PEDIATRICS

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PEDIATRICS ... CONT.

REGULAR VISITS

- ❑ purpose: prevention, screening, advocacy
- ❑ usual schedule: newborn, 1 week post-discharge, 1, 2, 4, 6, 9, 12, 15, 18, 24 months
  - yearly until age 6, then every other year
  - yearly after age 11
- ❑ history
  - pregnancy and neonatal history
  - feeding and diet (see Table 1)
  - immunizations (see Tables 2 and 3)
  - developmental assessment (see Table 4)
  - growth, energy, appetite, sleep and review of systems
  - past medical history, family and social history, allergies and medications
- ❑ physical exam
  - growth: serial height, weight, head circumference
  - head and neck: dysmorphic features, red reflex, palate, fontanelles (anterior closes between 9-18 months, posterior between 2-4 months), strabismus, vision, tympanic membranes, hearing
  - cardiovascular: auscultation, peripheral pulses (including femorals), BP yearly after age 3
  - chest, abdominal, GU, skin
  - MSK: hips (Barlow and Ortolani tests), scoliosis, lumbosacral spine (hairy patch, pigmentation, sinus tract)
  - neurologic: primitive reflexes in newborns and in early infancy
- ❑ counselling/anticipatory guidance (see Nutrition, Colic, and Child Injury Prevention Sections)
  - healthy infants should be positioned for sleep on side or back (decrease incidence of SIDS - see Sudden Infant Death Syndrome Sections)

NUTRITION

Breast Feeding

- ❑ colostrum (100 ml) for first few days – clear fluid with nutrients and immunologic protection for baby
- ❑ full milk production by 3-7 days (mature milk by 15-45 days)
- ❑ support for mothers who want to breast feed (e.g. La Leche League, lactation consultant) should start while in hospital
- ❑ assessment of adequate intake: weight gain, number of wet diapers, number of bowel movements, pause during sucking, swallowing
- ❑ feeding schedule
  - premature infants: q 2-3 hours
  - term infants: q 3.5-4 hours
- ❑ breast-fed babies require supplementation with
  - vitamin K (given IM at birth)
  - vitamin D (Tri-Vi-Sol or Di-Vi-Sol)
  - fluoride (after 6 months if not sufficient in water supply)
  - iron (premature infants): 8 weeks to 1st birthday
  - iron (exclusively breast-fed infants): after 6 months
- ❑ contraindications
  - mother receiving chemotherapy or radioactive compounds
  - mother with HIV/AIDS, active untreated TB, herpes (primary or in breast region)
  - mother using alcohol and/or drugs (affects breast milk in 2 ways: decrease milk production and/or directly toxic to baby)
  - mother taking certain medications (most are safe): e.g. antimetabolites, bromocriptine, chloramphenicol, high dose diazepam, ergots, gold, metronidazole, tetracycline

Advantages of Breast Feeding

- ❑ “breast is best"
- ❑ composition of breastmilk
  - energy: 20 kcal/oz.
  - carbohydrate: lactose
  - protein: whey 80% (more easily digested than casein), casein 20%, essential amino acids (lower content than cow’s milk, lower renal solute load for developing kidneys)
  - fat: cholesterol, triglycerides, essential free fatty acids (up to 50% energy from fat)
  - iron: higher bioavailability (50% of iron is absorbed vs. 10% from cow's milk), supply for first 6 months

- ❑ immunologic
  - lower allergenicity than cow's milk (protein)
  - IgA, macrophages, active lymphocytes, lysozyme, lactoferrin (lactoferrin inhibits *E.coli* growth in intestine)
  - lower pH promotes growth of lactobacillus in the GI tract (protective against pathogenic intestinal bacteria)
- ❑ bonding
- ❑ economical
- ❑ convenient

Complications of Breast Feeding
- ❑ sore/cracked nipples: try warm compresses, massage, frequent feeds
- ❑ breast engorgement: continue breast feeding and/or pumping
- ❑ mastitis (usually due to S. *aureus* acquired from baby): treat with cold compresses between feeds, cloxacillin for mother, continue nursing +/– incision and drainage
- ❑ breast milk jaundice: 1% of newborns (see Jaundice Section)
- ❑ poor weight gain: consider dehydration or failure to thrive
- ❑ thrush: check baby's mouth for white cheesy material; treat with antifungal

Alternatives to Breast Feeding
- ❑ formulae: 100-120 kcal/kg/day = 150-180 cc/kg/day (minimum)
  - cows based formulae, e.g. SMA, Similac, Enfalac with iron
  - soya protein based formulae e.g. Isomil, Prosobee with iron
  - iron fortified formula recommended
  - use one formula consistently
- ❑ special formulae: for protein hypersensitivity, lactose intolerance, galactosemia, PKU, other malabsorption syndromes (all rare)
- ❑ cow's milk
  - should not be used under 9 months of age because of high renal solute load, poor iron absorption and inappropriate energy distribution
  - homo milk starting 9-12 months until 24 months, then 2% or skim milk
- ❑ vegan diet is not recommended in first 2 years

Table 1. Dietary Schedule

Age	Food	Comments
0 to 4 months	breast milk, formula	can be used exclusively until 6 months of age
4 to 6 months	iron enriched cereals	rice cereals first because less allergenic
4 to 7 months	pureed vegetables	yellow/orange vegetables first and green last (more bulk) avoid vegetables with high nitrite content (beets, spinach, turnips) introduce vegetables before fruit
6 to 9 months	pureed fruits and juices pureed meats, fish, poulty, egg yolk	avoid desserts no egg white until 12 months (risk of allergy)
9 to 12 months	finger foods, peeled fruit, cheese and cooked vegetables	NO peanuts or raw, hard vegetables till age 3 to 4 years no added sugar, salt, fat or seasonings

COLIC
- ❑ rule of 3's: unexplained paroxysms of irritability and crying for > 3 hours/day and > 3 days/week for > 3 weeks in an otherwise healthy, well-fed baby
- ❑ occurs in 1:5 babies
- ❑ etiology: generally regarded as a lag in the development of normal peristaltic movement in GI tract
- ❑ other reasons why babies cry: hunger or gas pains, too hot or cold, overstimulated, need to suck or be held
- ❑ timing: onset 10 days to 3 months of age; peak 6-8 weeks
- ❑ average 40-120 minutes/day for first 3 months
- ❑ child cries, pulls up legs and passes gas soon after feeding

Notes

❑ suggestions for management
- parental relief, rest and reassurance (it is not their fault!)
- hold baby, soother, car ride, music, vacuum, check diaper
- drugs (ovol drops, ancatropine) are of little benefit
- elimination of cow milk protein from mother's diet (effective in small percentage of cases)

CHILD INJURY PREVENTION

Injuries
❑ not accidents - predictable and preventable
❑ leading cause of death from 1-44 years of age
❑ leading cause of potential years of life lost
❑ main causes of injury: motor vehicle, burns, drowning, suicide, falls

Newborn to 6 Months
❑ falls: do not leave infant alone on a bed, change table, in a bath; place in crib or playpen before answering phone or door; keep crib rails up
❑ burns: check water temperature before bathing, check milk temperature before feeding, do not hold cup of hot liquid and infant at same time
❑ sun exposure
❑ car seats, smoke and carbon monoxide detectors
❑ Poison Control Centre number next to telephone

6 to 12 Months
❑ stair barriers, discourage walkers
❑ plastic covers for electrical outlets, appliances unplugged when not in use
❑ keep small objects, plastic bags, and medications out of reach
❑ avoid play areas with sharp-edged tables and corners
❑ never leave unsupervised in tub

1 to 2 Years
❑ burns: turn pot handles to back of stove
❑ poisoning: keep drugs and cleaning products out of reach, Poison Control Centre number next to telephone, ipecac syrup in house
❑ choking: no nuts, raw carrots, orange segments, hot dogs, running while eating
❑ toddler seat at 20 lbs, fence around swimming pool
❑ watch for unsafe toys, balloons and plastic bags

2 to 5 Years
❑ street safety, bicycle helmet, seat belt and booster seat at 40 lbs
❑ stranger safety
❑ swimming lessons
❑ never leave child unsupervised at home, on driveway, in pool

IMMUNIZATION

Table 2. Immunization Schedule

Age	Vaccination	Route	Type	Contraindications
2 months	DTaP+IPV+Hib	IM	diptheria - toxoid pertussis - killed bacteria tetanus - toxoid polio - inactivated virus Hib - conjugated to diphtheria	previous anaphylaxis to vaccine; defer if progressive, evolving, unstable neurologic disease relative contraindication if child becomes hypotonic or hyporesponsive after vaccine
4 months	DTaP+IPV+Hib	IM		
6 months	DTaP+IPV+Hib	IM		
12 months	MMR	SC	live attenuated viruses	immunocompromise (but healthy HIV positive children should receive MMR vaccine); within 3 months of immunosuppressive therapy; pregnancy
18 months	DTaP+IPV+Hib	IM		
4-6 years	MMR DTaP+IPV	SC IM		 no Hib after age 7
grade 7 (in Ontario)	Hepatitis B vaccine in 3 doses	IM	purified HBsAg	
14-16 years and q 10 years thereafter	TdP	IM		immunodeficiency; pregnancy

Administration of Vaccines

- ❑ injection site
  - infants (<12 months old): anterolateral thigh
  - children: deltoid
- ❑ DTaP+IPV+Hib: these five vaccines are given as one IM injection (Pentacel)
- ❑ oral polio vaccine is available and used in some provinces, but not in Ontario

Contraindications to Any Vaccine

- ❑ moderate to severe illness +/– fever
- ❑ allergy to vaccine component (e.g. egg)

Possible Adverse Reactions to Any Vaccine

- ❑ local: induration or tenderness
- ❑ systemic: fever, rash
- ❑ allergic: urticaria, rhinitis, anaphylaxis

Possible Adverse Reactions to Regular Vaccines

- ❑ DTaP+IPV
  - minor: fever, local redness, swelling, irritability
  - major: prolonged crying (1%), hypotonic unresponsive state (1:1750), seizure (1:1950)
  - prophylaxis: acetaminophen 10-15 mg/kg 4 hours prior to injection and q4h afterwards
- ❑ Hib
  - safe; almost no reaction
- ❑ MMR
  - fever, measle-like rash in 7-14 days, lymphadenopathy, arthralgia, arthritis, parotitis
- ❑ TdP
  - anaphylaxis

TB Skin Test (Mantoux)

- ❑ screen high risk populations only (HIV, from foreign country with increased incidence, substance abuse in family, homeless, aboriginal)
- ❑ evidence against screening healthy populations

- ❑ intradermal injection (do not administer with MMR vaccine)
- ❑ positive result (TB-positive)
  - > 15 mm: children > 4 years with no risk factors
  - > 10 mm: children < 4 years, environmental exposure
  - > 5 mm: children with close TB contact, immunosuppressed
- ❑ BCG history irrelevant - does not usually give positive response
- ❑ positive reaction means active disease or previous contact

DELAYED IMMUNIZATION

Table 3. Delayed Immunization Schedule

Unimmunized Children Aged 1-6 Years		
Visit	**Vaccine**	**Notes**
initial visit	DTaP + Hib, MMR	no pertussis after age 7
2 months after first visit	DTaP	
2 months after second visit	DTaP	
12 months after third visit	DTaP	
4-6 years old	DTaP, MMR	
grade 7	Hepatitis B (0,1,6 months)	in Ontario
14-16 years old	TdP	
Unimmunized Children Aged 7 years and Over		
Visit	**Vaccine**	**Notes**
initial visit	TdP, MMR	
2 months after first visit	TdP	
6-12 months after second visit	TdP	
q 10 years thereafter	Td	no polio

OTHER VACCINES

BCG vaccine
- ❑ infants of parents with infectious TB at time of delivery
- ❑ groups/communities with high rates of disease/infection
- ❑ offered to aboriginal children on reserves

Pneumovax
- ❑ protects against 23 serotypes of S. *pneumoniae*
- ❑ for children with HIV or splenectomized children; e.g. sickle cell disease, splenic dysfunction, thalassemia
- ❑ for these high risk groups, give vaccine at 2 years of age, then revaccinate 3-5 years after initial dose

Influenza A
- ❑ given annually in the fall since strains vary from year to year
- ❑ for children with severe or chronic disease, e.g. cardiac, pulmonary, or renal diseases, sickle cell disease, diabetes, endocrine disorders, HIV, immunosuppressed, long-term aspirin therapy, residents of chronic care facilities
- ❑ contraindicated if allergic to eggs or < 6 months of age

Hepatitis B
- ❑ now recommended routinely in Canada
- ❑ set of 3 vaccinations given in mid-childhood to early teens (0, 1, 6 months)
- ❑ given in Grade 7 in Ontario schools (given at different grades in other provinces)
- ❑ if mother is HBsAg +ve, then give HBIG + vaccine at birth, and vaccine at 1 and 6 months

Varivax
- ❑ live attenuated varicella virus vaccine protects against chicken pox
- ❑ must be stored at -15ºC
- ❑ can be given after age 12 months (1 dose = 0.5 ml subcutaneous injection)
- ❑ after age 13, give two doses 4-8 weeks apart
- ❑ seroconversion rates of > 95% (20-30% yearly loss of antibody over 6 years); likely lifelong immunity, but longer studies are as yet unavailable

- ❑ mild local reactions in 5-10% (higher in immunocompromised)
- ❑ efficacy: protection rate is > 90%
- ❑ benefits
  - avoid chicken pox (5-7 days of fever, itchy rash, malaise, possible bacterial superinfection, encephalitis or pneumonia) (see Colour Atlas J1)
  - milder illness if chicken pox does develop
  - avoid parental cost of being off work or hiring babysitter
- ❑ costs $65-75, currently not covered by many drug plans
- ❑ contraindicated in pregnant women and in women planning to get pregnant in the next 3 months

DEVELOPMENTAL MILESTONES

Table 4. Developmental Milestones

Age	Gross Motor	Fine Motor	Speech and Language	Adaptive and Social Skills
6 weeks	prone-lifts chin intermittently			social smile
2 months	prone-arms extended forward	pulls at clothes	coos	
4 months	prone-raises head + chest, rolls over F —> B, no head lag	reach and grasp, objects to mouth	responds to voice	
6 months	prone-weight on hands, tripod sit	ulnar grasp	begins to babble, responds to name	stranger anxiety
9 months	pulls to stand	finger-thumb grasp	mama, dada - appropriate, imitates 1 word	plays games separation anxiety
12 months	walks with support, “cruises”	pincer grasp, throws	2 words with meaning besides mama, dada	plays peek-a-boo, drinks with cup
15 months	walks without support	draws a line	jargon	points to needs
18 months	up steps with help	tower of 3 cubes, scribbling	10 words, follows simple commands	uses spoon, points to body parts
24 months	up 2 feet/step, runs, kicks ball	tower of 6 cubes, undresses	2-3 words phrases uses “I”, “Me”, “you” 25% intelligible	parallel play, helps to dress
3 years	tricycle, up 1 foot/step, down 2 feet/step, stands on one foot, jumps	copies a circle and a cross, puts on shoes	prepositions, plurals, 75% intelligible, knows sex, age	dress/undress fully except buttons, counts to 10
4 years	hops on 1 foot, down 1 foot/step	copies a square, uses scissors	tells story, normal dysfluency, speech intelligible	cooperative play, toilet trained, buttons clothes
5 years	skips, rides bicycle	copies a triangle, prints name, ties shoelaces	fluent speech, future tense, alphabet	knows 4 colours

Table 5. Primitive Reflexes

Reflex	Appears	Disappears
grasp	birth	1-4 months
Moro	birth	3-4 months
rooting/sucking	birth	3-4 months
stepping/placing	birth	2-5 months
Galant	birth	2-3 months
tonic neck (“fencing”)	birth	2-3 months

Moro Reflex
- ❑ elicited by placing infant supine, head supported by examiner's hand, sudden withdrawal of support, head allowed to fall backward
- ❑ reflex is abduction and extension of the arms, opening of the hands, followed by adduction of the arms as if in an embrace
- ❑ absence of Moro suggests CNS injury
- ❑ asymmetry of Moro suggests focal motor lesions, e.g. brachial plexus injury or fracture of clavicle or humerus

Galant's Reflex
- ❑ stroking one side of the back along paravertebral line results in lateral curvature of the trunk toward the stimulated side

NORMAL PHYSICAL GROWTH
- ❑ newborn size influenced by maternal factors (placenta, in utero environment)
- ❑ premature infants: use corrected age until 2 years
- ❑ not linear: most rapid growth during first two years; growth spurt at puberty
- ❑ different tissue growth at different times
  - first two years: CNS
  - mid-childhood: lymphoid tissue
  - puberty: genital tissues
- ❑ body proportions: upper/lower segment ratio
  - newborn 1.7; adult male 0.97; female 1.0
  - increased ratio: achondroplasia, short limbs, hypothyroidism
  - decreased ratio: Marfan Syndrome

Weight Gain
- ❑ birth weight: 3-4.5 kg
- ❑ some weight loss after birth (maximum 10%); birthweight regained by 10 days
- ❑ 2x birth weight by 4-5 months; 3x birth weight by 1 year; 4x birth weight by 2 years
- ❑ half adult weight at 10 years

Linear Growth
- ❑ birth length: 50 cm
- ❑ 75 cm at 1 year, 87 cm at 2 years (half adult height); 93 cm at 3 years
- ❑ measure length until 2 years of age, then measure height

Head Circumference
- ❑ birth HC: 35 cm
- ❑ increase 2 cm/month for first 3 months, then 1 cm/month for 3-6 months, then 0.5 cm/month for 6-12 months

Dentition
- ❑ primary dentition (20 teeth)
  - first tooth at 5-9 months (lower incisor), then 1 per month to 20 teeth
  - 6-8 central teeth by 1 year
- ❑ secondary dentition (32 teeth)
  - first adult tooth is 1st molar at 6 years
  - 2nd molars at 12 years, 3rd molars at 18 years

FAILURE TO THRIVE (FTT)
- ❑ definition: weight < 3rd percentile, or falls across two major percentile curves, or < 80% of expected weight for height and age
- ❑ 50% organic, 50% non-organic
- ❑ inadequate caloric intake most important factor in poor weight gain
- ❑ energy requirements
  - 0-10 kg: 100 kcal/kg/day
  - 10-20 kg: 1000 cal + 50 cal/kg/day for each kg > 10
  - 20 kg+: 1500 cal + 20 cal/kg/day for each kg > 20
- ❑ may have other nutritional deficiencies, e.g. protein, iron, vitamin D deficiency

Approach to a Child with FTT
- ❑ history
  - detailed dietary and feeding history
  - pregnancy, birth, and postpartum history
  - developmental and medical history, including medications
  - social and family history (parental height and weight)

- assess 4 areas of functioning: child's temperament, child-parent interaction, feeding behaviour and parental psychosocial stressors

❑ physical examination
- height, weight, HC, arm span, upper:lower segment ratio
- assessment of nutritional status, dysmorphism, pubertal status
- observation of a feeding session and parent-child interaction
- signs of neglect or abuse

❑ laboratory investigations: as indicated by clinical presentation
- CBC, smear, electrolytes, urea, ESR, T4, TSH, urinalysis
- bone age x-ray
- karyotype in all short girls and in short boys where appropriate
- any other tests indicated from history and physical exam: e.g. renal or liver function tests, venous blood gases, ferritin, immunoglobulins, sweat chloride, fecal fat

❑ organic cause: usually apparent on full history and physical exam
❑ non-organic cause: often no obvious diagnosis from history and physical exam

Causes of Organic FTT
❑ inadequate intake
❑ inadequate absorption
❑ inappropriate utilization of nutrients
❑ increased energy requirements
❑ decreased growth potential

Causes of Non-Organic FTT
❑ inadequate nutrition, poor feeding technique, errors in making formula
❑ emotional deprivation, poor parent-child interaction, dysfunctional home
❑ child abuse and/or neglect
❑ parental psychosocial stress, childhood abuse and/or neglect
❑ treatment: most are managed as outpatients with multidisciplinary approach
- primary care physician, dietitian, psychologist, social work, child protection services

SHORT STATURE

Assessment of Short Stature
❑ height << 3rd percentile, height crosses 2 major percentile lines, low growth velocity (< 25th percentile)
❑ history: perinatal history, growth pattern, medical history, parental height and age of pubertal growth spurt
❑ physical exam: growth velocity (over 6 month period), sexual development (see Failure to Thrive Section)
❑ calculate Mid-Parental Height (predicted adult height) +/– 8 cm for 2 SD range
- boy = [father height (cm) + mother height (cm) + 13 cm]/2
- girl = [father height (cm) + mother height (cm) – 13 cm]/2

❑ true growth hormone deficiency is rare; associated with other congenital anomalies (midline defects, vocal abnormalities, micropenis, height affected more than weight)

