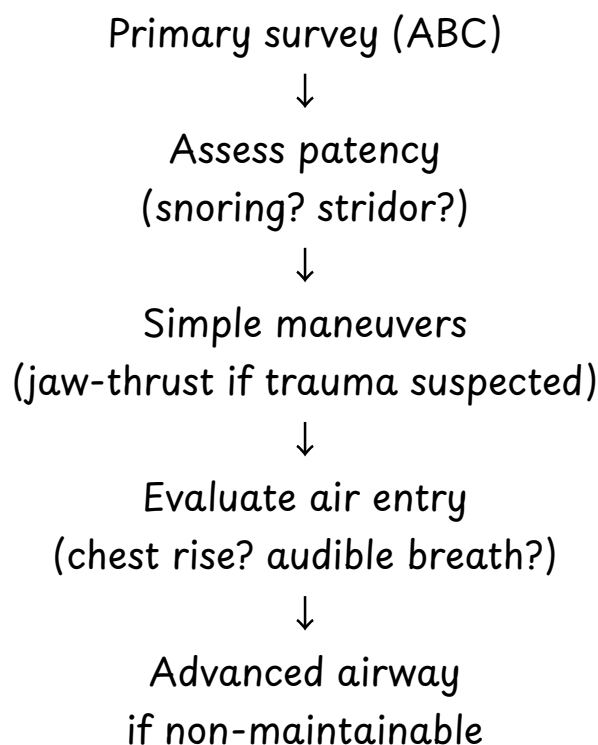
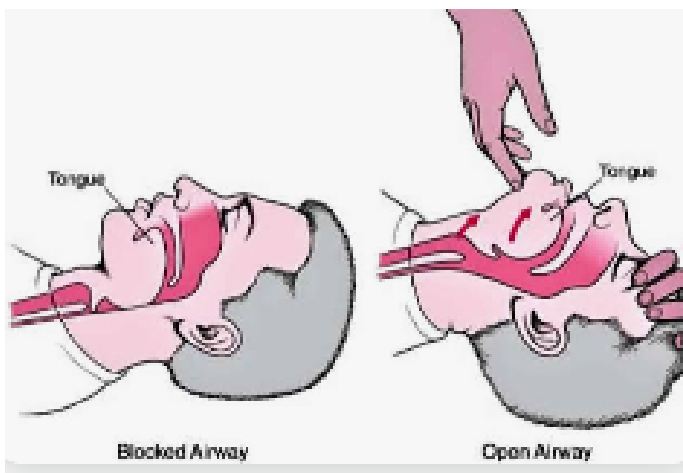
- **24-hour urine VMA and HVA (> 95% sensitive)** + marrow with small round blue cells in **Homer Wright rosettes**
- Staging imaging: **123I-MIBG** or **68Ga-DOTATATE PET-CT**
- **MYCN amplification (2p)** is the **single strongest marker of high risk**; segmental changes (**1p loss, 11q loss, 17q gain**) are also unfavorable

High-risk protocol

1. **Induction:** cisplatin, etoposide, cyclophosphamide, doxorubicin, vincristine
2. **Local control:** surgical debulking + focal radiation
3. **Consolidation:** myeloablative chemo + **tandem autologous SCT**
4. **Maintenance:** **isotretinoin (13-cis-retinoic acid)** + **dinutuximab (anti-GD2)** + cytokines (**IL-2 / GM-CSF**)

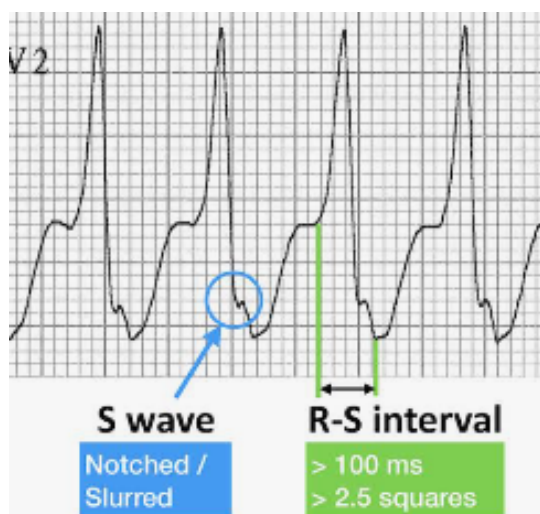
Airway assessment

- **Patent:** unobstructed, quiet, effortless breathing
- **Maintainable:** initially obstructed (e.g. snoring) but becomes patent with simple maneuvers (**head-tilt/chin-lift** or **jaw-thrust**)
- **Non-maintainable:** obstruction persists despite simple maneuvers, needs advanced intervention



💡 Heads up:

Rescue breaths in infants need a mouth-to-nose-and-mouth seal. In older kids it's mouth-to-mouth with the nose pinched. And don't overextend an infant's neck, it can narrow the airway.



💡 Heads up:

Endotracheal epinephrine is 0.1 mg/kg (1 mg/mL concentration), 10 times the IV/IO dose of 0.01 mg/kg (0.1 mg/mL). Mixing these up is a classic error.

💡 Heads up:

Hyperglycemia is NOT one of the reversible Hs in pediatric arrest.

Post-arrest care

- **TTM:** maintain **32-37°C**. Avoid fever, it worsens neurologic injury
- **Oxygenation:** target **SpO2 94-99%**, avoid hyperoxia
- **Hemodynamics:** keep **SBP >5th percentile** for age
- **Seizures:** **continuous EEG** needed, post-arrest seizures are often **non-convulsive**

Markers of good CPR

- **ETCO2 >10-15 mmHg**
- **Diastolic BP** (correlates with coronary perfusion)

3. Shock & hemodynamic instability

- Shock = impaired **oxygen delivery (DO2)** relative to tissue demand (**VO2**)
- **Compensated shock** in kids: tachycardia + poor perfusion with a **normal systolic BP** (robust systemic vasoconstriction)