Table 6. Short Stature

NORMAL GROWTH VELOCITY (non-pathological short stature)	DECREASED GROWTH VELOCITY (pathological short stature)
❑ constitutional (delayed bone age); delayed adolescence and may have family history of delayed puberty, may require treatment with androgen/estrogen short-term	❑ primordial (height, weight, and HC are affected) - chromosomal (e.g. Turner, Down syndrome, dysmorphic features) - skeletal dysplasias - IUGR (teratogen, placenta, infection)
❑ familial (normal bone age) (no treatment helpful)	❑ endocrine (height more affected than weight) - "short and fat" - growth hormone deficiency - hypothyroidism - Cushing's syndrome - hypopituitarism
	❑ chronic disease (weight more affected than height) - "short and skinny" - Celiac disease, IBD, CF - chronic infections - chronic renal failure (often height more affected)
	❑ psychosocial neglect (psychosocial dwarfism) - usually decreased height and weight (and HC if severe)

Investigations
❑ bone age x-ray
❑ karyotype in girls to rule out Turner syndrome
❑ other tests as indicated by history and physical

Management
❑ no treatment for the short normal child
❑ criteria for growth hormone (GH) therapy:
- GH has been shown to be deficient by physiological and pharmacological tests (2 required)
- patient is short (below 3rd percentile) and not growing
- x-rays show that there is still growth potential, with low growth velocity
- no etiological factor found that can be fixed
- signs and symptoms of GH deficiency - e.g. infantile features and fat distribution, hypoglycemia, prolonged hyperbilirubinemia in the newborn period, delayed puberty

❑ other endocrine abnormalities that are contributing to short stature should be corrected (e.g. thyroid hormone for hypothyroidism, insulin for diabetes)

TALL STATURE

❑ also constitutional and familial variants
❑ assessment
- history and physical examination: differentiate familial from other causes
- calculate Mid-Parental Height (predicted adult height)
- look for associated abnormalities (e.g. hyperextensible joints in Marfan syndrome)

❑ etiology
- constitutional: most common, advanced bone age/physical development in childhood but normal once adulthood reached
- endocrine: e.g. hypophyseal (pituitary) gigantism, precocious puberty, thyrotoxicosis, Beckwith-Wiedeman syndrome
- genetic: e.g. Marfan, Klinefelter syndromes

❑ treatment: depends on etiology
- estrogen used in females to cause epiphyseal fusion

OBESITY

- ❑ weight > 20% greater than expected for age and height
- ❑ history: diet, activity, family heights and weights, growth curves
- ❑ physical examination: may suggest secondary cause, e.g. Cushing's syndrome
  - caliper determination of fat is more sensitive than weight
- ❑ organic causes are rare (< 5%)
  - genetic, e.g. Prader-Willi, Carpenter, Turner syndrome
  - endocrine, e.g. Cushing's, hypothyroidism
- ❑ complications
  - low correlation between obese children and obese adults
  - some association with: hypertension, increased LDL, increased acute respiratory infection, slipped capital femoral epiphysis
  - may predispose to adult hypertension, diabetes, cardiovascular disease
  - boys: gynecomastia
  - girls: polycystic ovarian disease, early menarche
  - psychological: discrimination, teasing, isolation, decreased self-esteem, treated as stupid or inferior
- ❑ management
  - encouragement and reassurance
  - diet: qualitative changes; do not encourage weight loss but allow for linear growth to catch up with weight
  - evidence against very low kilojoule diets for preadolescents
  - behavior modification: increase activity, change meal patterns
  - insufficient evidence for or against exercise, family programs for obese children
  - education: multidisciplinary approach, dietitian, counselling

CHILD ABUSE AND NEGLECT

Definition

- ❑ an intentional act of commission or omission (physical, sexual, or emotional) by another person that harms a child in a significant way

Legal Obligation to Report

- ❑ upon suspicion of abuse, physicians in Canada are required by law to call the Children's Aid Society (CAS)

Risk Factors

- ❑ family factors
  - social isolation
  - poverty
  - stressful life events or situation
  - domestic violence
- ❑ caregiver factors
  - parents were abused as children (most commonly associated)
  - psychological dysfunction / psychiatric illness
  - substance abuse
  - parenting style
  - poor social and vocational skills, below average intelligence
- ❑ child factors
  - difficult child (temperament)
  - handicap or disability
  - special needs, e.g. mental retardation

Physical Abuse

- ❑ history inconsistent with physical findings
- ❑ “doctor shopping”, multiple visits to different hospitals
- ❑ delay in seeking medical attention
- ❑ injuries of varied ages, recurrent or multiple injuries
- ❑ distinctive marks: e.g. belt buckle, cigarette burns, hand
- ❑ atypical patterns of injuries: face, abdomen, buttocks, inner thighs, upper back, symmetrical pattern
- ❑ altered mental status: head injury, drug ingestion, poisoning

- ❑ shaken baby syndrome
  - most common cause of severe closed head injury in infants < 1 year old
  - violent shaking of infant resulting in intracranial hematomas and retinal hemorrhages
  - diagnosis confirmed by CT or MRI
  - poor prognosis for infants presenting in coma: 50% die, 25% have significant neurologic damage

Sexual Abuse

- ❑ prevalence: 1 in 4 females, 1 in 10 males
- ❑ peak ages at 2-6 and 12-16 years
- ❑ most perpetrators are known to child
  - most common: father, stepfather, uncle
- ❑ diagnosis usually depends on child telling someone
- ❑ clinical signs
  - specific or generalized fears, depression
  - social withdrawal, lack of trust
  - psychosomatic symptoms, school failure
  - sexual preoccupation, play
  - behavior: seductive, acting out, aggressive, pseudomature
  - recurrent UTIs, pregnancy, STDs, vaginitis, vaginal bleeding, genital injury
- ❑ investigations depend on presentation, age, sex, and maturity of child
  - up to 72 hours: rape kit
  - R/O STD, UTI, pregnancy (consider STD prophylaxis or morning after pill)
  - R/O other injuries

Neglect

- ❑ failure to thrive, developmental delay
- ❑ inadequate or dirty clothing, chronic lack of personal hygiene
- ❑ child exhibits poor attachment to parents

Management of Child Abuse and Neglect

- ❑ history: from child and caregiver(s)
- ❑ physical exam: head to toe (do not force), emotional state, development
- ❑ document all injuries: type, location, size, shape, colour, pattern
- ❑ report all suspicions to CAS and/or police
- ❑ acute medical care; hospitalize if indicated or if concerns about further or ongoing abuse
- ❑ investigations: bloodwork, throat and/or genital swabs, skeletal survey, bone scan, CT/MRI, photos
- ❑ arrange consultation to social work, psychiatry
- ❑ arrange appropriate follow-up
- ❑ D/C directly to CAS or to responsible guardian under CAS supervision

DEVELOPMENTAL DELAY

Differential Diagnosis

- ❑ chromosomal: Down syndrome, trisomy 13, trisomy 18
- ❑ metabolic: Tay-Sachs, PKU, adrenoleukodystrophies
- ❑ cerebral degenerative: Huntington's chorea, SSPE
- ❑ prenatal infection: TORCHS, HIV
- ❑ postnatal infection: meningitis, encephalitis, HIV
- ❑ toxic agents/drugs: alcohol, street drugs
- ❑ trauma/hypoxia: birth trauma, intracerebral hemorrhage
- ❑ other syndromes: cerebral malformations, neurofibromatosis, autism
- ❑ sensory defects: vision, hearing

LANGUAGE DELAY

Differential Diagnosis

- ❑ hearing impairment
  - not responsive to sounds out of sight
  - prelinguistic skills (e.g. cooing, babbling) may initially develop normally but may decrease due to lack of feedback
  - no impairment in social interaction
  - causes
    - genetic (30-50%)
    - congenital infection (e.g. rubella, CMV)
    - meningitis
    - ototoxic medications (e.g. aminoglycosides)
- ❑ cognitive disability
  - global developmental delay, mental retardation
  - both receptive and expressive language components affected
  - child often has interest in communication
- ❑ pervasive developmental disorder (including autism)
  - poor social interaction and language impairment, especially expressive (see Pervasive Developmental Disorder Section)
- ❑ selective mutism
  - only speaks in certain situations, usually at home
  - usually starts at age 5-6 years when child goes to school
  - healthy children with no hearing impairment
  - often above average intelligence
- ❑ Landau-Kleffner syndrome (acquired epileptic aphasia)
  - presents in late preschool to early school age years
  - child begins to develop language normally, then sudden regression of language
  - child has severe aphasia with EEG changes
  - often has overt seizure activity
  - initial presentation may be similar to autism
- ❑ mechanical problems
  - cleft palate
  - cranial nerve palsy
- ❑ social deprivation

PERVASIVE DEVELOPMENTAL DISORDER (PDD)

- ❑ broad generic term which describes a spectrum of related disorders, including autism, Asperger's syndrome, child disintegrative disorder, and PDD not otherwise specified
- ❑ autism
  - prevalence M:F = 4:1
  - risk in sibling 8-9%
  - onset prior to 3 years of age
- ❑ Asperger's syndrome
  - prevalence M>F
  - impaired social interaction
  - language and cognition better than in autism
  - restricted, repetitive, stereotyped patterns of behaviour, interests and activities
  - better prognosis than in autism
- ❑ 4 main areas of functioning affected

- ❑ 1) lack of reciprocal social interaction
  - lack of interest in peers and poor group participation
  - higher functioning individuals with PDD lack depth in their interactions with people: inflexibility, lack of reciprocity and empathy
- ❑ 2) problems with verbal and non-verbal communication
  - delay in onset of expressive and receptive language
  - characteristics of autism: echolalia, perseveration, abnormalities in volume, pitch and rate of speech
- ❑ 3) restricted and repetitive behaviours
  - stereotypic: hand-flapping, head-banging, rocking, repetitive finger movements, spinning, etc.
  - ritualistic: checking, touching
- ❑ 4) abnormal cognitive function
  - majority exhibit mental retardation
  - may have good memory and visuospatial function
  - poor symbolization and understanding of abstract ideas and theoretical concepts
  - higher functioning PDD children may have consuming interest in one topic to the exclusion of other topics

FETAL ALCOHOL SYNDROME (FAS) AND FETAL ALCOHOL EFFECTS (FAE)

- ❑ prevalence
  - FAS: 1 in 500-600
  - FAE: 1 in 300-350
- ❑ not known how much alcohol is harmful during pregnancy
- ❑ no "safe" level of alcohol consumption during pregnancy

Criteria for Diagnosis of Fetal Alcohol Syndrome

- ❑ A: Growth deficiency
  - low weight and/or short length at birth that continues through childhood
- ❑ B: Abnormal craniofacial features
  - small head, small eyes, long smooth philtrum, thin upper lip, maxillary hypoplasia
- ❑ C: Central nervous system dysfunction
  - microcephaly and/or neurobehavioral dysfunction (e.g. hyperactivity, motor problems, attention deficits, learning disabilities, cognitive disabilities)
- ❑ D: Strong evidence of maternal drinking during pregnancy

Fetal Alcohol Effects

- ❑ child born to a mother who was known to be drinking heavily during pregnancy
- ❑ child has some but not all of physical characteristics of FAS

CHRONIC RECURRENT ABDOMINAL PAIN

- ❑ prevalence: 10% of school children
  - common in early childhood and early adolescence
- ❑ < 10% have organic disease
- ❑ characteristics of psychogenic abdominal pain
  - seldom wakes child
  - poorly localized, periumbilical, constant
  - aggravated by exercise, alleviated by rest
  - school avoidance
  - psychosocial factors related to onset and/or maintenance of pain
  - absence of organic illness
- ❑ psychiatric comorbidity: anxiety, somatoform, mood, learning disorders, sexual abuse, eating disorders, elimination disorders
- ❑ assessment: interview child alone and with parents, R/O organic illness
- ❑ management
  - identify psychosocial stressors
  - individual and family psychotherapy

ELIMINATION DISORDERS

ENURESIS

- ❑ involuntary urinary incontinence by day and/or night in a child > 5 years old
- ❑ not due to neurological disorder resulting in poor bladder control, epilepsy, or structural abnormality of the urinary tract
- ❑ prevalence: 10% of 6 year olds, 3% of 12 year olds, 1% of 18 year olds

Primary Nocturnal Enuresis (90%)

- ❑ wet only at night during sleep
- ❑ developmental disorder or maturational lag in bladder control while asleep
- ❑ more common in boys, family history common
- ❑ investigations: urinalysis
- ❑ treatment
  - time and reassurance (~20% resolve spontaneously each year)
  - bladder retention exercises
  - conditioning: "wet" alarm wakes child upon voiding (40-75% success rate)
  - medications: DDAVP

Secondary Enuresis

- ❑ develops after child has sustained (3 months or more) period of bladder control
- ❑ nonspecific regression in the face of stress or anxiety, e.g. birth of sibling, significant loss, family discord
- ❑ may be secondary to UTI, DM, DI, neurogenic bladder, CP, sickle cell disease, seizures, pinworms
- ❑ may occur if engrossed in other activities

Diurnal Enuresis

- ❑ daytime wetting (60-80% also wet at night)
- ❑ timid, shy, temperamental problems
- ❑ R/O structural anomaly, e.g. ectopic ureteral site, neurogenic bladder
- ❑ treatment depends on cause
  - remind child to go to toilet
  - mental health treatment
  - focus on verbal expression of feelings

ENCOPRESIS

- ❑ fecal incontinence in a child at least 4 years of age
- ❑ prevalence: 1-1.5% of school aged children (rare in adolescence)
- ❑ M:F = 6:1
- ❑ must exclude medical causes, e.g. Hirschsprung's disease, hypothyroidism, hypercalcemia, spinal cord lesions, anorectal malformations

Retentive Encopresis (psychogenic megacolon)

- ❑ causes
  - physical: anal fissure (painful stooling)
  - emotional: disturbed parent-child relationship, coercive toilet training
  - genetic: 75% have enuretic relative, MZ > DZ twins
- ❑ history
  - child withholds bowel movement, develops constipation, leading to fecal impaction and seepage of soft or liquid stool
  - crosses legs to resist urge to defecate
  - distressed by symptoms, soiling of clothes
  - toilet training: coercive or lackadaisical
- ❑ physical exam
  - rectal exam: large fecal masses in rectal vault
- ❑ treatment
  - clean out bowel completely (e.g. Golytely, fleet enemas)
  - stool softeners (e.g. Senokot, Lansoyl at bedtime)
  - enemas and suppositories
  - regular schedule to defecate
  - positive reinforcement

Non-Retentive Encopresis

- ❑ continuous: present from birth (never gained primary control of bowel function)
  - bowel movement randomly deposited without regard to social norms
  - family structure usually does not encourage organization and skill training
  - child has not had adequate consistent bowel training
  - treatment: consistent, firm and kind toilet training
- ❑ discontinuous: previous history of normal bowel control
  - bowel movements as an expression of anger or wish to be seen as a younger child
  - breakdown occurs in face of stressful event, regression
  - displays relative indifference to symptoms
  - treatment: psychotherapy if persists for many weeks

Toilet Phobia

- ❑ relatively young child
- ❑ views toilet as a frightening structure
- ❑ child thinks they may be swept away by toilet
- ❑ treatment
  - gradual series of steps with rewards
  - desensitization

GENETICS

APPROACH TO THE DYSMORPHIC CHILD

- ❑ 3/100 infants are born with a congenital defect, many are associated with a degree of developmental disability
- ❑ genetic disorders and birth defects account for approximately 40% of childhood deaths
- ❑ history
  - prenatal/obstetrical history: maternal age and past health, alcohol/drug/meds use, difficulties during pregnancy/labour/delivery, investigations done and results (see Obstetrics Notes)
  - complete 3 generation family pedigree: consanguinity, stillbirths, neonatal deaths, specific illnesses, mental retardation, multiple miscarriages, ethnicity (thalassemia, Tay-Sachs)
  - developmental milestones and growth in an older child
- ❑ physical examination
  - careful observation
  - growth parameters (height/weight/head circumference)
  - compare child's features with parents and sibs
- ❑ investigation
  - ask for serial photographs if child is older
  - x-rays if bony abnormalities or if suspect a congenital infection
  - cytogenetic/chromosome studies +/– skin fibroblasts
  - biochemistry: specific enzyme assays
  - molecular biology for specific testing
  - genetic probes now available e.g. Fragile X
- ❑ counselling and recurrence risk assessment

Patterns of Inheritance

- ❑ autosomal dominant
  - 50% risk with an affected parent
  - e.g. Neurofibromatosis I and II, Marfan syndrome, Achondroplasia
- ❑ autosomal recessive
  - risk is 25% when both parents carry the affected gene
  - carrier states can sometimes be detected; consanguinity increases chance
  - e.g. sickle cell anemia, CF, Tay-Sachs
- ❑ X-linked recessive
  - gene for the disease carried on X chromosome, inherited through mother; most are recessive with homozygous females being rare
  - female carriers may sometimes be detected, e.g. G6PD deficiency
  - cannot have male to male transmission
  - e.g. Duchenne MD, Fragile X, G6PD, Hemophilia A and B

- ❑ multifactorial
  - genetic predisposition with environmental factors required for disease to be expressed
  - recurrence risk 4-10% (disease specific) ; if mother and one child affected, risk is up to 15%
  - e.g. neural tube defects, cleft lip and palate
- ❑ mitochondrial
  - genes from mother only; M=F
  - e.g. Leber optic neuropathy, MELAS
- ❑ spontaneous mutations

DOWN SYNDROME

- ❑ in humans, the most common abnormality of autosomal chromosomes
- ❑ trisomy 21
  - 80-90% nondisjunction
  - 5% translocations
  - 3% mosaics (may be less noticeable/less severe)
- ❑ incidence: most common autosomal chromosomal abnormality, 1 in 600-800 live births, rises with advanced maternal age to 1 in 20 by age 45 years
- ❑ affected fetuses have increased risk of spontaneous abortion
- ❑ clinical features
  - hypotonia at birth (80%), low IQ, developmental delay
  - neurologic: hypotonia, premature senility, Alzheimer's onset in 40's
  - facies: flat occiput, microcephaly, small midface, small mandible and maxillae, upslanting palpebral fissures, epicanthal folds, Brushfield's spots in iris
  - ENT: furrowed prominent tongue, high arched palate, ear anomalies, frequent acute otitis media
  - CVS: 40% have congenital cardiac defects, particularly endocardial cushion defects
  - GI: duodenal, anal atresia and TE fistula
  - MSK: lax joints including dysplastic hips, vertebral anomalies, atlantoaxial instability
  - skin: Simian (palmar) crease, abnormal dermatoglyphics
  - hematologic: leukemias (1% lifetime risk)
  - endocrine: hypothyroidism
- ❑ prognosis: shorter life expectancy
- ❑ management
  - recommended testing: echo, thyroid tests, atlanto-occipital x-ray at 2 years (controversial)
  - treat any life-threatening defects immediately (e.g. duodenal atresia)
  - mainly symptomatic
  - wide range of severity, early intervention programs to help children reach full potential

OTHER TRISOMIES

Trisomy 13

- ❑ incidence 1:5000 live births
- ❑ increased risk of spontaneous abortions
- ❑ features: seizures, deafness, microcephaly, cleft lip/palate, polydactyly, retinal anomalies, single umbilical artery, cardiac defects, scalp defects
- ❑ midline anomalies: scalp, pituitary, palate, heart, umbilicus, anus
- ❑ prognosis: 44% die in 1 month
  < 10% survive past 1 year (profound MR in survivors)

Trisomy 18

- ❑ incidence: 1/8000 live births, female: male = 3:1
- ❑ increased risk of spontaneous abortion
- ❑ features: prominent occiput, micrognathia, ocular abnormalities, cleft lip and palate, low set ears, rocker bottom feet, short stature, clenched fist with overlapping digits, hypoplastic nails, clinodactyly, polydactyly, cardiac defects, hernia, severe CNS malformation, urogenital abnormalities (cryptorchidism, polycystic kidneys)
- ❑ key point: small babies (SGA, microcephaly, short)
- ❑ prognosis of severe FTT: 33% die in 1 month, 50% by 2 months, 90% by 12 months, profound MR in survivors

TURNER SYNDROME

- ❑ most common genotype is 45X; mosaic also possible with most common being (45X/46XX)
- ❑ incidence 1:2,500 live female births
- ❑ risk not increased with advanced maternal age
- ❑ clinical features
  - intelligence usually normal, may have mild learning disabilities
  - lymphedema, cystic hygroma in the newborn with polyhydramnios, lung hypoplasia
  - short stature, wide carrying angle at elbows
  - short webbed neck, low posterior hair line
  - broad chest, wide spaced nipples
  - infertility, gonadal dysgenesis
  - primary amenorrhea, lack of development of secondary sexual characteristics
  - heart defects: coarctation of the aorta, bicuspid aortic valve
  - renal abnormalities, increased risk of HTN
- ❑ prognosis: normal life expectancy if no complications; risk of X-linked diseases increases to that of males
- ❑ management
  - to facilitate growth and development of secondary sexual characteristics
  - hormone/estrogen replacement
  - growth hormone (controversial)

KLINEFELTER SYNDROME

- ❑ 1/1,000 live male births, 47 XXY (most common)
- ❑ associated with late maternal age
- ❑ doesn't present until male post-pubertal
- ❑ mild mental retardation, long limbs, hypogonadism, hypospermia gynecomastia, lack of facial hair
- ❑ treatment: testosterone in adolescence

FRAGILE X

- ❑ most common genetic cause of developmental delay in boys
- ❑ incidence 1/1250; X-linked recessive
- ❑ clinical features
  - overgrowth: prominent jaw, forehead, ears; elongated, narrow face; marcroorchidism
  - hyperextensibility, high arched palate, mitral valve prolapse
  - often hyperactive and/or autistic
  - IQ typically 30-65 but 20% of affected males have normal intelligence
  - female carriers may show some intellectual impairment
- ❑ diagnosis
  - cytogenetic studies: region on Xq which fails to condense during mitosis
  - molecular testing: overamplification of a trinucleotide repeat, length of segment is proportional to severity of clinical phenotype (genetic anticipation)

MUSCULAR DYSTROPHY

- ❑ a group of inherited diseases characterized by progressive skeletal (+ cardiac) muscle degeneration

Duchenne Muscular Dystrophy

- ❑ X linked recessive, 1/3000 males, 1/3 spontaneous mutations
- ❑ missing structural protein dystrophin, leads to muscle fibre fragility, fibre breakdown, necrosis and regeneration
- ❑ clinical features
  - by age 3, proximal muscle weakness, Gower's sign
  - pseudo-hypertrophy of muscles
  - decreased reflexes
  - may develop mild mental retardation, obesity
- ❑ diagnosis
  - pedigree
  - creatine phosphokinase, lactate dehydrogenase increased
  - muscle biopsy, EMG
- ❑ complications
  - patient usually wheelchair bound by 12 years old
  - early flexion contractures, scoliosis
  - death due to pneumonia/respiratory failure or congestive heart failure

❑ treatment
- supportive (physiotherapy, wheelchairs, braces), prevent obesity
- surgical (for scoliosis)
- use of steroids experimental
- gene therapy trials underway

Becker's Muscular Dystrophy
❑ dystrophin gene abnormal, symptoms similar to Duchenne but onset is later and progression is slower

CLEFT LIP AND PALATE
❑ multi-factorial inheritance
❑ see ENT section

INBORN ERRORS OF METABOLISM
❑ an inherited disorder of intermediary metabolism
❑ treatment is sometimes possible because the biochemical basis of the disorder is understood
❑ presentation
- seizures, encephalopathy
- developmental delay, FTT
- renal tubular disease, diffuse liver disease
- hypoglycemia, hyperammonemia, wide anion gap metabolic acidosis

VACTERL ASSOCIATION
❑ number of congenital anomalies occuring together
❑ v=vertebral anomalies,a=imperforate anus, c=cardiac abnormalities, te=tracheoesophageal fistula, r= radial and renal dysplasia, l=limb deformity

NEONATOLOGY

INFANT MORTALITY
❑ 9-10/1,000 births
❑ causes
- congenital
- prematurity (RDS, intracranial hemorrhage)
- asphyxia
- infections
- sudden infant death syndrome

NORMAL BABY AT TERM
❑ HR 120-160/per min
❑ RR 40-60/per min
❑ weight 2500-4500 g
❑ glucose > 2.2
❑ BP systolic 50-80, diastolic 30-40 (dependent on GA)

GESTATIONAL AGE AND SIZE

Definitions
❑ gestational age
- pre-term: <37 weeks
- term: 37-42 weeks
- post-term: > 42 weeks

❑ SGA: measurements < 2 SD below mean for gestational age (GA)
❑ AGA: within 2 SD of mean for GA
❑ LGA: > 2 SD above the mean for GA
❑ GA can be estimated using the Ballard Score

Table 7. Infant Maturity			
Sites	< = 36 Weeks	37-38 Weeks	> = 39 Weeks
skin	pale, translucent	pinker, smoother	pink, thick
sole creases	smooth progresses to anterior creases	anterior progresses to heal creases	increasing depth of sole creases
breast nodule diameter	≤ 2 mm	4 mm	5-10 mm
scalp hair	fine and fuzzy	fine and fuzzy	thick and silky
ear lobe	flat, pliable, no cartilage	some cartilage	stiffened by thick cartilage
testes and scrotum	testes in lower canal, small scrotum, few rugae	intermediate scrotum full	pendulous, covered with rugae
labia and clitoris	prominent clitoris, small labia	clitoris nearly covered by prepuce	clitoris covered by prepuce large labia

Table 8. Abnormalities of Gestational Size and Maturity Features		
Features	Causes	Problems
pre-term infants < 37 weeks	infection (TORCH) maternal pathology drugs/EtOH chromosomal smoking multiple pregnancy infections placental causes	RDS, respiratory diseases recurrent apnea feeding difficulties hypocalcemia, hypoglycemia anemia jaundice intracranial hemorrhage, cerebral anoxia hypothermia edema NEC retinopathy of prematurity
SGA infants • asymmetric undergrowth: late onset, growth arrest	extrinsic causes: diabetes, nutrition, hypertension, multiple pregnancies, drugs, EtOH, smoking	asphyxia hypoglycemia hypocalcemia
• symmetric undergrowth: early onset, lower growth potential	intrinsic causes: infections (TORCH) meconium aspiration, chromosomal, genetic, congenital abnormalities, syndromal, idiopathic	hypothermia hyperviscosity (polycythemia) NEC PDA
LGA infants - large features	maternal DM, racial or familial factors	asphyxia, meconium aspiration, respiratory distress, TTN, PPH jaundice, hypoglycemia, hypocalcemia polycythemia, congenital abnormalities
post-term infants • wisened looking, leathery skin • meconium staining		severe asphyxia, meconium aspiration hypoglycemia birth trauma if large infant

NEONATAL RESUSCITATION

❑ How Ready Is This Child?

❑ Assess Apgar at 1, 5 minutes, if < 7 at 5 min then q 5 min

Table 9. Apgar Score			
Sign	0	1	2
Heart Rate	absent	< 100/minute	> 100/minute
Respiratory Effort	absent	slow, irregular	good, crying
Irritability	no response	grimace	cough or sneeze
Tone/Muscle	limp	some flexion of extremities	active motion
Color	blue, pale	body pink, extremities blue	completely pink

Initial Resuscitation
- ❑ always remember ABC's
- ❑ anticipation - know maternal history, history of pregnancy, labor, and delivery
- ❑ all infants
  - prevent heat loss by drying, warming (on radiant heater, remove wet towels)
  - position head and neck to open airway for suction
  - stimulate infant
- ❑ Airway
  - gentle suction of mouth then nose: < 100 mmHg, < 5 seconds
  - with thick meconium, suction the nasopharynx as the head is delivered, then intubate and suction trachea prior to first breath if possible
- ❑ Breathing
  - check for spontaneous respirations
  - bag and mask if apneic/gasping/HR < 100, bag at a rate of 40-60/minute with 90-100% O_2
  - intubation is indicated if
    - prolonged ventilation is required
    - bag and mask are not effective
    - tracheal suctioning is needed (thick meconium)
    - HR remains < 100
    - diaphragmatic hernia is suspected
- ❑ Circulation
  - heart rate is the most important indicator of the need for intervention
  - "80 or less compress" - if bradycardic (apex < 80 and no improvement with bagging) or asystolic, compressions begin at rate of 120/minute
  - coordinate 3 compressions with 1 ventilation (120 compressions/minute, 40 ventilations/minute) - check after 30 seconds
    - if HR > 80 stop compressions but continue ventilation
- ❑ Drugs
  - epinephrine - for asystole or severe bradycardia
  - HCO_3 (4.2% solution given slowly)
  - $CaCO_3$ - electrical abnormalities
  - Narcan - if mother given opioids, general anesthetic

ROUTINE NEONATAL CARE
- ❑ eye care - erythromycin ointment to prevent ophthalmia neonatorum - gonorrhea, chlamydia
- ❑ vitamin K - to avoid hemorrhagic disease of newborn
- ❑ HBIG plus vaccine if mother is Hep B +ve
- ❑ screening test
  - in all neonates: PKU, TSH usually after 24 hours of life
  - if indicated: blood group, sickle cell, G6PD deficiency (varies by province)
  - blood group and direct antiglobulin test if mother Rh-ve

RESPIRATORY DISTRESS IN THE NEWBORN

Presentation
- ❑ tachypnea > 60 / per min
- ❑ audible grunting
- ❑ intercostal retractions/indrawing
- ❑ nasal flaring
- ❑ duskiness/central cyanosis
- ❑ decreased A/E on auscultation
- ❑ tachycardia > 160 / per min

Diagnosis
- ❑ chest x-ray
- ❑ ABG, CBC, blood glucose
- ❑ blood cultures, Gram stain

Differential Diagnosis
- ❑ pulmonary
  - respiratory distress syndrome (RDS)
  - transient tachypnea of the newborn (TTN)
  - meconium aspiration (group B strep and others)
  - atelectasis

- pleural effusions
- pneumothorax
- congenital lung malformations

❑ cardiac
- congenital heart disease (cyanotic, obstructive, LR shunt)
- persistent pulmonary hypertension (PPHN)

❑ hematologic
- blood loss
- polycythemia

❑ infectious
❑ anatomic
- tracheoesophageal fistula
- congenital diaphragmatic hernia

❑ metabolic
- hypoglycemia
- inborn errors of metabolism

❑ neuromuscular
- CNS damage (trauma, hemorrhage)
- medication (maternal sedation)
- anomalies (e.g. Werdnig-Hoffmann disease)
- drug withdrawal syndromes

Upper Airway Obstruction
❑ Choanal Atresia
❑ Pierre-Robin syndrome
❑ laryngeal obstruction (stenosis, atresia, malacia)
❑ tracheal obstruction (mass, stenosis, malacia, vascular ring)
❑ mucous plug
❑ cleft palate

RESPIRATORY DISTRESS SYNDROME (RDS)
❑ also known as Hyaline Membrane Disease
❑ most common cause of respiratory distress in the pre-term infant

Pathophysiology
❑ surfactant deficiency —> poor lung compliance due to high alveolar surface tension and atelectasis —> respiratory distress—> hypoxia + acidosis
❑ surfactant decreases alveolar surface tension, lung compliance and functional residual capacity
❑ hypoxia, hypotension, and hypothermia may impair surfactant production/secretion

Risk Factors
❑ premature babies 5% risk @ 33 weeks, 65% risk @ 29 weeks
❑ infants of diabetic mothers (insulin inhibits the cortisol surge necessary for surfactant synthesis)
❑ C-section (reduced with antenatal steroids to mother)
❑ asphyxia, acidosis
❑ second of twins
❑ males:females = 2:1

Clinical Features
❑ onset within first few hours of life, worsens over next 24-72 hours, with symptoms of respiratory distress
❑ infants may develop edema, apnea, respiratory failure, and require ventilation
❑ chest x-ray: decreased aeration and lung volumes, reticulogranular pattern throughout lung fields with air bronchograms, atelectasis, may resemble pneumonia

Prevention
❑ minimize prematurity
❑ monitor L/S ratio
❑ steroid therapy (Celestone) for mothers 24 hours prior to delivery of premature infants

Treatment
❑ supportive: O_2, assist ventilation with PEEP or CPAP, fluids, nutrition
❑ surfactant administration (bovine or synthetic)

Prognosis
- ❑ self-limited disease, tends to improve after 72 hours without complications
- ❑ in severe prematurity and/or prolonged ventilation, increased risk of bronchopulmonary dysplasia

Complications
- ❑ PDA
- ❑ bronchopulmonary dysplasia
- ❑ retinopathy of prematurity
- ❑ pulmonary air leaks (pneumothorax)
- ❑ intracerebral/intraventricular hemorrhage

TRANSIENT TACHYPNEA OF THE NEWBORN (TTN)

- ❑ also known as
  - persistent postnatal pulmonary edema
  - "wet lung syndrome"
  - respiratory distress syndrome type II

Pathophysiology
- ❑ delayed resorption of fetal lung fluid —> accumulation of fluid in peribronchial lymphatics and vascular spaces —> tachypnea

Increased Risk In
- ❑ full term or slightly premature infants
- ❑ C-section babies (whose lungs are not compressed during passage through the pelvic floor)
- ❑ males

Clinical Features
- ❑ tachypnea within the first few hours of life (usually within the first 30 minutes); mild retractions, grunting, without signs of severe respiratory distress
- ❑ usually resolves in 24-72 hours
- ❑ chest x-ray: hazy lungs, fluid in fissures, increased vascularity, slight cardiomegaly

Treatment
- ❑ supportive: O_2, fluids, nutrition

MECONIUM ASPIRATION SYNDROME (MAS)

- ❑ 10-15% of all births are meconium stained, ~5% of meconium stained infants get MAS
- ❑ usually associated with fetal distress in utero, or post-term infant
- ❑ higher incidence with thick meconium
- ❑ respiratory distress within hours of birth - tachypnea, hypercarbia, small airway obstruction, chemical pneumonitis
- ❑ chest x-ray: hyperinflation, streaky atelectasis, patchy infiltrates
- ❑ complications: hypoxemia, acidosis, PPHN, 11% pneumothorax, 30% mechanical ventilation, 4% mortality
- ❑ treatment: supportive care and ventilation, may benefit from surfactant replacement as surfactant function is inhibited by meconium
- ❑ prevention: careful in utero monitoring, suction naso/oropharynx at perineum, then intubate and suction below cords at birth

PNEUMONIA

- ❑ consider in infants with prolonged rupture of membranes or maternal fever
- ❑ suspect if temperature unstable, WBC elevated, or neutropenic
- ❑ chest x-ray: hazy lung (as in TTN) + distinct infiltrates, normal lung volume

DIAPHRAGMATIC HERNIA

- ❑ Posterolateral or Anteromedial
- ❑ clinical features
  - respiratory distress, cyanosis
  - scaphoid abdomen
  - affected side dull to percussion and breath sounds absent; may hear bowel sounds instead
  - asymmetric chest movements, trachea deviated away from affected side
  - may present outside of neonatal period

- ❑ chest x-ray: portion of GI tract in thorax (usually left side), displaced mediastinum
- ❑ treatment: surgical
- ❑ prognosis: 50% survival overall
  - associated with a high incidence of pulmonary vascular anomalies, hypoplastic lungs

PERSISTENT PULMONARY HYPERTENSION (PPHN)

- ❑ R —> L shunt through PDA / foramen ovale / intrapulmonary channels, decreased pulmonary blood flow creates hypoxemia leading to further pulmonary vasoconstriction
- ❑ risk factors: abruption / placenta previa, asphyxia, MAS, RDS, sepsis, structural abnormalities (Potters / diaphragmatic hernia)
- ❑ treatment: O_2 given early, tapered slowly, minimize stress / hypoxia, if mechanical ventilation is unsuccessful, extracorpreal membrane oxygenation (ECMO) may be required

BRONCHOPULMONARY DYSPLASIA (BPD)

- ❑ usually after prolonged intubation/ventilation with high oxygen concentration (incidence with maturity)
- ❑ persistent respiratory distress
  - decreased compliance, increased resistance, pulmonary edema
  - hypoxemia, hypercapnia, may have apnea and bradycardia
- ❑ may have cardiac component (congestive heart failure)
- ❑ treatment: gradual weaning from ventilator, feed and grow, avoid stress, dexamethasone may help decrease inflammation and encourage weaning
- ❑ 15% mortality in severe cases

CYANOSIS OF THE NEWBORN

- ❑ central cyanosis means poor oxygenation - decreased SaO_2 decreased PaO_2
- ❑ peripheral cyanosis can be normal, or it could mean sepsis, temperature instability, congestive heart failure, vessel abnormalities
- ❑ Do ABGs if cyanosis seen in resting state/sleep after 30 min of life
- ❑ SaO_2 < 90% or PaO_2 < 60 mmHg = emergency
- ❑ hemoglobin abnormalities cause decreased SaO_2, normal PaO_2
- ❑ always check the pO_2 on 100% oxygen x 10-15 min (hyperoxic test)
  - if < 100 think congenital heart disease (see Pediatric Cardiology Section)
  - if > 100 think respiratory (airway, chest, lungs), brain or blood

Table 10. Differential Diagnosis of Cyanosis in the Newborn

Pulmonary
- see Neonatology Respiratory Distress Section

Cardiovascular
- see Pediatric Cardiology Section

Central Nervous System
- maternal sedative drugs
- asphyxia
- intracranial hemorrhage, intraventricular hemorrhage
- nerve-muscle disease

Hematologic
- acute blood loss
- chronic blood loss
- polycythemia
- methemoglobinemia

Metabolic
- hypoglycemia
- adrenogenital syndrome
- shock

Differential
- ❑ pink upper, blue lower (more common)
  - PPHN
  - left heart obstruction/hypoplasia
  - coarctation of aorta post subclavian/interrupted aortic arch
- ❑ blue upper, pink lower
  - TGA with R to L shunt across PDA

APNEA

Definition
- ❑ absence of respiratory gas flow for 20 seconds in the preterm infant and 15 seconds in the term infant (less if associated with bradycardia or cyanosis)
- ❑ central: no chest wall movement
- ❑ obstructive: chest wall movement continues
- ❑ mixed: combination of central and obstructive apnea

Differential Diagnosis
- ❑ apnea < 24 hrs – strongly associated with sepsis
- ❑ apnea > 24 hrs – if not pathological, apnea of prematurity
- ❑ in term infant apnea always requires full W/U
- ❑ CNS
  - apnea of prematurity presents in the first week of life due to prematurity of CNS and resolves by 36 weeks GA.
  - seizures
  - intracranial hemorrhage
- ❑ sepsis
- ❑ GI: GE reflux, esophagitis
- ❑ metabolic: low glucose, low calcium, low Na
- ❑ cardiovascular
  - low and high blood pressure
  - anemia, hypovolemia, PDA
- ❑ drugs: demerol, morphine

Treatment
- ❑ correct underlying cause
- ❑ tactile stimulation, reduce warming of face
- ❑ monitoring
- ❑ oxygen, CPAP, ventilation
  - medications: methylxanthines (caffeine, theophylline) which stimulate CNS and diaphragm,
  - doxapram (direct CNS stimulant) used in some centres

JAUNDICE
- ❑ very common - 65% of newborns
- ❑ 85-102 umol/L (5-6 mg/dl) bilirubin in blood to be visible
- ❑ look at sclera, mucous membranes, palm creases

Risk Factors
- ❑ prematurity
- ❑ acidosis
- ❑ sepsis
- ❑ hypoalbuminemia
- ❑ dehydration

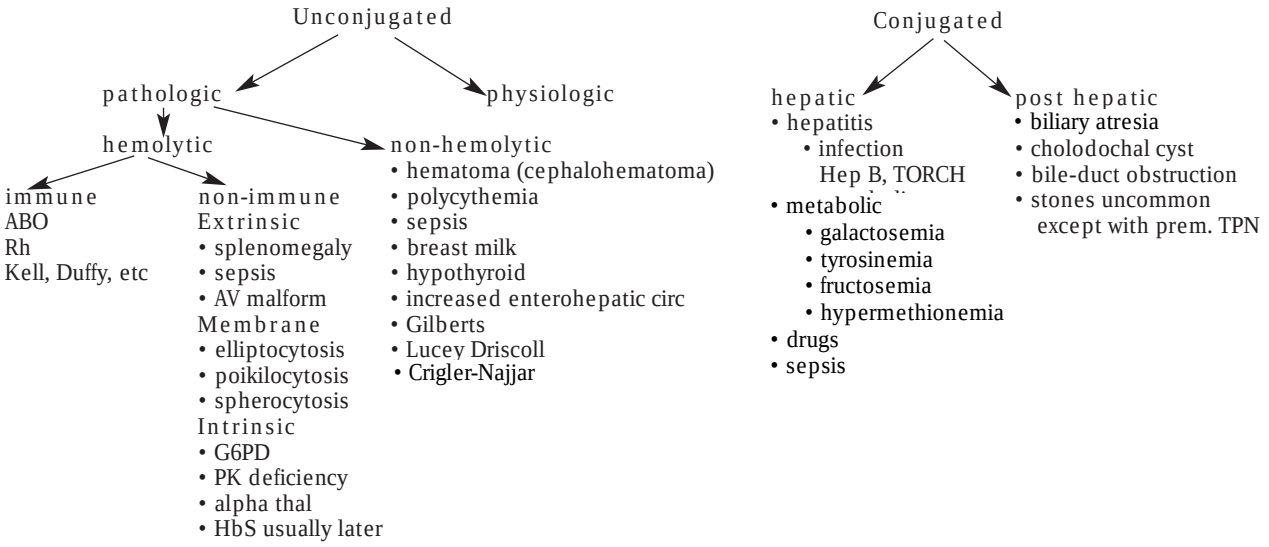


Figure 2. Approach and Differential for Neonatal Jaundice

< 24 Hours of Age

- ❑ always pathologic and requires investigation
  - blood group, Coombs, hemoglobin, peripheral smear
- ❑ hemolysis
  - Rh or ABO incompatibility
  - internal hemorrhage
- ❑ sepsis/congenital infection: TORCH

> 24 Hours of Age

- ❑ physiologic
  - immature liver enzymes, increased hematocrit with decreased RBC lifespan overload the liver
  - onset day 2-5 in fullterm, 6-7 in preterm infants, usually peaks 2 days after onset
  - doesn't increase faster than 85 umol/L /day, doesn't exceed 220 umol/L
- ❑ if not physiologic, then investigate: blood group, Coombs, hemoglobin, peripheral smear
- ❑ consider septic workup CBC, diff, C&S urine and blood, ± CSF, ± chest x-ray
- ❑ increased hemolysis
  - G6PD deficiency, pyruvate kinase, spherocytosis
- ❑ bruising, hemorrhage, hematoma, cephalohematoma
- ❑ polycythemia
- ❑ drugs
- ❑ sepsis/congential infection: TORCHS
- ❑ dehydration

Prolonged Neonatal Jaundice (> 1 Week of Age)

- ❑ breast milk
  - 1/200 breast fed infants
  - inhibition of glucuronyl transferase activity
  - may persist up to 4-6 weeks
- ❑ hypothyroidism
- ❑ neonatal hepatitis
- ❑ conjugation dysfunction (e.g. Gilbert's disease, Crigler-Najjar Syndrome)
- ❑ inborn error of metabolism (e.g. galactosemia)
- ❑ impaired excretion (e.g. biliary atresia, choledochal cyst)
  - conjugated hyperbilirubinemia
  - pale stools, dark urine
  - failure to thrive, malabsorption

Kernicterus
- ❑ CNS toxicity (associated with increased unconjugated bilirubin + saturation of albumin or open blood brain barrier, basal ganglia targeted)
- ❑ clinical features include hearing loss, CP (athetoid), motor dysfunction, severe mental retardation, death

Treatment
- ❑ maintain good hydration and normal acid-base status
- ❑ 1st line therapy: phototherapy - photoisomerization (blue light most effective)
- ❑ exchange transfusion, depending on level of bilirubin, age, weight
- ❑ treat any underlying cause
- ❑ do not interrupt breastfeeding in healthy term newborns

NECROTIZING ENTEROCOLITIS (NEC)
- ❑ intestinal inflammation associated with focal or diffuse ulceration and necrosis primarily affecting terminal ileum and colon

Etiology
- ❑ multifactorial associations
- ❑ prematurity —> immature defenses
- ❑ asphyxia, acidosis and hypoxia leading to bowel ischemia
- ❑ infection: C. *difficile* toxin, coagulase negative staph in NICU
- ❑ hypertonic feedings / enteral alimentation
- ❑ hypovolemia, hypothermia
- ❑ milk substrate (?cow's milk protein, ?osmolality)

Clinical Features
- ❑ distended abdomen and signs of obstruction (vomiting)
- ❑ increased amount + bile stained gastric aspirate/vomitus
- ❑ frank or occult blood in stool
- ❑ feeding intolerance
- ❑ diminished bowel sounds
- ❑ signs of bowel perforation - sepsis, shock, peritonitis

Investigation
- ❑ abdomen x-ray: intramural air, perforation, fixed loops, thickened bowel wall
- ❑ high WBC, low plt, electrolyte imbalances, acidosis, hypoxia, hypercarbia

Treatment
- ❑ NPO, vigorous IV fluid resuscitation, NG decompression
- ❑ TPN
- ❑ antibiotics for infection
- ❑ serial abdominal x-rays detect early perforation
- ❑ surgery for complications (e.g. perforation)

SUDDEN INFANT DEATH SYNDROME (SIDS)
- ❑ sudden and unexpected death of an infant < 12 months of age in which the cause of death cannot be found by history, examination and a thorough postmortem
- ❑ 1-2/1,000 (leading cause of death between 1-12 months of age)
- ❑ frequency varies widely in different populations

Epidemiology
- ❑ more common in children placed in prone position (? cause vs. association)
- ❑ number of deaths peak at age 2 months
- ❑ increase in deaths during peak respiratory virus season
- ❑ most deaths occur between midnight and 8:00 am
- ❑ more common in prematurity, smoking in household, minorities, socially disadvantaged
- ❑ 3:2 male predominance
- ❑ risk of SIDS is increased 3-5X in siblings of infants who have died of SIDS

Prevention
- ❑ do not place infant in prone position
- ❑ alarms/other monitors not recommended ~ increase anxiety and do not prevent life-threatening events
- ❑ avoid overheating and overdressing babies
- ❑ appropriate infant bedding

SEIZURE DISORDERS

Classification and description – see Neurology section

Childhood Epileptic Syndromes

- ❑ infantile spasms
  - onset 4-8 months
  - brief, repeated contractions of neck, trunk and extremities (flexion and extension) lasting 10-30 seconds
  - occur in clusters; often association with developmental delay
  - 40% unknown etiology but association with syndromes e.g. tuberous sclerosis
  - treatment includes ACTH, oral steroids, benzodiazepines, valproate, vigabatrin
- ❑ Lennox-Gastaut
  - preschool children
  - multiple seizure types common with frequent status epilepticus
  - seen with previous encephalopathy and brain malformations
  - treatment includes valproic acid, benzodiazepines and ketogenic diet; however, responses often poor
- ❑ Juvenile myoclonic epilepsy
  - adolescent onset (12-16 years of age); autosomal dominant
  - myoclonus particularly in morning (generalized T-C)
  - requires lifelong valproic acid; prognosis excellent
- ❑ Benign childhood epilepsy with rolandic spikes
  - onset peaks at 9-10 year of age
  - focal motor seizures involving tongue, mouth and face
  - remains conscious but aphasic post-ictally
  - remits spontaneously in adolescence; no sequellae

Generalized Tonic Clonic Seizures

- ❑ most common type of nonfebrile seizures in childhood
- ❑ generalized from onset (does not include partial seizures that become generalized)
- ❑ often associated with tongue biting and incontinence

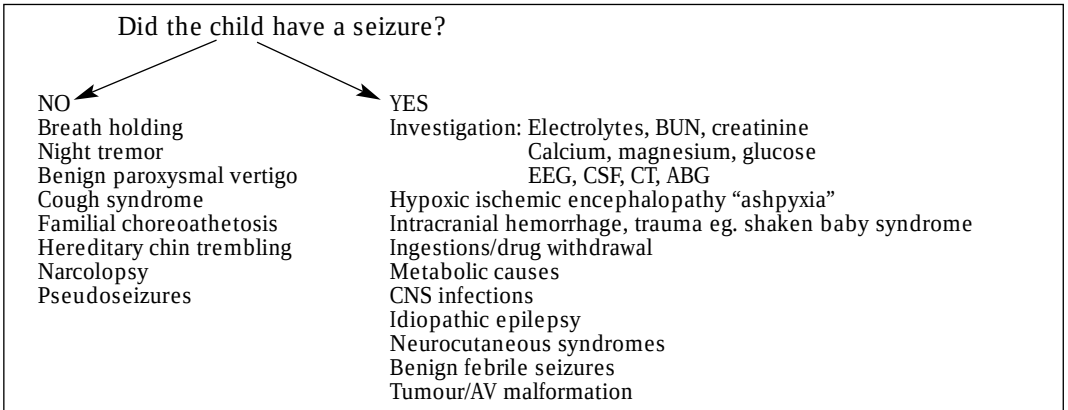


Figure 3. An Approach to the Child with a Suspected Convulsive Disorder

Table 11. Anticonvulsive Treatment by Seizure Type

Seizure Type	Treatment
absence	ethosuximide or valproic acid if > 2 years
generalized tonic-clonic	phenobarbital in first 12 months, carbamazepine after
myoclonic	ethosuximide, valproic acid, primidone, clonazepam
partial seizures	carbamazepine or phenytoin (Gabapentin, Lamotrigine, Vigabatrin as add-on therapy)

Treatment
- ❑ treat with drug appropriate to clinical situation
- ❑ start with one drug and increase dosage until seizures controlled
- ❑ if no effect, switch over to another before adding a second anticonvulsant
- ❑ education for patient and parents
    - privileges and precautions in daily life (e.g. buddy system)
- ❑ continue anticonvulsant treatment until patient free of seizures for 2 years or more

BENIGN FEBRILE SEIZURES
- ❑ most common cause of seizure in children
- ❑ 3-5% of all children, M > F

Criteria
- ❑ age 6 months - 6 years
- ❑ thought to be associated with initial rapid rise in temperature
- ❑ no interictal neurologic abnormalities
- ❑ no evidence of CNS infection/inflammation or acute systemic metabolic disorder
- ❑ no history of non-febrile seizures
- ❑ most common seizure type is generalized tonic-clonic; however may be any type
- ❑ risk factors include
    - family history of febrile seizures (40% positive)
    - high fever
    - slow development of child

Simple Febrile Seizure
- ❑ duration < 15 minutes (95% < 5 minutes)
- ❑ generalized, symmetric
- ❑ does not recur in a 24 hour period

Atypical Febrile Seizure
- ❑ focal origin
- ❑ > 15 minute duration, multiple (> 1 in 24 hours)
- ❑ followed by transient neurologic deficit

Risk Factors for Recurrence
- ❑ 33% chance of recurrence
- ❑ age of onset < 1 year
    - 50% chance of recurrence if < 1 year
    - 28% chance of recurrence if > 1 year
- ❑ risk of epilepsy is < 5%; risk factors include abnormal development of child previous to seizures, family history of afebrile seizures and a complex initial seizure

Workup
- ❑ history: determine focus of fever, description of seizure, meds, trauma history, development, family history
- ❑ exam: LOC, signs of meningitis, neurologic exam
- ❑ R/O meningitis – do LP if signs and symptoms of meningitis
- ❑ EEG not warranted unless atypical febrile seizure or abnormal neurologic findings
- ❑ investigations unnecessary except for determining focus of fever

Management
- ❑ COUNSELLING AND REASSURANCE TO PATIENT AND PARENTS
- ❑ antipyretics (e.g. acetaminophen), tepid baths, fluids for comfort (will not prevent seizure)
- ❑ prophylaxis not given except in very unusual circumstances
- ❑ if high risk for recurrent or prolonged seizures carry rectal Ativan at home

FLOPPY BABY (HYPOTONIA)
- ❑ decreased resistance to movement
- ❑ proper assessment of tone requires accurate determination of gestational age
- ❑ history – obstetrical/perinatal, family, exposures, regression in milestones
- ❑ evaluate
    - spontaneous posture (spontaneous movement? against gravity?) important in evaluation of muscle weakness
    - joint mobility (hyperextensibility?)

- shaking of limbs
- postural maneouvres

❑ postural manoeuvres include
- traction response – pull to sit and look for flexion of arms to counteract traction; no response at <33 weeks gestation
- axillary suspension – suspend infant by holding at axilla and lifting; hypotonic babies will slip through the grasp because of low shoulder girdle tone
- ventral suspension – infant is prone and supported under the abdomen by one hand; infant should be able to hold up extremities; inverted "U" posturing demonstrates hypotonia, that is, baby will drape self over examiner's arm

❑ investigations
- R/O systemic disorders
- lytes, blood glucose, Ca^{2+}, Mg, creatinine
- enhanced CT of brain
- peripheral CK, EMG, muscle biopsy
- chromosome analysis, genetic testing

❑ differential diagnosis of
- hypotonia with associated weakness
  - cerebral – malformation, infections, kernicterus, hypoxia
  - toxins (via mother) – narcotics, benzodiazepines, general anaesthetic, magnesium sulphate
  - spinal cord – trauma, tumour, myelodysplasia, infection, vascular lesion
  - anterior horn cell – spinal muscular atrophies
  - peripheral nerve – post-infectious neuropathy
  - neuromuscular junction – botulism, infantile myasthenia
  - muscle – Duchenne muscular dystrophy, myotonic dystrophy
- hypotonia without weakness
  - systemic – sepsis, heart failure, chromosomal (Down and Prader-Willi syndromes)
  - connective tissue – Marfan syndrome, Ehler-Danlos
  - cerebral – birth trauma, hemmorhage, intrapartum hypoxia
  - metabolic – nutritional (rickets), renal tubular acidosis, celiac disease

CEREBRAL PALSY

❑ nonprogressive central motor impairment syndrome due to prenatal/perinatal events (trauma, lesions, metabolic abnormalities anomalies of brain); a symptom complex, NOT a disease
❑ association with low birth weight babies
❑ incidence 1.5-2.5/1000 live births (developing countries)
❑ extent of mental retardation varies
❑ life expectancey is dependent on the degree of mobility and mental retardation, not on severity of CP

Types

❑ spastic i.e. increased tone - diplegia: lower limbs > upper limbs often due to interventricular hemorrhage or periventricular leukomalacia; hemiplegia: one-sided paralysis; quadraplegia
❑ extrapyramidal – choreoathetoid (kernicterus), dystonic (fluctuating high/low tone)
❑ hypotonic
❑ ataxic
❑ mixed

Etiology

❑ often obscure or multiple
❑ no definite etiology identified in 1/3 of cases
❑ 10% due to postnatal insult - infections, asphyxia and trauma

Other Signs

❑ swallowing incoordination - aspiration
❑ microcephaly (25%)
❑ seizures
❑ mental retardation, learning disabilities
❑ delay in motor milestones

Investigations
- ❑ include metabolics, chromosome studies, tissue exam, serology, neuroimaging, evoked potentials, EEG (if seizures), ophthalmology, audiology

Treatment
- ❑ maximize potential through multidisciplinary services; important for family to be connected with various support systems
- ❑ orthopedic management (e.g. dislocations, contractures, rhizotomy)

HYDROCEPHALUS (see Neurosurgery Notes)
- ❑ excessive accumulation of CSF associated with progressive ventricular dilatation
- ❑ pathophysiology/etiology
  - increased production or CSF e.g. choroid plexus papilloma
  - decreased absorption of CSF e.g. hyperplasia of arachnoid villi, infection/hemorrhage destroying arachnoid villi
  - obstruction to flow of CSF e.g. congenital malformations (Dandy-Walker, Arnold-Chiari), masses, infections, congenital bone defects

Clinical Signs
- ❑ in utero - large head
- ❑ ventricular distention leads to stretching of the pathways surrounding ventricles which may cause ataxia, spasticity (lateral ventricle), hypothalamic dysfunction (3rd); impaired vertical gaze (4th)
- ❑ acute (increased ICP)
  - irritability, lethargy, loss of appetite, vomiting
  - large fontanelle; splayed sutures
  - headache
  - cranial nerve deficits
  - herniation/coma
- ❑ chronic
  - onset < 2 years: macrocephaly and excessive rate of head growth
  - ataxia, spasticity
  - papilledema, optic atrophy, impaired upward gaze,
  - endocrine dysfunction (primarily causing growth failure)

Diagnosis
- ❑ prenatal ultrasound
- ❑ post natal ultrasound/CT/MRI

Treatment
- ❑ medical – treat underlying cause; acetazolamide (transiently decreases CSF production)
- ❑ surgical – remove lesion; ventriculoperitoneal shunt

NEURAL TUBE DEFECTS
- ❑ defective closure of caudal neural tube in fourth week gestation to varying degrees
- ❑ spina bifida occulta: vertebrae only (L5, S1), may have identifying dimple or tuft of hair; generally asymptomatic
- ❑ meningocele: vertebrae, meninges involved whereas myelomeningocele also includes spinal cord; neurologic deficits depend on level of lesion (include bowel/bladder dysfunction, paralysis and sensory deficits)

Etiology
- ❑ most neural tube defects are polygenic
- ❑ folic acid administration prior to conception lowers the risk of NTDs > 75%

Screening
- ❑ antenatal screening: triple screen, amniotic fluid AFP
- ❑ U/S + triple screen will detect 90% of NTDs
- ❑ examine backs of all newborns for pigmented spots or hairy patches

Management
- ❑ essential to have multidisciplinary approach for the family
- ❑ closure of the skin defect to prevent infection
- ❑ shunting to address associated hydrocephalus

- ❑ intermittent catheterization to decrease UTIs, reflux nephropathy
- ❑ orthopedics/orthotics and physiotherapy to help with posture and ambulation
- ❑ anesthetic skin care (e.g. bed sores)
- ❑ tethered cord release
- ❑ also important to address social issues

NEUROCUTANEOUS SYNDROMES

- ❑ characterized by tendency to form tumours of CNS, PNS, viscera and skin
- ❑ Neurofibromatosis type I
  - cafe-au-lait spots, axillary freckles, Lisch nodules of the iris, neurofibromas (progressive and potential to invade)
  - seizures, scoliosis, optic glioma
  - type II does not have above lesions; associated with brain tumours; bilateral acoustic neuromas are diagnostic
- ❑ Sturge-Weber's: port-wine nevus in V-1 distribution with associated angiomatous malformation of brain, seizures, contralateral hemiparesis
- ❑ Tuberous Sclerosis: adenoma sebaceum, “ash leaf” hypopigmentation, cardiac rhabdomyomas, kidney angioleiomyomas, mental retardation and seizures

GASTROINTESTINAL DISEASE

VOMITING

Approach

- ❑ consider: infection, inflammation, mechanical obstruction, motility disorders, others (e.g. eating disorder)
- ❑ Non GI causes: CNS, UTI, systemic infections, others

Assessment

- ❑ history
  - age of onset, duration, severity
  - quality: bilious, bloody, regurgitation
  - associated symptoms e.g. fever, abdominal pain
  - effect on growth and development, concurrent disease
- ❑ physical exam: assess hydration (see Table 14)
- ❑ lab investigation
  - bloody emesis: investigate for causes of upper GI bleed
  - bilious emesis: rule out obstruction (upper GI series, U/S)
  - regurgitation: evaluate for reflux (barium swallow with fluoroscopy, 24 hour esophageal pH probe)
- ❑ useful tests (based on history and physical exam)
  - CBC, lytes, BUN, Cr, ESR
  - urine, blood, stool C&S
  - amylase, lipase
  - arterial blood gases
  - abdominal x-ray, ultrasound, contrast radiology
  - endoscopy
- ❑ management
  - treat the underlying cause
  - rehydration

VOMITING IN THE NEWBORN

- ❑ congenital anomalies are a frequent cause, e.g. atresia, Hirshprung’s
- ❑ differential diagnosis: gastroenteritis, gastroesophageal reflux, overfeeding, food allergy, milk protein intolerance

Tracheoesophageal Fistula

- ❑ incidence: 1:3000-1:4500
- ❑ clinical features vary with type
  - vomiting, coughing and gagging
  - cyanosis with feeds
  - respiratory distress
  - may have history of maternal polyhydramnios
  - associated anomalies: VATER = Vertebral anomalies, Anal atresia, TEF and Renal disease plus cardiac abnormalities and radial defects of the upper limb

- ❑ x-ray —> plain and contrast studies show anatomic abnormality, NG tube curled in pouch
- ❑ treatment: early repair to prevent lung damage and maintain nutrition
- ❑ complications
  - pneumonia, lung damage, chronic reactive airways
  - stenosis and strictures at repair site
  - gastroesophageal reflux and poor swallowing following repair

Duodenal Atresia

- ❑ clinical features
  - bile-stained vomiting if distal to bile duct
  - abdominal distention, peristaltic waves
  - dehydration
  - associated with Down syndrome
  - may have history of maternal polyhydramnios
- ❑ abdominal x-ray —> air-fluid levels on upright film
  - "double bubble" sign (dilated stomach and duodenum)
- ❑ differential diagnosis: annular pancreas, aberrant mesenteric vessels, pyloric stenosis
- ❑ treatment
  - decompression with NG tube
  - correction of metabolic abnormalities
  - surgical correction

Pyloric Stenosis

- ❑ incidence: most common in first-born males, often family history
  - M:F = 5:1
- ❑ clinical features
  - non-bilious projectile vomiting that occurs after feeding
  - usually starts at 2-6 weeks of age
  - infant hungry and alert, will re-feed
  - FTT, wasting
  - dehydration, may lead to prolonged jaundice
  - gastric peristalsis goes from LUQ to epigastrium
  - “olive sign” (olive-shaped mass on right at margin of rectus abdominis muscle)
- ❑ lab: hypochloremic metabolic alkalosis
- ❑ diagnosis: clinical, abdominal ultrasound
- ❑ treatment: pyloromyotomy

Malrotation of the Intestine

- ❑ 3 presentations: recurrent vomiting (bilious intermittently); FTT with vomiting; sudden onset abdominal pain and then shock
- ❑ if vomiting with bilious material, malrotation with volvulus until proven otherwise
- ❑ 80% experience symptoms in first two months of life
- ❑ clinical features
  - distended abdomen
  - vomiting due to volvulus and bands across duodenum
  - cecum free
- ❑ diagnosed by upper GI studies: duodenum not fixed, spiral jejenum, mobile cecum (may not be in RLQ)
- ❑ treatment: surgical

Other

- ❑ meconium ileus (see Cystic Fibrosis Section)

VOMITING AFTER THE NEWBORN PERIOD

- ❑ distinguish from regurgitation (passive ejection of gastric contents secondary to reflux)

Infectious

- ❑ GI causes: gastroenteritis, peritonitis, appendicitis, hepatitis, ulcers, pancreatitis
- ❑ non-GI causes: UTI, otitis media, CNS infection, raised ICP, almost any infection, drugs, foreign body

Anatomic
- ❑ GI tract obstruction
  - intussusception (see below)
  - foreign body e.g. bezoar
- ❑ gastroesophageal reflux
  - usually temporary relaxation of lower esophageal sphincter —> decreased gastric emptying
  - presents with recurrent vomiting after feeds and FTT
  - most outgrow reflux by 18 months of age
  - conservative management: thickened feeds, elevate bed to 30 degrees
  - esophagograms may miss, pH studies are preferred
  - treat only if symptomatic or poor weight gain
  - medication e.g. cisapride, H2 blockers
  - if unresponsive to medication: surgery - Nissen fundoplication
  - complications: aspiration, esophageal bleeding, stricture formation, apnea

Central Nervous System
- ❑ increased ICP
  - hydrocephalus
  - neoplasm
- ❑ drugs/intoxicants
- ❑ migraine
- ❑ meningitis, encephalitis

Other
- ❑ metabolic/endocrine e.g. DKA, inborn errors, liver failure
- ❑ poisons/drugs: e.g. lead, digoxin, erythromycin, theophylline
- ❑ psychogenic: e.g. rumination syndrome, bulimia, anorexia, cyclic vomiting
- ❑ food allergy
- ❑ regurgitation, overfeeding

ACUTE DIARRHEA
- ❑ get a good history (daycare, travel, drugs, foods, other symptoms)

Etiology
- ❑ viral infection
  - most common in Canada, e.g. Rotavirus
  - associated with URTIs
  - slight fever, malaise, vomiting, vague abdominal pain
  - resolves in 3-7 days
- ❑ bacterial infection
  - Salmonella, Campylobacter, Shigella, pathogenic E. *coli*, Yersinia
  - more severe abdominal pain, high fever, bloody diarrhea
- ❑ parasitic infection
  - Giardia lamblia, E. histolytica
- ❑ toxin-induced: staphylococcal food poisoning, C. *difficile* toxin
- ❑ allergic: food intolerance
- ❑ antibiotic-induced
- ❑ non-specific: associated with any non-GI infection, generalized sepsis or shock

Complications
- ❑ dehydration (see Table 14)
- ❑ electrolyte disturbances: hyper or hyponatremia, hypokalemia, metabolic acidosis
- ❑ secondary disaccharidase deficiency (transient, due to villous damage)

Table 14. Signs of Dehydration

	None	Some	Severe
decrease body weight	–	3-5%	9-10%
neurological status	alert, well	irritable	lethargic or unconscious; floppy
sunken eyes	–	+	++
prolonged skin fold	–	+	++
dry oral mucosa	–	+	++
thirst	N, not thirsty	thirsty, drinks eagerly	drinks poorly or not able to drink
tears	present	absent	absent
urine output	N	↓	anuria
HR	N	slight ↑	↑
BP	N	N	↓

Investigations
- ❑ stool for C&S and O&P, blood and WBC, C. *difficile* toxin, Rotazyme assay

Management
- ❑ rehydration: most children managed with oral fluids e.g. Oral Rehydration Solution (Pedialyte, Gastrolyte)
- ❑ fluid replacement: consider deficit (% of body weight), maintenance and ongoing losses
- ❑ maintenance fluid requirements
  - newborn: 120-160 cc/kg/day (may vary with weight)
  - 100 cc/kg/24 hours for first 10 kg or 4 cc/kg/h
  - 50 cc/kg/24 hours for second 10 kg or 2 cc/kg/h
  - 20 cc/kg/24 hours thereafter or 1 cc/kg/h
  - IV fluid rate per hour = total per day divided by 24 (or use 4:2:1 rule)
- ❑ commonly used IV fluids
  - first week of life: D5W + 0.2 NS
  - 2/3 D5W 1/3 NS
  - NS: as bolus to restore circulation in very dehydrated child
- ❑ continue breast feeding when possible
- ❑ DRUGS NOT INDICATED: kaolin, pectin, anticholinergics, antispasmotics, opiate derivatives
- ❑ antibiotics used in: *Salmonella sepsis*, *Shigella/Yersinia/enterotoxic* E. *coli* (Septra), C. *difficile* (oral Flagyl/Vancomycin), *Campylobacter* (Erythromycin)

Table 15. Correction of Fluid and Electrolyte Deficits

Dehydration[1]	5%	10%	Rate
Isotonic	Na 4-5 mmol/kg	Na 8-10 mmol/kg K 4-5 mmol/kg	1/2 deficit over 1st 8 hours, then 1/2 over 16 hours
Hypotonic[2] Na < 130 mmol/L	Na 5-6 mmol/kg K 3 mmol/kg	Na 10-12 mmol/kg K 5 mmol/kg	If Na ≥ 105, correct as above If Na < 105, correct by 20 mmol/L maximum over 0.5-4 hour with hypertonic saline
Hypertonic Na > 150 mmol/L	Na 2-4 mmol/kg K 2-4 mmol/kg	Na 2-4 mmol/kg K 2-4 mmol/kg	Correct over 48-72 hours Do not allow serum Na to drop faster than 10-15 mmol/L/day[3]

Note: 1. For all types dehydration, H_2O for 5% dehydration = 50ml/kg; for 10% dehydration = 100 ml/kg
2. To calculate exact deficit: [Na] deficit = ($[Na]_{target} - [Na]_{actual}$) x body weight (kg) x total body H_2O (L/kg)
3. To lower serum Na by a predictable amount, remember: 4 ml/kg of free H_2O lowers serum Na by 1 mmol/L

CHRONIC DIARRHEA

Clinical Assessment
- ❑ > 14 days
- ❑ onset, nature of stool
- ❑ nutritional status (chronic diarrhea with FTT suggests malabsorption)
- ❑ history of infection
- ❑ hydration status

Investigations for Diarrhea of Unknown Etiology
- ❑ serial heights, weights, growth percentiles
- ❑ stools for C&S, O&P, occult blood, C. *difficile*, pH, reducing substances
- ❑ malabsorption work-up if indicated (see Chronic Diarrhea with FTT below)
- ❑ x-rays
  - upper GI series
  - barium enema
- ❑ mucosal biopsy

CHRONIC DIARRHEA WITHOUT FAILURE TO THRIVE

Infectious
- ❑ bacterial (e.g. *Campylobacter, Salmonella*)
- ❑ antibiotic induced: C. *difficile* colitis - often bloody stool
- ❑ parasitic: *Giardia lamblia*
- ❑ post-infectious: secondary lactase deficiency

Toddler's Diarrhea
- ❑ most common cause of chronic diarrhea during infancy, but still diagnosis of exclusion in thriving child
- ❑ onset between 6-36 months of age, ceases spontaneously between 2-4 years
- ❑ stool may contain undigested food particles, 4-6 BM per day
- ❑ excoriated diaper rash
- ❑ diet history: lots of juice overwhelms small bowel resulting in disaccharide malabsorption
- ❑ four F's: adequate fiber, normal fluid intake, 35-40% fat, discourage excess fruit juice
- ❑ management: reassurance, self-limiting

Lactase Deficiency (Lactose Intolerance)
- ❑ clinical features
  - chronic, watery diarrhea
  - abdominal pain, bloating, borborygmi
- ❑ two scenarios
  - primary lactose intolerance: crampy abdominal pain with loose stool in older children, usually in Orientals, Blacks
  - secondary lactose intolerance: old infant, persistent diarrhea post viral/bacterial infection, Celiac disease, or inflammatory bowel disease
- ❑ diagnosis
  - clinical trial off milk
  - watery stool, acid pH, positive reducing sugars
  - positive breath hydrogen test if > 6 years
- ❑ management
  - lactose tolerance test
  - milk free diet, soy formula
  - Lacteeze, Lactaid tabs/drops

CHRONIC DIARRHEA WITH FAILURE TO THRIVE
- ❑ suggests malabsorption (with frequent bulky, foul smelling stools)
- ❑ investigation of malabsorption
  - stool consistency, pH, reducing substances, microscopy, occult blood
  - stool: O&P, C&S, C. *difficile* toxin, 3-day fecal fat
  - chest x-ray
  - urinalysis
  - CBC, differential, ESR, smear, electrolytes, total protein, immunoglobulins
  - absorptive and nutritional status: albumin, carotene, Ca^{2+}, PO_4, Mg, Zn, Fe, ferritin, folate, fat-soluble vitamins, PT, PTT
  - sweat chloride

- if indicated, α-antitrypsin level, thyroid function tests, urine VMA and HVA, HIV test, lead levels
- upper GI series + follow-through
- specialized tests: small bowel biopsy, endoscopy and biopsy

1. Intestinal Causes

Celiac Disease (Gluten-sensitive enteropathy)
❑ defect at the mucosal level (BROW: barley, rye, oats, wheat)
❑ toxic or immunologic reaction
❑ clinical features
- presents at any age, usually 6-18 months
- FTT with poor appetite, irritability, apathy
- anorexia, nausea, vomiting, edema
- wasted muscles, distended abdomen and flat buttocks
- anemia, bleeding
- rickets
- clubbing of fingers

❑ diagnosis
- fat malabsorption studies
- small bowel biopsy: flat atrophic mucosa with resolution after trial of gluten-free diet (villous atrophy)
- antigliadin, antiendomysial antibodies, low D-xylose absorption

❑ treatment
- gluten-free diet for life
- avoid BROW

❑ complications if untreated
- small bowel lymphoma
- malnutrition

Milk Protein Allergy
❑ immune-mediated mucosal injury
❑ can be associated with soy protein, anemia, hypoalbuminemia
❑ often atopic individuals

Other
❑ specific enzyme deficiencies
❑ liver disease, biliary atresia
❑ a-ß-lipoproteinemia
❑ short gut syndrome
❑ blind loop syndrome
❑ protein-losing enteropathy (Celiac, IBD, Giardia)

Inflammatory Bowel Disease
❑ see Gastroenterology Notes
❑ incidence: increasing in North America, mostly older children, teenagers

2. Pancreatic Insufficiency

Cystic Fibrosis (see Cystic Fibrosis Section)
❑ loss of exocrine pancreatic function
❑ clinical features
- meconium ileus in the newborn
- FTT with good appetite
- rectal prolapse
- steatorrhea
- respiratory symptoms, nasal polyps

❑ diagnosis: elevated sweat chloride (> 60 mEq/L), increased fecal fat, DNA mutation
❑ management (GI)
- pancreatic enzyme replacement
- fat soluble vitamins (A,D,E,K)

Shwachman Syndrome
❑ pancreatic insufficiency (autosomal recessive)
❑ cyclic neutropenia
❑ skeletal abnormalities (metaphyseal dystosis leading to short stature)
❑ dry skin, eczematous, ichthyosiform lesions

3. Diet-Induced
❑ food allergy

4. Other
❑ diets rich in sorbitol, fructose (poorly absorbed CHO)
❑ metabolic/endocrine
- thyrotoxicosis
- Addison's disease
- galactosemia

❑ immune defects
- IgA deficiency, hypogammaglobulinemia
- SCID
- AIDS

❑ neoplastic
- pheochromocytoma
- lymphoma of small bowel

ACUTE ABDOMINAL PAIN

Assessment
❑ most common GI complaint
❑ accurate description of pain and its characteristics
❑ vomiting before pain suggests gastroenteritis
❑ vomiting after pain suggests a surgical condition
❑ physical examination: rebound tenderness, bowel sounds, rectal exam
❑ labs
- CBC and differential
- urinalysis to rule out UTI

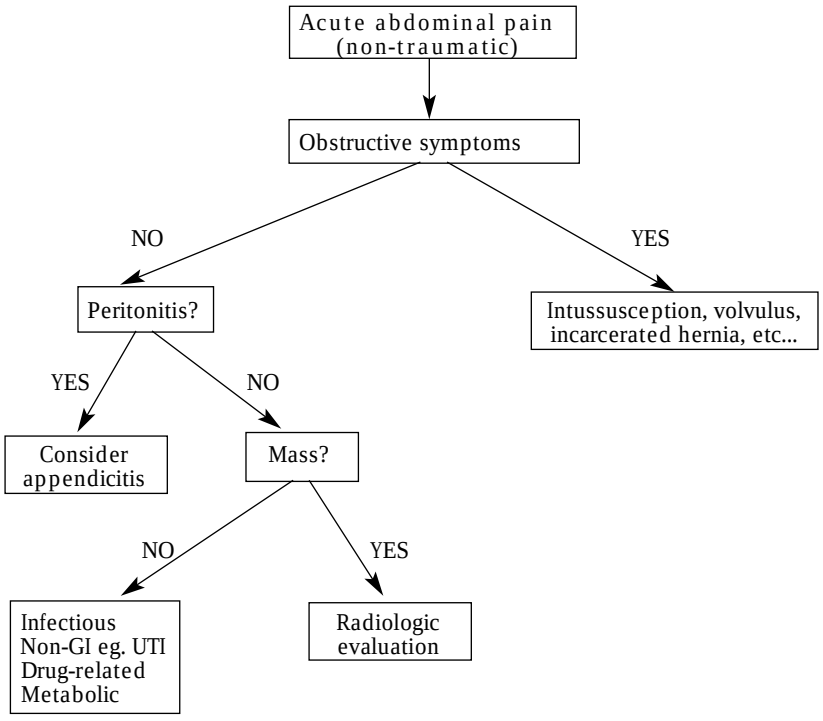


Figure 4. Approach to Acute Abdominal Pain

Differential Diagnosis
- gastroenteritis
- incarcerated hernia
- UTI
- appendicitis
- intussusception
- malrotation
- volvulus
- Henoch-Schönlein Purpura
- sickle cell crisis
- pneumonia
- DKA
- mesenteric adenitis

Notes

1. Appendicitis
- ❑ most common inflammatory bowel disorder from 5 years on
- ❑ clinical features
  - low grade fever
  - anorexia
  - abdominal pain: periumbilical then RLQ
  - nausea, vomiting (after onset of pain)
  - peritoneal signs
  - generalized peritonitis is a common presentation in infants/young children
- ❑ treatment: surgical
- ❑ complications
  - perforation
  - abscess

2. Intussusception
- ❑ 90% idiopathic, children with CF at significantly at risk
- ❑ 50% between 3 – 12 months, 75% before 2 years of age
- ❑ telescoping of segment of bowel into distal segment —> ischemia and necrosis
  - usual site: ileocecal junction
- ❑ lead point may be swollen Peyer's patches, Meckel's diverticulum, polyp, malignancy in older child
- ❑ clinical features
  - sudden onset of recurrent, paroxysmal, severe periumbilical pain
  - pain-free remissions
  - later vomiting and rectal bleeding (“red currant jelly” stools)
  - sausage-shaped mass often in upper to mid abdomen
  - shock and dehydration
  - “classic triad” of abdominal pain, palpable sausage-shaped mass and red currant jelly stools only in 10-15% of patients
- ❑ diagnosis and treatment
  - air enema —> see reverse "E" sign
  - U/S
  - reduction under hydrostatic pressure, air enema
  - surgery rarely needed

CHRONIC ABDOMINAL PAIN
- ❑ 10-15% of children
- ❑ definition: three or more episodes of pain severe enough to affect activities, occurring over a period of 3 months

Assessment
- ❑ distinguish organic from non organic
- ❑ history
  - weight loss, appetite, energy
  - associated vomiting, diarrhea
  - characteristics of pain
  - psychosocial issues
- ❑ physical exam: abnormalities suggest organic nature
- ❑ red flags for organic etiology
  - age < 5 years old
  - pain away from midline
  - localized pain awakens child at night
  - prominent vomiting, diarrhea
  - joint pain
  - fever
  - anemia
  - travel history
  - weight loss or failure to gain weight

Organic (< 10%)
- ❑ chronic infection
- ❑ GI
  - constipation - cause or effect?
  - inflammatory bowel disease
  - anatomic anomalies, masses
  - esophagitis
  - peptic ulcer disease, lactose intolerance
  - pancreatic, hepatobiliary
- ❑ genitourinary disease
- ❑ gynecological
- ❑ cardiovascular
- ❑ neoplastic

Functional, Recurrent Abdominal Pain (RAP) (90%)
- ❑ school age, peak 8-10 years
- ❑ F > M
- ❑ vague, crampy periumbilical or epigastric pain, vivid imagery to describe pain
- ❑ should not awaken child
- ❑ no precipitating or relieving factors, no consistent pattern
- ❑ child appears well with normal growth
- ❑ associated with school absenteeism
- ❑ diagnosis
  - must consider kidney disease, malrotation of bowel, IBD
  - school phobia?
- ❑ investigations as indicated
  - CBC, ESR, urinalysis, stools for O&P, C&S, occult blood
- ❑ treatment
  - manage any emotional or family problems
  - trial of high fibre diet, trial of lactose-free diet
  - reassurance

CONSTIPATION
- ❑ as many as 20% of children < 5 years of age

Assessment
- ❑ history
  - age of onset, dietary history
  - associated symptoms: abdo pain, encopresis, overflow diarrhea
- ❑ physical exam
  - examine lower back for evidence of occult cord lesion (NTD)
  - abdominal exam
  - rectal exam
- ❑ most often diet-related with no specific disease
- ❑ Hirschsprung's disease

Functional Constipation
- ❑ 99% of cases of constipation
- ❑ lack of bulk or fibre in diet or change in diet
- ❑ poor fluid intake
- ❑ in children, can occur during toilet training, or due to pain on defecation, stool witholding
- ❑ in infants, often when introducing cow's milk after breast milk
- ❑ treatment
  - increase fluids, increase dietary fibre
- ❑ complications
  - anal fissures and pain—> withhold passing stool —> chronic dilatation and overflow incontinence, encopresis = Pain Retention Cycle
- ❑ treatment
  - increase fluids, increase dietary fibre
  - may need mineral oil, laxatives
  - appropriate toilet training technique

Specific Organic Disorders

1. Hirschsprung's Disease (congenital aganglionic megacolon)
- ❑ rectosigmoid in 75% of cases
- ❑ incidence: M:F=3:1, 1/5 000 live births
- ❑ associated with Down syndrome
- ❑ clinical features
  - severity depends on length of involvement
  - no meconium within first 24 hours
  - palpable stool on abdominal exam with empty rectum on DRE
  - intermittent diarrhea, BM only with rectal stimulation
  - constipation
  - abdominal distention
  - vomiting
  - FTT
- ❑ complications
  - enterocolitis: may be fatal, peak incidence 2-3 months of age
  - toxic megacolon and perforation

❑ diagnosis
- barium enema: proximal dilatation due to functional obstruction, empty rectum
- manometric studies: may have false positives
- rectal biopsy: definitive diagnosis (absent ganglion cells)

❑ treatment
- nonsurgical if short segment
- surgery: colostomy and re-anastomosis

2. Other
❑ intestinal obstruction
❑ endocrine
- hypothyroidism
- diabetes mellitus
- hypercalcemia

❑ neurogenic bowel (i.e. spina bifida)
❑ anal fissure/stricture/stenosis
❑ collagen vascular disease
❑ drugs: lead, chemotherapy, opioids

ABDOMINAL MASS

Table 16. Differential Diagnosis of Abdominal Mass

	Benign	Malignant
Renal	hydronephrosis polycystic kidney disease hamartoma	nephroblastoma (Wilm's) renal cell carcinoma
Adrenal		neuroblastoma
Ovarian	ovarian cysts	ovarian tumors
Other	splenomegaly pyloric stenosis abdominal hernia teratoma	lymphoma retroperitoneal rhabdomyoscarcoma

❑ 50% of abdominal masses in the newborn are renal in origin

GASTROINTESTINAL HEMORRHAGE

Assessment
❑ assess hemodynamic stability
❑ NG tube to determine if upper or lower bleed
❑ history: acute or chronic, age of child
- associated symptoms, etc...

❑ management
- volume resuscitation and stabilization
- treat underlying condition

Upper GI Bleeding
❑ mucosal lesions
- gastritis/gastroenteritis
- esophagitis
- duodenal/gastric ulcer
- Mallory-Weiss tear
- epistaxis, foreign body

❑ vascular
- coagulopathy
- vitamin K deficiency (hemorrhagic disease of the newborn)
- esophageal varices

❑ other
- swallowed blood, food colouring

❑ investigations
- CBC, stool OB, NG aspirate: blood, pH, Apt test in newborn
- endoscopy, colonscopy when stable

❑ treatment
- underlying cause, may use H_2 blockers

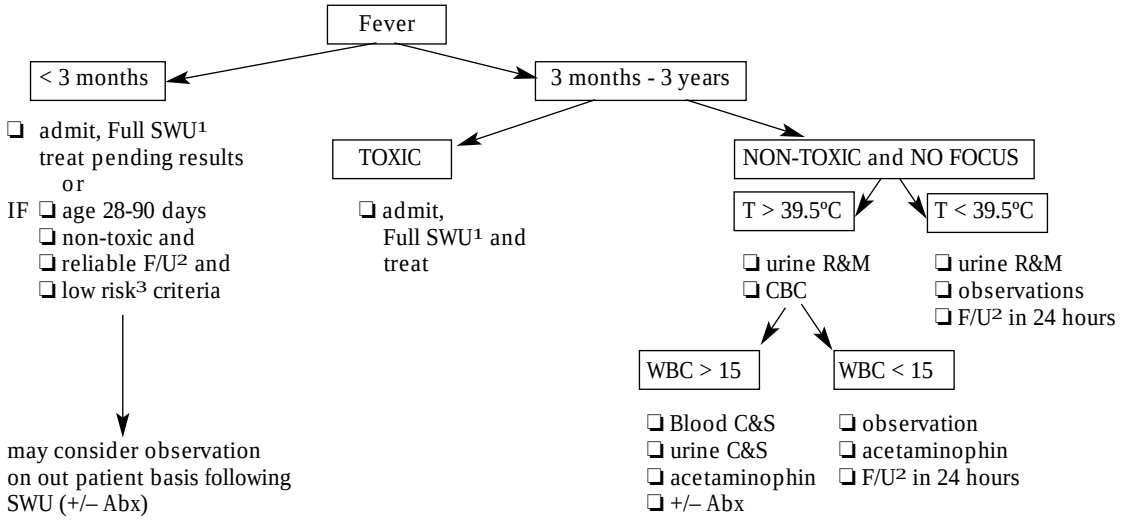
Notes

Lower GI Bleeding

1. Acute
- ❑ infection
  - bacterial, parasitic, antibiotic-induced (C. *difficile*)
- ❑ anatomic
  - malrotation/volvulus
  - intussusception "red currant jelly" stools
  - Meckel's diverticulum
  - anal fissures
- ❑ vascular/hematologic
  - Henoch-Schönlein Purpura
  - hemolytic-uremic syndrome (E. *coli*)
  - coagulopathy

2. Chronic
- ❑ anal fissures most common
- ❑ colitis
  - inflammatory: IBD
  - allergic (milk protein)
- ❑ structural
  - polyps: most are hamartomas
  - neoplasms: rare
- ❑ coagulopathy

INFECTIOUS DISEASES



NOTES:
1. Full septic workup - blood C&S, CBC and differential, urine R&M, C&S, LP, chest x-ray if respiratory SSx, stool C&S if GI SSx
2. Follow-up is crucial - if adequate F/U is not assured, a more aggressive diagnostic and therapeutic approach may be indicated
3. Low-Risk Criteria - previously healthy, normal physical exam (non-toxic), negative lab screen (WBC 5-15, < 1.5 x 10^9 bands, urine < 10 WBC/hpf, stool < 5 WBC/hpf)
4. Important Principles - the younger the child, the greater the difficulty to clinically assess the degree of illness

Figure 5. Approach to the Febrile Child

Clinical Pearl
- ❑ Teething may cause a temperature elevation >37.5ºC on the first day of the eruption in 50% of infants. However, significant temperature elevation should never be attributed solely to teething!

SEPSIS IN THE NEONATE

Table 17. Neonatal Sepsis

Early Onset (birth-8 days)	Late Onset (8-28 days)
• begins in utero • Risk Factors: maternal UTI, GBS positive, 1º maternal infection maternal fever/ leukocytosis/ chorioamnionitis prolonged rupture of membranes, prematurity, large inocculum • GBS, *E. coli*, *Listeria*, *Klebsiella*	• acquired after birth • usually healthy, full-term • same pathogens plus: *pneumococcus, meningococcus*, HSV, *Staphylococcus*

Signs of Sepsis
- respiratory distress, cyanosis, apnea
- tachycardia/bradycardia
- lethargy, poor feeding
- hypotonia, seizures, bulging fontanelle
- jaundice
- temperature instability (hypo/hyperthermia)

Table 18. Antibiotic Treatment of Serious Bacterial Infections

Neonate pathogens: GBS, *E.coli*, *Listeria*, S. *aureus*	ampicillin + gentamicin or ampicillin + cefotaxime +/– cloxacillin if risk of S. *aureus*
1-3 months same pathogens as above and below	ampicillin + cefotaxime +/– cloxacillin if risk of S. *aureus*
> 3 months pneumococcus, H. *influenzae* type b (> 5 years),* meningococcus	cefuroxime ceftriaxone or cefotaxime, if risk of meningitis vancomycin, if penicillin/ cephalosporin-resistant pneumococci

*Hib has dramatically decreased since introduction of Hib vaccine

MENINGITIS

❑ peak age: 6-12 months; 90% occurs < 5 years old

Risk Factors

❑ compromised immunity e.g. HIV, asplenia, prematurity
❑ neuroanatomical defects e.g. dermal sinus, neurosurgery
❑ parameningeal infection e.g. sinusitis, mastoiditis
❑ environmental e.g. day-care centres, household contact, travel to endemic regions

Pathophysiology

❑ URTI —> blood stream invasion from respiratory tract —> hematogenous seeding of meninges —> meningeal and CNS inflammation

Clinical Features

❑ +/– URI prodrome
❑ fever, toxic, lethargy, irritability
❑ headache, photophobia, nausea/vomiting
❑ younger infants may not demonstrate localizing signs, may have non-specific symptoms (poor feeding, irritability, lethargy)bulging fontanelle
❑ signs of meningismus: Brudzinski's, Kernig's, opisthotonous, nuchal rigidity, CN III and IV paralysis
❑ increasing head circumference (if sutures not closed)
❑ seizure in 20-30% of patients with bacterial meningitis
❑ petechial rash (meningococcus)

Diagnosis
- ❑ LP for CSF
  - raised opening pressure (norms: recumbent and relaxed, less flexed position < 160 mm H_2O, flexed lateral decubitus position = 100-280 mm H_2O)
  - cloudy in bacterial infection
- ❑ viral meningitis
  - Enterovirus, EBV, Influenza, Herpes, Adenovirus
  - WBC < 300 x 10^6/L (usually lymphocytes),
  - glucose normal, protein normal to high
- ❑ bacterial meningitis
  - WBC > 1000 x 10^6/L, increased PMNs; WBC may be < 100 x 10^6/L in early disease
  - elevated protein > 0.4 g/L
  - decreased glucose < 2.1 mmol/L (< 50 % serum glucose)
  - Gram stain positive in 80-90% of cases
  - CSF culture
  - Ziehl-Neelson stain, if TB suspected
  - latex agglutination tests if partially treated meningitis
- ❑ CBC (< 2 x 10^9/L WBC = bad prognostic marker)
- ❑ blood glucose
- ❑ blood cultures (positive in 90% cases)
- ❑ electrolytes (SIADH)
- ❑ if partially treated meningitis, LP may show persistent abnormalities, plus a positive CSF culture

Complications
- ❑ mortality: neonate 15-20%, children < 10%, pneumococcus > meningococcus > Hib
- ❑ acute
  - SIADH —> hyponatremia —> brain edema
  - seizures
  - subdural hematoma
  - brain abscess, disseminated infection (osteomyelitis, septic arthritis, abscess)
  - shock/DIC
- ❑ chronic
  - hearing loss
  - mental retardation/ learning disability
  - neurological deficit, seizure disorder
  - hydrocephalus

Treatment
- ❑ antibiotics (see Table 18) should be immediate, do not wait for LP results
  if viral: supportive, acyclovir for herpes
- ❑ fluid restriction if SIADH
- ❑ monitor glucose, acid-base and volume status
  steroids in Hib meningitis may reduce neurologic sequelae if given very early
- ❑ anticonvulsants may be needed to treat seizures
- ❑ isolation
- ❑ prophylaxis
  - active immunization
  - H. *influenzae* type b vaccine - routinely
  - meningococcal vaccine - if asplenic, complement deficient or for outbreaks
  - pneumococcal vaccine- if immunocompromised/splenectomized
  - BCG vaccine - if born in TB-endemic area
  - chemoprophylaxis for contacts and index case
    - H. *influenzae* - rifampin
    - N. *meningitidis* - rifampin (ceftriaxone or sulfisoxazole)
- ❑ report to public health if H. *influenzae* or N. *meningitidis*

HIV INFECTION

Epidemiology
- ❑ risk of infection 20-30% born to untreated HIV infected women
- ❑ transmission
  - infants and children: transplacental most common, maternal blood, rarely through breast milk
  - adolescents: sexual intercourse, needles, blood products
- ❑ incubation period: months to years (short incubation in 25%)
- ❑ signs and symptoms occur often within the first year, most within two years

HIV Testing
- ❑ viral nucleic acid by PCR
- ❑ viral culture
- ❑ viral antigen - p24
- ❑ HIV antibody - ELISA and Western blot to confirm
  - maternal HIV antibodies can persist up to 18 months
  - if child breastfeeding repeat test 3 months after stopping breastfeeding

Clinical Features of AIDS in Infants and Children
(see Infectious Diseases Notes)
- ❑ FTT, hepatomegaly, lymphadenopathy
- ❑ recurrent/persistent thrush
- ❑ chronic interstitial pneumonitis (relatively common); PCP
- ❑ opportunistic infections
- ❑ encephalopathy

Management
- ❑ prompt treatment of infections
- ❑ adequate nutrition
- ❑ prophylaxis
  - TMP/SMX for PCP
  - +/– IVIG
- ❑ nystatin, cotrimoxazole, ketoconazole, acyclovir if indicated
- ❑ suppression of HIV
  - Zidovudine, other e.g. didanosine
- ❑ immunizations
  - all routine immunizations (including MMR if well)
  - avoid OPV and BCG
  - pneumococcal, influenza and varicella vaccines

PERIORBITAL/ORBITAL CELLULITIS
- ❑ medical emergency
- ❑ periorbital vs. orbital (proptosis, compromised visual acuity, strabismus and extraocular movements, deep eye pain)

Clinical Features
- ❑ unilateral eyelid swelling with erythema
- ❑ conjunctive usually normal
- ❑ if bacteremic, other systemic features present (fever, WBC)
- ❑ orbital cellulitis: proptosis, ophthalmoplegia, pain on eye movement, decreased visual acuity

Pathophysiology
- ❑ secondary to sinusitis, dental sepsis, eye or skin infection
- ❑ primary infection with hematogenous spread to orbit
- ❑ *H.influenzae, S.pneumonia, S.aureus*

Treatment
- ❑ blood C&S
- ❑ urgent IV antibiotics
  - traumatic, any age: cloxacillin or cefazolin
  - nontraumatic, < 5 years: cefotaxime or cefuroxime
  - nontraumatic, > 5 years: cloxacillin or cefazolin
- ❑ may require urgent drainage
  - rifampin for contacts if H. *influenzae*
- ❑ mild early cases can be treated as outpatients with close follow-up

Complications
- ❑ cavernous sinus thrombosis
- ❑ meningitis
- ❑ brain abscess

OTITIS MEDIA (see Otolaryngology Notes)

Etiology
- ❑ S. *pneumoniae* (30%)
- ❑ nontypable H. *influenzae* (20%)
- ❑ M. *catarrhalis* (20%)
- ❑ group A Strep (5%)
- ❑ viral (20-25%)

Risk Factors
- ❑ daycare attendance
- ❑ bottle feeding in bed
- ❑ second-hand smoke
- ❑ formula-fed infants
- ❑ cleft lip, Down syndrome
- ❑ low socioeconomic status
- ❑ Inuit, Aboriginals

Clinical Features
- ❑ may follow URI
- ❑ painful ear, tugging, tinnitus, vertigo
- ❑ discharge if perforated
- ❑ hearing loss
- ❑ fever, vomiting, irritability in younger infants
- ❑ first stage —> slightly retracted, red tympanic membrane
- ❑ second stage —> bulging, red TM with fluid level, ± perforation

Treatment
- ❑ 1st line: amoxicillin
- ❑ if no improvement after 48 hours or child received amoxicillin in last 4 weeks, consider 2nd line:
  - erythromycin-sulfonamide (Pediazole)
  - trimethoprim/sulfamethoxazole
  - amoxicillin/clavulanate
  - cefixime (once daily regimen)
  - cefuroxime PO
- ❑ 10 day oral regimen for uncomplicated acute episodes
- ❑ ± daily prophylaxis if recurrent episodes
- ❑ ± tympanostomy tubes +/– adenoidectomy

Complications
- ❑ hearing loss, chronic effusion
- ❑ cholesteatoma, mastoiditis
- ❑ meningitis

STREPTOCOCCAL INFECTIONS

1. Pharyngitis and Tonsillitis
- ❑ viral etiology more common than bacterial in > 3 years of age group
- ❑ bacterial etiology (Group A Strep)
  - > 3 years old
  - sore throat, fever, exudate on red tonsils, tender cervical nodes, associated headache, abdominal pain
  - exudate on red tonsils also seen in EBV, adenovirus, diphtheria
- ❑ viral etiology (adenovirus, enterovirus, and EBV in older age group)
  - < 3 years old
  - runny nose, cough, diarrhea, rash

Management of Strep throat
- ❑ symptomatic
- ❑ antibiotics to prevent rheumatic fever, shorten illness duration

- ❑ > 3 years old, culture before treatment or do rapid Strep Antigen test
- ❑ rapid Strep test only 70-90% sensitive, do cultures if negative
- ❑ can prevent rheumatic fever if treated within 9-10 days
- ❑ antibiotics do not alter the risk of glomerulonephritis
- ❑ antibiotics for proven bacterial infection
    - penicillin or erythromycin x 10 days

Indications for Tonsillectomy

- ❑ proven, recurrent Strep tonsillitis
- ❑ peritonsillar abscess (rare)
- ❑ symptomatic tonsillar hypertrophy
    - sleep apnea
    - hypoxia
    - cor pulmonale
- ❑ suspected tumour

2. Scarlet Fever

- ❑ erythrogenic strain of Group A hemolytic Strep
- ❑ acute onset of fever, sore throat, strawberry tongue
- ❑ 24-48 hours after pharyngitis, rash develops which begins in the groin, axillae, neck, antecubital fossa
- ❑ within 24 hours, rash becomes generalized with perioral sparing
- ❑ rash fades after 3-4 days, may be followed by peeling
- ❑ penicillin (or erythromycin)

3. Post-Infectious Complications - Rheumatic Fever

- ❑ Jones Criteria (revised)
    - requires 2 major OR 1 major and 2 minor PLUS evidence of preceding Strep infection (increased ASOT, throat swab, recent scarlet fever)
    - major criteria: "SPACE"
        - subcutaneous nodules
        - pancarditis
        - arthritis (migratory)
        - chorea (Sydenham's)
        - erythema marginatum
    - minor critera
        - previous rheumatic fever or rheumatic heart disease
        - polyarthralgia
        - fever
        - elevated ESR or C reactive protein or leukocytosis
        - prolonged PR interval
- ❑ treatment
    - penicillin for acute course
    - secondary prophylaxis for at least 5 years or until 21 years old
    - anti-inflammatory drugs (ASA)
- ❑ complications
    - mitral insufficiency/stenosis
    - aortic insufficiency/stenosis

4. Invasive Group A Strep

- ❑ bacteremia post streptococcal disease of skin, resp tract, rectum, or vagina
- ❑ DIC, shock, and peripheral gangrene can occur
- ❑ hematogenous dissemination —> meningitis, osteomyelitis, arthritis, soft tissue abscesses, pneumonia, or endocarditis
- ❑ necrotizing fascitis
- ❑ streptococcal toxic shock-like syndrome may occur after streptococcal superinfection of varicella lesions
- ❑ treatment: IV penicillin. If allergic, erythromycin or clindamicin

5. Impetigo (see Dermatology Section)

6. Group B Strep

- ❑ common cause of neonatal infection

Table 19. Features of GBS Infections

Feature	Early Onset	Late Onset	Late-late Onset
Age range	< 7 days	7 days - 3 months	> 3 months
Median age of onset	1 hour	27 days	unknown
Incidence of prematurity	30%	uncommon	common
Clinical presentation	- sepsis ± signs of resp distress - meningitis (5-10%)	- sepsis ± signs of resp distress - meningitis (30%) - soft tissue, bone, joint localization	- in VLBW, premature and immunocompromised: bacteremia, sepsis, septic arthritis
Mortality rate	5-20%	2-6%	low

- ❑ treatment
  - initial suspected GBS infection: IV ampicillin and gentamicin until CSF or bloodstream sterility documented
  - upon confirmation of GBS: IV penicillin x 14 days (meningitis) to 4 weeks (endocarditis)
  - meningitis: repeat LP at 24 hours after initial treatment (controversial)

PERTUSSIS/WHOOPING COUGH

- ❑ Bordetella pertussis
- ❑ incubation: 6-20 days
- ❑ communicable from 1 week before paroxysms to 3 weeks after
- ❑ decreased incidence due to immunizations
- ❑ highly contagious; airborne —> transmitted via air droplets released during intense coughing

Clinical Features

- ❑ prodromal catarrhal stage
  - 1-2 weeks, most contagious
  - coryza, mild cough, low grade fever
- ❑ paroxysmal stage
  - 2-4 weeks
  - paroxysms of cough, sometimes followed by inspiratory whoop
  - +/– vomiting with coughing spells
  - can have severe symptoms for 6 weeks, cough for 6 months
  - pressure effect - subconjunctival hemorrhage, rectal prolapse, hernias, epistaxis
- ❑ convalescent stage
  - 1-2 weeks, noninfectious
  - occasional paroxysms of cough but decreased frequency and severity

Complications

- ❑ respiratory
  - secondary pneumonia (most common), otitis media
  - atelectasis
  - apnea (infants)
- ❑ neurological
  - seizures
  - encephalopathy (1:100 000)
  - intracranial hemorrhage

Diagnosis

- ❑ clinical: URTI symptoms followed by paroxysms of cough in an afebrile child
- ❑ lymphocytosis
- ❑ culture of nasopharyngeal swab or aspirate
- ❑ fluorescent antibody staining of pharyngeal specimen (most sensitive); PCR

Treatment

- ❑ supportive care is mainstay of treatment
- ❑ hospitalize if paroxysms of cough are associated with cyanosis and/or apnea
- ❑ erythromycin x 14 days
  - isolate until 5 days of treatment
  - treatment will decrease infectivity but not change course
  - shortens period of communicability
- ❑ chemoprophylaxis: erythromycin for all household contacts

INFECTIOUS MONONUCLEOSIS

- ❑ the “great imitator"
- ❑ Epstein-Barr virus (EBV)
- ❑ systemic viral infection that affects many organ systems
- ❑ transmission through saliva - “kissing disease”

Presentation

- ❑ tonsillar exudate
- ❑ lymphadenopathy
- ❑ fever
- ❑ +/– rash - pathognomonic rash with amoxicillin/ampicillin
- ❑ +/– hepatosplenomegaly
- ❑ any -itis, including arthritis, hepatitis, nephritis

Blood Picture

- ❑ atypical lymphocytes, lymphocytosis, Downey cells
- ❑ ± anemia
- ❑ ± thrombocytopenia
- ❑ heterophil antibody test (Monospot test) not sensitive in children < 4 years
- ❑ EBV titres

Treatment

- ❑ throat culture to rule out streptococcal pharyngitis
- ❑ bed rest, fluids, saline gargles for sore throat, acetaminophen
- ❑ if airway obstruction, admit - steroids
- ❑ avoid contact sports if organomegaly present
- ❑ resolves over 2-3 weeks although fatigue may persist

URINARY TRACT INFECTION

- ❑ see Urology Notes
- ❑ in newborns - more common in males
- ❑ in children - more common in females due to straight short urethra

Risk Factors

- ❑ female (after 2 years), neurogenic bladder, reflux, GU tract abnormalities, diabetes, immunocompromised, sexual intercourse, uncircumcised male, poor hygiene

Signs and Symptoms

- ❑ non-specific - fever, vomiting, irritability
- ❑ specific - dysuria, flank pain

Diagnosis

- ❑ MSU: > 10^5 colonies/ml of single organism OR catheter: > 10^3 colonies/ml OR
- ❑ suprapubic: any growth
- ❑ urine R&M diagnostic sensitivity: WBC 40%, bacteria 60%, WBC + bacteria 99%

Treatment

- ❑ hydration and antibiotics
- ❑ 7-10 days eg: TMP/SMX, amoxicillin/pivampicillin, nitrofurantoin, TMP
- ❑ if toxic, give IV initially (amp + gent/ceftriaxone/cefotaxime)
- ❑ prophylaxis if reflux, neurogenic bladder, recurrent UTIs (> 3 UTIs/year)
- ❑ later investigations
  - U/S and VCUG - for anatomical abnormalities, reflux
  - renal nuclear scans
- ❑ indications for investigations: < 1 year old with symptomatic UTI, all boys, all febrile UTIs with significant systemic symptoms
- ❑ prophylaxis if reflux, neurogenic bladder, recurrent UTIs (> 3 UTIs/year)

COMMON BENIGN NEONATAL CONDITIONS

- ❑ vascular instability (cutis marmorata, phlebectasia, acrocyanosis) may be normal particularly in premature infants
- ❑ vernix caseosa is a soft creamy white layer which is common in pre-term babies and disappears by term; in contrast to post-term in which peeling of extremeties is common
- ❑ Mongolian spots are bluish black macules over lower back and buttocks seen commonly in Negroid, Indian and Asian infants (may look like bruises)
- ❑ capillary hemangioma is a raised red lesion which increases in size after birth and generally resolve between 1-4 years of age
- ❑ erythema toxicum is an erythematous papular-vesicular rash which is self-limited
- ❑ pustular melanosis is defined by brown macular base with dry vesicles more common in Negroid infants

DIAPER DERMATITIS

- ❑ differential diagnosis
    - 1. irritant contact dermatitis
    - 2. seborrheic dermatitis
    - 3. candidiasis
    - 4. psoriasis

Primary Irritant Dermatitis

- ❑ intertriginous areas not involved (differentiates from candida)
- ❑ chemical irritation (urine, feces) – very common
- ❑ seen in infants with diarrhea or home diapering

Treatment

- ❑ use disposable diapers
- ❑ 1% hydrocortisone cream
- ❑ use protective ointments e.g. vaseline, zinc oxide

SEBORRHEIC DERMATITIS

- ❑ usually appears in the first few days of life
- ❑ thick yellow greasy scale
- ❑ sites include scalp (cradle cap), eyebrows, nose, diaper area including intertriginous areas
- ❑ non-pruritic
- ❑ usually happy baby

Treatment

- ❑ scale removal with oils and physical means, tar shampoos, hydrocortisone

CANDIDA

- ❑ red confluent lesions with “satellite" lesions
- ❑ intertriginous areas involved (distinguish from diaper dermatitis)
- ❑ may have concomitant oral thrush

Treatment

- ❑ topical antifungal

ITCHY ERUPTIONS IN CHILDHOOD

1. Atopic dermatitis
2. Contact dermatitis
3. Scabies
4. Urticaria
5. Bites (mosquito, flea)
6. Chicken pox

ATOPIC DERMATITIS (ECZEMA)

- ❑ family history positive for atopy (asthma, allergy, ASA sensitivity)
- ❑ those affected thought to have a decreased threshold for pruritis and for reaction to irritants
- ❑ serum IgE levels are higher in 80-85% of those affected

Clinical Stages	Location
infantile (3 months to 3 years)	face and extensors of lower legs
childhood (3 years to puberty)	flexural areas
adult (puberty onwards)	diffuse on face and extremities

- ❑ diagnostic criteria include
  - characteristics of lesions (acute and chronic)
  - follows typical distribution
  - chronic relapsing course
  - family history of atopy
- ❑ acutely: erythema, vesicles, exudate and crusts, pruritis
- ❑ chronic: scaling, xerosis, lichenification and pigment changes
- ❑ prognosis – approximately 75% have remission by adolescence

Treatment
- ❑ general: stress chronicity of illness; prevent scratching by physical means
- ❑ specific therapy
  - topical steroids: hydrocortisone 1% to face and folds, medium strength on rest of body (no systemic steroids)
  - antihistamines are effective against pruritis
  - skin hydration by vaseline application while wet
  - skin hygiene to prevent infection
  - avoid harsh soaps, chemicals, perfumes, wool, etc.
- ❑ systemic medication
  - antihistamines; antibiotics when infected
  - do not use systemic steroids

Complications
- ❑ secondary infection (Staph, herpes simplex)

IMPETIGO
- ❑ contagious infection by S. *aureus* and Group A Strep
- ❑ honey-coloured, crusting erosions - *Streptococcus*
- ❑ may have bullous lesions (bullous impetigo) - *Staphylococcus*
- ❑ occurs on exposed areas (face)
- ❑ satellite lesions by autoinoculation
- ❑ non-pruritic

Treatment
- ❑ topical antibiotics (fucidin/bactroban)
- ❑ penicillin, erythromycin, cephalexin
- ❑ local crust removal
- ❑ careful hygiene to prevent spread

Complications
- ❑ local cellulitis
- ❑ post-streptococcal glomerulonephritis

SCABIES
- ❑ very itchy papules; hand and feet commonly involved
- ❑ track marks (S-shaped burrows)
- ❑ infants or immunosuppressed patients can get very severe scabies (sparing of head and neck in adults)
- ❑ may have excoriations, honey-coloured crusts and pustules from secondary infection

Treatment
- ❑ premethrin (Nix) or gamma benzene hexachloride/lindane
- ❑ precipitated sulfur
- ❑ treat family and contacts
- ❑ antihistamine e.g. hydroxyzine (Atarax) or diphenhydramine (Benadryl)

ERYTHEMA MULTIFORME MINOR (80%)
- ❑ 1-2 cm erythematous papules; center clears to a purpuric or cyanotic lesion i.e. target lesions
- ❑ symmetrical; common to dorsum of hands/feet, elbows, knees and face
- ❑ may have mild mucous membrane involvement
- ❑ no systemic signs

Etiology
- ❑ idiopathic (most common)
- ❑ infectious – HSV implicated
- ❑ drugs

Treatment
- ❑ attempt to identify agent, symptomatic
- ❑ no antihistamines, NSAIDs or salicylates necessary

Prognosis
- ❑ self-limited

ERYTHEMA MULTIFORME MAJOR (STEVENS-JOHNSON SYNDROME) (20%)

- ❑ lesions of EM minor plus bullous lesion with mucous membrane involvement (oral, nasal, conjunctival and genital)
- ❑ etiology: drugs (sulfa, phenytoin, penicillin, phenobarbital)
- ❑ may have non-specific viral prodrome
- ❑ treatment: supportive-IV fluids, analgesia, ophthalmology consult, prophylactic antibiotics, systemic steroids controversial

PEDIATRIC EXANTHEMS

Table 20. Pediatric Exanthems

Disease	Incubation	Infectivity	Spread	Clinical S/SX	Complications
roseola (HHV-6, others)	5-15 days	unknown	unknown	high fever x 72 hours mild rash on trunk after defervescence, spreads to neck	febrile seizures
rubella (rubivirus)	14-21 days	7 days pre-rash and 5 days post	droplet	fever and 3 day pink descending maculopapular rash, initially discrete. Sub-occipital-lymphadenopathy	arthritis, thrombocytopenia (rare), encephalitis (rare)
measles (morbillivirus)	10-14 days	4 days pre-rash	droplet	fever, cough, coryza, conjuctivitis x 72 hours as prodrome, Koplik's spots, then red maculopapular confluent rash (face to feet)	secondary bacterial infection, acute otitis media, bronchopneumonia, encephalitis, SSPE
varicella	10-21 days	1-2 days pre-rash until all vesicles have crusted	droplet and direct contact	prodrome variable from none to low grade fever and malaise, maculopapular rash on trunk progresses to vesicles, then to crusts	pneumonia, encephalitis, cerebellar ataxia, ITP, dissemination and death in immunosuppressed, herpes zoster, Reye syndrome
mumps (paramyxovirus)	12-25 days	7 days pre-parotitis, 7 days post-parotitis occasionally abdominal pain due to pancreatitis	droplet	uni- or bilateral parotitis +/– mild resp symptoms	meningoencephalitis, pancreatitis, orchitis, sterility, labyrinthitis, deafness
erythema infectiosum (parvovirus)	4-14 days	unknown	?droplet	usually no prodromal symptoms, sudden appearance of livid erythema on cheeks, progressing to macuopapular rash on trunk and extremities, later lacy appearance, duration 3-5 weeks	increased fetal wastage *in utero*, aplastic crisis in patients with chronic hemolytic anemia eg. sickle cell, arthritis, vasculitis

HEART MURMURS

- ❑ 50-80% of children have audible heart murmurs at some point in their lives
- ❑ most murmurs are functional (i.e. "innocent") without associated structural abnormalities
- ❑ murmurs can become audible or accentuated in high output states, e.g. fever

Table 21. Differentiating Innocent and Pathological Heart Murmurs

	Innocent	Pathological
history and physical	asymptomatic	symptoms and signs of cardiac disease
timing	systolic ejection murmur (except venous hum)	all diastolic, pansystolic or continuous
grade	≤ 2/6	> 2/6
splitting	physiologic S2	fixed splitting or single S2
extra sounds/clicks	none	present
change of position	murmur varies	unchanged

Table 22. Five Innocent Heart Murmurs

Type	Description	Differential Diagnosis
Still's murmur	vibratory, LLSB or apex	subaortic stenosis, small VSD
pulmonary ejection	soft, blowing, ULSB	ASD, PS
venous hum	infraclavicular hum, continuous, R > L	PDA
supraclavicular arterial bruit	low intensity, above clavicles	AS, bicuspid aortic valve
peripheral pulmonic stenosis	neonates, low-pitched radiates to axilla and back	PDA, PS

CONGENITAL HEART DISEASE

- ❑ 8/1000 live births, can present with heart murmur, heart failure, or cyanosis
- ❑ increased risk
  - maternal factors
    - diabetes, phenylketonuria
    - medication, alcohol or drug use
    - infection (e.g. rubella, CMV)
  - infant factors
    - prematurity (e.g. PDA)
    - chromosomal abnormalities (e.g. Down syndrome)
    - positive family history (2-4% risk if sibling affected)
- ❑ most common lesion: VSD
- ❑ congenital heart disease can be categorized as:
  - L to R shunts: e.g. VSD, ASD, PDA, endocardial cushion defect
  - cyanotic e.g. Tetralogy of Fallot, Transposition of Great Arteries (TGA)
  - obstructive lesions: e.g. aortic stenosis, pulmonic stenosis, coarctation of aorta, hypoplastic left heart syndrome
- ❑ subacute bacterial endocarditis (SBE) prophylaxis should be given to all patients with congenital heart disease except those with an isolated secundum ASD, corrected VSD or PDA without residua at greater than 6 months after repair, or mitral valve prolapse without mitral regurgitation

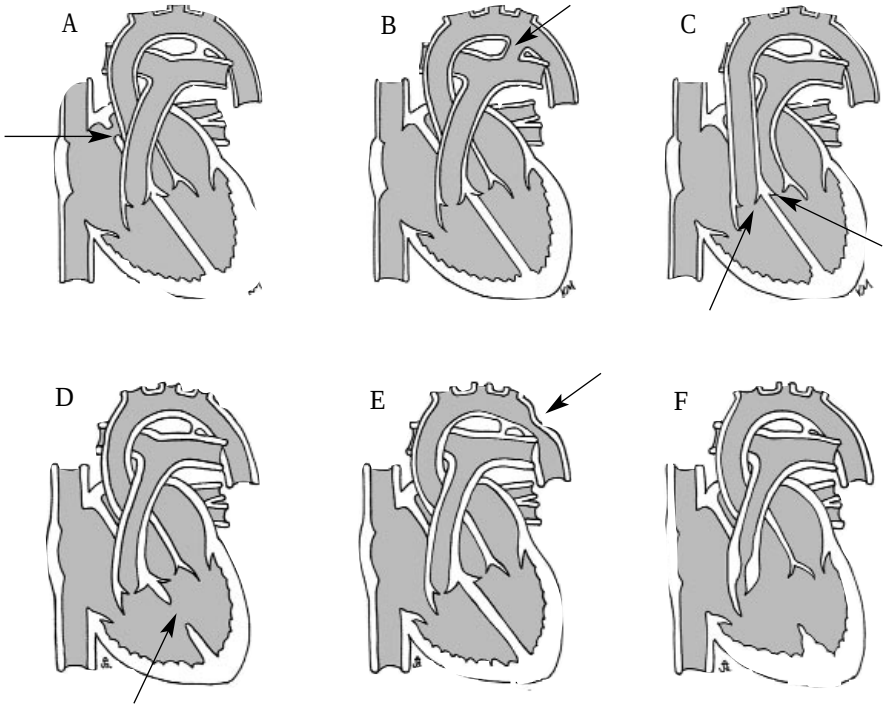


Figure 7. Common Congenital Heart Diseases

Drawing by Kevin Millar and Jacquelyn Shaw

LEFT TO RIGHT SHUNT LESIONS

- ❑ extra blood is displaced through a communication from the left to the right side of the heart, resulting in increased pulmonary blood flow
- ❑ shunt volume dependent upon three factors: size of defect, pressure gradient between chambers or vessels, peripheral outflow resistance
- ❑ untreated shunts can result in pulmonary vascular disease, RVH, and R to L shunts

Atrial Septal Defect (ASD)

- ❑ three types
  - ostium primum - common in Down syndrome
  - ostium secundum - most common type (50-70%)
  - sinus venosus - defect located at entry of SVC into right atrium
- ❑ often asymptomatic in childhood
- ❑ murmur: often grade II-III/VI pulmonic outflow murmur with widely split and fixed S2
- ❑ ECG: RAD, mild RVH, RBBB
- ❑ CXR: increased pulmonary vasculature
- ❑ natural history: 80-100% spontaneous closure rate if ASD diameter < 8 mm
- ❑ if remains patent, CHF and pulmonary HTN can develop in adult life
- ❑ management: elective surgical or catheter closure (low risk procedures) between 2-5 years of age

Ventricular Septal Defect (VSD)

- ❑ most common congenital heart defect (30-50%)
- ❑ small VSD (majority)
  - asymptomatic, normal growth and development
  - murmur: early systolic to holosystolic, best heard at LLSB
  - ECG and CXR are normal
  - most close spontaneously, does not need surgical closure even if remains patent
- ❑ moderate to large VSD
  - delayed growth and development, decreased exercise tolerance, recurrent URTIs or "asthma" episodes, CHF
  - murmur: holosystolic at LLSB with thrill, mid-diastolic rumble at apex
  - ECG: LVH, LAH, RVH
  - CXR: increased pulmonary vasculature, cardiomegaly, CHF

- natural history: secondary pulmonary HTN, CHF by 2 months of age
- management: treatment of CHF; surgical closure

Patent Ductus Arteriosus (PDA)
- ❑ patent vessel between descending aorta and pulmonary artery
- ❑ 5-10% of all congenital heart defects
- ❑ common in premature infants (1/3 of infants < 1750 grams)
- ❑ may be asymptomatic or have apneic or bradycardic spells, exertional dyspnea
- ❑ associated tachycardia, bounding pulses, hyperactive precordium, wide pulse pressure
- ❑ murmur: continuous "machinery" murmur, best heard at left infraclavicular area
- ❑ ECG: may show LVH, RVH
- ❑ CXR: normal to mildly enlarged heart, increased pulmonary vasculature
- ❑ diagnosis by echocardiography
- ❑ natural history: spontaneous closure common in premature infants, less common in term infants
- ❑ management: indomethacin, surgical ligation, or catheter closure
- ❑ high risk of SBE, antibiotic prophylaxis required until 6 months after closure

Endocardial Cushion Defect
- ❑ spectrum from endocardial cushion VSD and ostium primum ASD to complete AV canal with common AV valve
- ❑ commonly associated with Down syndrome
- ❑ natural history depends on size of defect and valvular involvement
- ❑ complete AV canal require early complete surgical repair, preferably before 3 months of age

CYANOTIC CONGENITAL HEART DISEASE
- ❑ systemic venous return re-enters systemic circulation directly
- ❑ most prominent feature is cyanosis (O_2 sat < 75%)
- ❑ differentiate between cardiac and other causes of cyanosis with hypoxia test
- ❑ survival depends on mixing via shunts (e.g. ASD, VSD, PDA)

Transposition of the Great Arteries
- ❑ most common cardiac lesion in the cyanotic newborn
- ❑ aortic root arises anteriorly from the right ventricle and the main pulmonary artery arises posteriorly from left ventricle, resulting in parallel pulmonary and systemic circulations (Figure 8)
- ❑ newborn presents with progressive cyanosis unresponsive to oxygen therapy as the ductus arteriosus closes and mixing between the two circulations diminishs; severe hypoxemia, acidosis, and death can occur rapidly
- ❑ if VSD present, cyanosis is not prominent, infant presents with CHF after a few weeks of life
- ❑ murmur: none or grade II/VI SEM
- ❑ ECG: RAD, RVH
- ❑ CXR: egg-shaped heart with narrow mediastinum ("egg on a string")
- ❑ management:
  - prostaglandin E1 infusion to keep ductus open
  - balloon atrial septostomy with catheter
  - surgical correction: arterial switch procedure

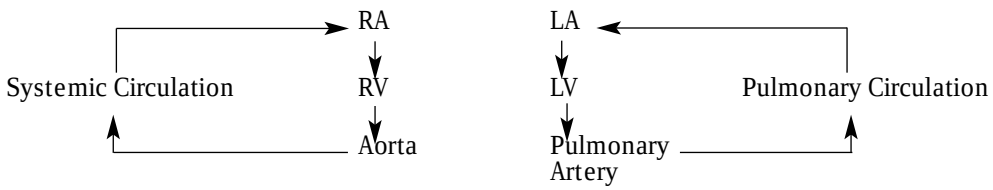


Figure 8. Parallel Circulations of TGA

Tetralogy of Fallot
- ❑ 10% of all congenital heart defects, most common cyanotic heart defect beyond infancy
- ❑ embryologically a single defect with hypoplasia of the conus causing:
  - VSD
  - RV outflow tract obstruction (RVOTO)

- overriding aorta
- RVH

❑ direction and degree of shunt are functions of the relative outflow resistance
❑ infants may initially have a left to right shunt and therefore are not cyanotic but the RVOTO is progressive, resulting in increasing right to left shunting with hypoxemia and cyanosis
❑ "tet" spells
- caused by increased right to left shunting due to exercise or crying which decreases systemic resistance
- paroxysm of rapid and deep breathing, irritability and crying
- increased cyanosis and decreased intensity of murmur
- peak incidence at 2-4 months of age
- if severe may lead to seizures, loss of consciousness, death (rare)
- management: oxygen, knee-chest position, morphine sulfate, propanolol

❑ murmur: single loud S2 due to severe pulmonic stenosis
❑ ECG: right axis deviation, RVH
❑ CXR: boot shaped heart, decreased pulmonary vasculature, right aortic arch
❑ management: surgical repair including closure of VSD and widening of RVOTO

Clinical Pearl
❑ Characteristic Chest X-Ray Findings in Congenital Heart Disease
Boot-Shaped Heart - Tetralogy of Fallot, tricuspid atresia
Egg-Shaped Heart - Transposition of Great Arteries
"Snowman" Heart - Total Anamolous Pulmonary Venous Return

OBSTRUCTIVE LESIONS

❑ present with pallor, decreased urine output, cool extremities and poor pulses

Coarctation of the Aorta

❑ narrowing of aorta almost always at the level of the ductus arteriosus
❑ commonly associated with bicuspid aortic valve (50%)
❑ if severe, presents with shock in the neonatal period when the ductus closes
❑ often asymptomatic with upper extremity systolic pressures of 140-145 mm Hg
❑ weak pulses, decreased blood pressure in lower extremities, radial-femoral delay
❑ if associated with other lesions (e.g. PDA, VSD), can cause CHF
❑ murmur: absent or systolic with late peak at apex, left axilla, left back
❑ management: balloon arterioplasty or surgical correction
❑ complications: essential hypertension

Aortic Stenosis

❑ valvular (75%), subvalvular (20%), supravalvular and idiopathic hypertrophic subaortic stenosis (IHSS) (5%)
❑ often asymptomatic but may be associated with CHF, exertional chest pain, syncope or sudden death
❑ murmur: SEM at URSB with aortic ejection click at the apex
❑ management: surgical or balloon valvuloplasty, repeated interventions and valve replacement may be necessary
❑ SBE prophylaxis and exercise restriction required

Pulmonary Stenosis

❑ valvular (90%), subvalvular or supravalvular
❑ usually part of other congenital heart lesions (e.g. Tetralogy of Fallot) or in association with other syndromes (e.g. congenital rubella, Noonan syndrome)
❑ critical pulmonic stenosis: inadequate pulmonary blood flow, dependent on ductus for oxygenation, progressive hypoxia and cyanosis
❑ presentation varies from asymptomatic to CHF
❑ murmur: wide split S2 maximal on expiration, SEM at ULSB, pulmonary ejection click
❑ ECG: RVH
❑ CXR: dilated poststenotic pulmonary artery
❑ management: balloon valvuloplasty

Hypoplastic Left Heart Syndrome

❑ a spectrum of hypoplasia of left ventricle, atretic mitral and/or aortic valves, small ascending aorta, coarctation of the aorta with resultant systemic hypoperfusion
❑ most common cause of death from congenital heart disease in first month of life
❑ presents with circulatory shock and metabolic acidosis on closure of the ductus

- ❑ management
  - intubate and correct metabolic acidosis
  - IV infusion of PGE1 to keep ductus open
  - treatment options
    - surgical correction (overall survival 50% to late childhood)
    - transplantation
    - no treatment

CONGESTIVE HEART FAILURE

Etiology
- ❑ congenital heart defects
- ❑ ateriovenous malformations
- ❑ cardiomyopathy
- ❑ arrhythmias
- ❑ acute hypertension
- ❑ anemia
- ❑ cor pulmonale

Pathophysiology
- ❑ see Cardiology Notes

Symptoms
- ❑ infant: feeding difficulties, easy fatigability, exertional dyspnea, diaphoresis when sleeping or eating, respiratory distress, vomiting, lethargy, cyanosis
- ❑ child: decreased exercise tolerance, fatigue, decreased appetite, failure to thrive, respiratory distress, syncope, frequent URTIs or "asthma" episodes
- ❑ orthopnea, paroxysmal nocturnal dyspnea, edema are uncommon in children

Physical Findings
- ❑ four key features: tachycardia, tachypnea, cardiomegaly, hepatomegaly (2 tachy's, 2 megaly's)
- ❑ failure to thrive
- ❑ respiratory distress, wheeze, crackles, cyanosis and clubbing
- ❑ alterations in peripheral pulses, four limb blood pressures
- ❑ dysmorphic features associated with congenital syndromes

Management
- ❑ general: sitting up, oxygen, sodium and water restriction, increased caloric intake
- ❑ pharmacologic: diuretics, inotropic agents, afterload reduction
- ❑ correction of underlying cause

INFECTIVE ENDOCARDITIS
- ❑ see also Cardiology Notes
- ❑ 10-15% of cases are culture negative
- ❑ Osler's nodes, Janeway's lesions, splinter hemorrhages are late findings in children
- ❑ antibiotic prophylaxis for prevention is necessary for all patients with:
  - congenital heart disease (except for isolated secundum ASD)
  - rheumatic valve lesions
  - prosthetic heart valves
  - surgical shunts
  - previous endocarditis
  - pacemaker leads

DYSRHYTHMIAS
- ❑ see also Cardiology Notes
- ❑ can be transient or permanent, congenital (structurally normal or abnormal) or acquired (toxin, infection)

Sinus Arrhythmia
- ❑ phasic variations with respiration
- ❑ heard in almost all normal children

Premature Atrial Contractions
- ❑ may be normal variant or can be caused by electrolyte disturbance, hyperthyroidism, cardiac surgery, digitalis toxicity

Premature Ventricular Contractions (PVCs)
- ❑ common in adolescents
- ❑ benign if single, uniform, disappear with exercise, no associated structural lesions
- ❑ if not benign, may degenerate into more severe dysrhythmias

Supraventricular Tachycardia (SVT)
- ❑ most frequent sustained dysarrhythmia in children
- ❑ not lifethreatening but can lead to symptoms
- ❑ caused by re-entry via accessory connection, AV node most common site
- ❑ characterized by a rate of greater than 210 bpm
- ❑ treatment: vagal manouver, adenosine, digoxin (except in WPW)

HEMATOLOGY

APPROACH TO ANEMIA

History
- ❑ acute anemia: poor exercise tolerance, headache, fatigue, syncope
- ❑ chronic anemia: usually well tolerated
- ❑ diet history; milk excess —> iron deficiency anemia
- ❑ melena/hematochezia —> blood loss —> iron deficiency anemia
- ❑ family history of cholecystectomy or splenectomy —> hereditary hemolytic disorder
- ❑ ethnic origin —> thalassemia, sickle cell anemia
- ❑ exposure to oxidant drugs (sulpha drugs) —> G6PD deficiency
- ❑ underlying chronic illness (renal, hepatic, inflammatory)
- ❑ social history —> lead intoxication increased in older housing

Physical Exam
- ❑ heart rate, blood pressure, orthostatic changes
- ❑ flow murmur, pallor, level of activity
- ❑ jaundice —> hemolysis
- ❑ petechiae, purpura —> bleeding tendency
- ❑ hepatomegaly, splenomegaly —> infiltrative disorder
- ❑ failure to thrive —> chronic disease, organ failure
- ❑ stool —> occult blood

Table 23. Differential Diagnosis of Anemia

microcytic
- iron deficiency
  - blood loss or dietary lack
- thalassemia trait
- chronic inflammation
- sideroblastic anemia
- lead poisoning

normocytic

low reticulocyte count
- bone marrow infiltration
- transient erythroblastopenia of childhood
- chronic disease
- aplastic crisis

high reticulocyte count
- blood loss
- hemolysis
  - extrinsic
    - antibody-mediated
    - fragmentation: DIC, HUS, heart valve
  - intrinsic
    - membrane disorders: spherocytosis
    - enzyme deficiencies: G6PD
    - hemoglobin disorders: thalassemia

macrocytic
- folic acid deficiency
- vitamin B12 deficiency
- hypothyroidism
- liver disease

PHYSIOLOGIC ANEMIA
- ❑ elevated hemoglobin (> 170 g/L) and reticulocyte count at birth result of relatively hypoxic environment in utero
- ❑ after birth, levels start to fall due to shorter RBC lifespan, decreased RBC production, and increasing blood volume secondary to growth
- ❑ lowest levels at 6-12 weeks age (earlier in premature infants), about 100 g/L, levels rise again after 3 months
- ❑ no treatment required if asymptomatic

IRON DEFICIENCY ANEMIA
- ❑ most common cause of childhood anemia (see Colour Atlas E1)
- ❑ premature infants at increased risk - low iron stores at birth

Etiology
- ❑ dietary, typically between 6-24 months age, particularly in bottlefed infants receiving large volumes of cow's milk
- ❑ blood loss or malabsorption
- ❑ beware iatrogenic blood loss through repeated blood sampling (especially in neonates)
- ❑ cow's milk/cow's milk-based formula may result in blood loss and protein-losing enteropathy secondary to GI inflammation

Prevention
- ❑ for breast-fed infants after 6 months, give iron-fortified cereals and iron-rich foods
- ❑ if not breast fed, give iron-fortified formula from birth
- ❑ premature infants should start iron supplements at 6-8 weeks of age and continue until 1 year old

Management
- ❑ determine cause
- ❑ oral iron therapy - black stools suggest compliance
  - subjective improvement in 24-48 hours
  - increased reticulocyte count in 48-72 hours
  - increased hemoglobin in 4-30 days
  - repletion of iron stores in 1-3 months

SICKLE CELL DISEASE
- ❑ describes syndrome of hemoglobin SS, S-C and rare variants
- ❑ identification of specific genotypes important due to differences in frequency, type, and severity of clinical complications

Pathophysiology
- ❑ red blood cells sickle with low pO_2, dehydration, fever, acidosis
- ❑ acute intravascular sickling results in infarction of tissue
- ❑ hemolysis causes chronic, well-compensated, severe anemia; not routinely transfusion dependent (see Colour Atlas E5)
- ❑ increased incidence in Blacks and Mediterraneans

Presentation
- ❑ trait —> asymptomatic ± microscopic hematuria
- ❑ disease —> after 6-9 months age with fall in fetal Hgb, anemia, jaundice, splenomegaly

Types of Crises (usually have more than 1 crisis by age 1)
- ❑ vaso-occlusive crises - in any organ, most commonly in long bones of arms and legs, chest, abdomen, CNS, dactylitis (swollen hands and feet) in young children
- ❑ aplastic crisis - transient RBC aplasia after parvovirus B19 infection of red cell precursors in bone marrow
- ❑ splenic sequestration - sickling in spleen, large pooling of blood with acute fall in hemoglobin, shock

Functional Asplenia
- ❑ splenic dysfunction as early as 4 months, usually by 5 years
- ❑ susceptible to infection by encapsulated organisms, especially *Streptococcus pneumoniae*
- ❑ requires prophylactic oral penicillin daily, pneumococcal vaccine, and immediate evaluation of fever

Management
- ❑ acute
  - supportive and symptomatic
  - fluids, analgesia, exchange transfusions
  - oxygen if respiratory distress or chest crisis
  - incentive spirometry
- ❑ chronic
  - early aggressive treatment of infections, prophylactic antibiotics
  - pneumococcal, meningococcal, H. *influenzae*, Hepatitis B, and influenza vaccines
  - folate supplementation
  - hydroxyurea
  - chronic transfusion program if history of stroke
  - genetic counselling and education

SPHEROCYTOSIS

- ❑ red cell membrane disorder, causes a sphering of red blood cells which are removed by the spleen (see Colour Atlas E16)
- ❑ genetics
  - autosomal dominant
  - may have positive family history but high spontaneous mutation rate
- ❑ clinical severity can range from well-compensated, mild hemolytic anemia to severe hemolytic anemia with growth failure, splenomegaly, and chronic transfusion requirements in infancy
- ❑ management
  - splenectomy as needed
  - genetic counselling

GLUCOSE-6-PHOSPHATE DEHYDROGENASE (G6PD) DEFICIENCY

- ❑ X-linked recessive, different variants of the disease
- ❑ higher prevalence in Mediterraneans, Blacks, Orientals
- ❑ enzyme deficient red blood cells are unable to defend against oxidant stress (infection, drugs) and forms Heinz bodies (denatured hemoglobin) which are phagocytosed by splenic macrophages, creating "bites" on cells
- ❑ presents with acute hemolytic anemia with jaundice and dark urine
- ❑ management: supportive, hydration, transfusion, phototherapy
- ❑ prevention: avoid known oxidants e.g. fava beans, ASA, antimalarials, sulfonamides, infections

BLEEDING DISORDERS (see Hematology Notes)

Coagulation Defects

- ❑ characterized by deep bleeding into joints and muscles
- ❑ large spreading ecchymotic lesions and hematoma

Platelet Abnormalities

- ❑ characterized by petechiae, purpura, bruises, mucocutaneous bleeding, bleeding from superficial cuts (i.e. epitaxis, gum bleeding menorrhagia)

Table 23. Classification of Bleeding Disorders

	Mechanism	Examples
Blood Vessels	vasculitis	HSP
Platelets	low production	drugs, marrow infiltration, leukemia
	high destruction	ITP, infection, drugs
	high consumption	DIC, giant hemangioma, hypersplenism
	dysfunctional	vW disease, drugs (ASA), uremia
Coagulation Pathway	Vitamin K deficiency	hemorrhagic disease of newborn
	Factor VIII deficiency	Hemophilia A
	Factor IX deficiency	Hemophilia B
	abnormal vWF	vonWillebrand's disease

Immune Thrombocytopenia Purpura of Childhood (childhood ITP)

- ❑ peak age: 2-6 years, M=F
- ❑ usually follows an acute viral infection, rarely a presenting symptom of autoimmune disease e.g. SLE
- ❑ caused by antibodies that bind to platelet membranes
- ❑ splenic destruction of antibody-coated platelets
- ❑ typically presents 1-4 weeks after viral illness with sudden onset of petechiae, purpura, epitaxis in an otherwise well child
- ❑ self-limited in children; spontaneous recovery in 80% of cases
- ❑ differential diagnosis: drug-induced thrombocytopenia, HIV, leukemia, infection (viral), SLE

- ❑ clinically: no lymphadenopathy, no hepatosplenomegaly
- ❑ labs: thrombocytopenia with normal RBC, WBC
- ❑ if atypical presentation, do bone marrow to rule out leukemia
- ❑ management: consider prednisone or IVIG if clinically bleeding or severe thrombocytopenia, splenectomy only for life-threatening bleeding

Neonatal Thrombocytopenia

- ❑ transplacental passage of maternal antiplatelet antibodies
- ❑ two types
  - neonatal alloimmune thrombocytopenia (NAIT)
    - mother mounts immune response against antigens on fetal platelets
    - suspect in thrombocytopenic newborn who is otherwise well, normal maternal platelets, no history of maternal autoimmune disease or ITP
    - diagnosis: maternal serum (with immunoglobulins) reacts with father or child's platelets
    - treatment: transfusion of infant with washed maternal platelets
  - neonatal ITP
    - caused by antiplatelet antibodies from maternal ITP
    - similar presentation to NAIT but must distinguish, if infant is transfused with maternal platelets, the transfused platelets will also be destroyed
    - treatment: steroids to mother x 10-14 days prior to delivery or IVGG to mother defore delivery or to infant after delivery

Hemorrhagic Disease of the Newborn

- ❑ caused by vitamin K deficiency
- ❑ factors II, VII, IX, X are vitamin K-dependent, therefore both PT and PTT are abnormal
- ❑ presents at 2-7 days of life with generalized ecchymoses, GI hemorrhage, bleeding from a circumcision or umbilical stump
- ❑ prevention: vitamin K administration at birth to all newborns

Hemophilia A: Factor VIII Deficiency

- ❑ X-linked recessive, 5 times more common than Hemophilia B
- ❑ lack of factor VIII delays formation of thrombin which is crucial to forming a normal, functional fibrin clot and solidifying the platelet plug at areas of vascular injury
- ❑ severity determined by level of factor VIII, severity of bleeds, and presence of antibodies to factor VIII
  - severe hemophilics (<1% factor VIII) have spontaneous bleeding or bleeding from minor trauma and manifests in infancy, hallmark: hemarthrosis
  - mild hemophilics (>5% factor VIII) have bleeding with significant trauma (e.g. surgery) and may go undiagnosed for many years
- ❑ DDAVP for mild disease, factor VIII replacement

Hemophilia B (Christmas Disease): Factor IX Deficiency

- ❑ X-linked recessive, treated with factor IX replacement or plasma
- ❑ presentation same as Hemophilia A

von Willebrand's Disease

- ❑ defect: variable abnormality in von Willebrand factor (vWF)
- ❑ vWF is an adhesive protein that bridges subendothelial collagen and platelets, and protects factor VIII from rapid clearance
- ❑ autosomal dominant (more common, mild) or autosomal recessive (rarer, more severe)
- ❑ presents with mucocutaneous bleeding, epistaxis, gingival bleeding, ecchymosis, menorrhagia
- ❑ abnormal PTT and bleeding time
- ❑ DDAVP for mild disease (increases release of vWF), cryoprecipitate

Table 24. Evaluation of the Child with Abnormal Bruising/Bleeding

	BT	PT	PTT	VIII:C	vWF	Platelets	Fibrinogen
hemophilia A	N	N	↑	↓	N	N	N
hemophilia A	N	N	↑	N	N	N	N
vonWillebrand's	↑	N	N or ↑	↓	↓	N	N
DIC	N or ↑	↑	↑	↓	N	↓	↓
vit K deficiency	N	↑	↑	N	N	N	N
thrombocytopenia	↑	N	N	N	N	↓	N

BT=bleeding time, VIII:C=factor VIII coagulant activity

❑ extensive bruising in the absence of lab abnormalites: consider child abuse

ONCOLOGY

❑ cancer is second most common cause of death in children after 1 year of age (#1=injuries)
❑ usually occur sporadically, but increased risk with
- neurocutaneous syndromes
- chromosomal syndromes
- immunodeficiency syndromes
- prior malignancy
- family history
- exposure to radiation, chemicals, biologic agents

❑ leukemia (25-35%) and brain tumours (20%) most common
❑ some malignancies may be more prevalent in certain age groups
- newborns: neuroblastoma, congenital leukemia
- infancy and childhood: leukemia, neuroblastoma, Wilms', retinoblastoma
- adolescence: lymphoma, gonadal tumours, bone

LEUKEMIA

❑ most common childhood malignancy
❑ heterogenous group of diseases; types: ALL (80%), AML (15%) and CML (5%)
❑ etiology unknown; EBV associated with African Burkitt lymphoma, retrovirus with T cell leukemia
❑ signs and symptoms due to infiltration of leukemic cells into bone marrow (bone pain, anemia, neutropenia, thrombocytopenia) and into tissues (lymphadenopathy, hepatosplenomegaly, CNS, testes)
❑ prognosis: low-risk - 90% long-term remission, high-risk - 70% long-term remission
❑ see also Hematology Notes

Table 25. Prognostic Indicators in Childhood Acute Lymphocytic Leukemia

	Good	Poor
age	2-10 years	<2 or >10 years
ethnicity	white	black
sex	female	male
lymphadenopathy	no	yes
hepatosplenomegaly	no	yes
mediastinal mass	no	yes
initial WBC	< 20 x 10^9/L	> 20 x 109/L
hemoglobin	> 100 g/L	< 100 x g/L
LDH	low	high
lymphoblasts	typical	undifferentiated
hyperploidy	yes	no
translocation	no	yes

LYMPHOMA

- ❑ third most common childhood tumour
- ❑ Hodgkin's lymphoma
  - older children (age > 15), similar to adult Hodgkin's
  - presents with painless, firm lymphadenopathy
  - B symptoms only in 30% of children
- ❑ Non-Hodgkin's lymphoma
  - younger children (7-11 years)
  - rapidly growing tumour with distant metastases
  - signs and symptoms related to disease site, most commonly abdomen, chest (mediastinal mass), head and neck region
- ❑ see also Hematology Notes

BRAIN TUMOURS

- ❑ predominantly infratentorial involving cerebellum, midbrain, brainstem
- ❑ glial (astrocytomas most common) or primitive neuroectodermal (medulloblastoma, germ cell tumours, ependymothera)
- ❑ signs and symptoms
  - infratentorial: vomiting, morning headache, increased head circumference, ataxia, diplopia, nystagmus, papilledema
  - supratentorial: focal deficits, seizure, long tract signs
- ❑ evaluation
  - history, physical exam including complete neurological exam
  - CT and/or MRI of head as indicated
- ❑ see also Neurosurgery Notes

WILMS' TUMOUR (NEPHROBLASTOMA)

- ❑ mean age at diagnosis 3-3 1/2 years, M=F
- ❑ 5% of all childhood cancers
- ❑ 1/3 hereditary and 2/3 sporadic
- ❑ associated with a number of congenital abnormalities: sporadic anridia (often with 11p13 deletion), hemihypertrophy, genitourinary abnormalities
- ❑ presentation
  - 80% with asymptomatic abdominal mass
  - hypertension, hematuria
- ❑ differential diagnosis: hydronephrosis, polycystic kidney, renal cell carcinoma, neuroblastoma, lymphoma
- ❑ management
  - nephrectomy
  - staging
  - chemotherapy (pre- or post-op)
  - radiation
- ❑ generally good prognosis

NEUROBLASTOMA

- ❑ neural crest cell tumour arising from sympathetic tissues of the adrenal medulla (45%) or the sympathetic chain (25% retroperitoneal, 20% posterior mediastinal, 4% pelvis, 4% neck)
- ❑ most common malignancy in infancy, median age of onset 20 months

Presentation

- ❑ abdominal mass (most common), neck mass, chest mass (may be incidental finding on chest x-ray)
- ❑ direct extension: spinal cord compression, Horner syndrome
- ❑ metastases:periorbital ecchymosis, bone pain, hepatomegaly, "blueberry muffin" skin nodules
- ❑ paraneoplastic: hypertension, diarrhea (VIP secretion), opsoclonus, myoclonus

Diagnosis and Staging

- ❑ LFTs, renal function tests, serum ferritin
- ❑ VMA, HVA urine
- ❑ CT scan chest, abdomen
- ❑ bone scan
- ❑ bone marrow exam - for neuroblastoma cells in "rosettes"
- ❑ tissue biopsy

Management

- ❑ surgery, radiation, chemotherapy
- ❑ +/– bone marrow transplantation

Good Prognostic Factors
- ❑ < 1 year old
- ❑ female
- ❑ primary site - posterior mediastinum and neck
- ❑ stage I, II, IVS disease
- ❑ low serum ferritin
- ❑ VMA/HVA ratio > 1
- ❑ aneuploidy
- ❑ no N-myc oncogene amplification

RHEUMATOLOGY

EVALUATION OF LIMB PAIN

History
- ❑ pain: onset, duration, location, character, intensity, frequency, aggravating/alleviating factors, limitations in daily activity
- ❑ trauma, injury
- ❑ morning stiffness, limp, swelling/redness of joints
- ❑ general: fever, rash, fatigue, weight loss, cough, chest pain, hair loss
- ❑ family history: arthritis, psoriasis, IBD, bleeding disorders

Physical Exam
- ❑ complete physical exam
- ❑ all joints: inspection, palpation, range of motion
- ❑ gait, leg length discrepency
- ❑ tenderness on tendons or tendon insertion sites
- ❑ muscle weakness or atrophy

Investigations
- ❑ CBC, differential, smear, ESR
- ❑ X-rays of painful joints/limbs
- ❑ as indicated: ANA, RF, PTT, sickle cell prep, viral serology, immunoglobulins, complement, urinalysis, synovial analysis and culture

Table 26. Differential Diagnosis of Limb Pain

Cause	< 3 years	3-10 years	> l0 years
trauma	x	x	x
infection			
septic arthritis	x	x	x
osteomyelitis	x	x	x
inflammatory			
transient synovitis		x	
JRA	x	x	x
seronegative spondyloarthropathy			x
SLE			x
dermatomyositis		x	
Henoch-Schonlein Purpura		x	
anatomic/orthopedic			
Legg-Calve-Perthes disease		x	x
slipped capital femoral epiphysis			x
neoplastic			
leukemia	x	x	x
neuroblastoma	x	x	x
bone tumours		x	x
hematologic			
hemophilia	x	x	x
sickle cell anemia	x	x	x
pain syndromes			
growing pains		x	
fibromyalgia			x
reflex sympathetic dystrophy			x

GROWING PAINS

- ❑ age 2-12 years, M=F
- ❑ pain
  - poorly localized affecting shins, rarely calves
  - usually bilateral
  - occurs in evening or awakens child at night
  - responds to reassurance, massage or analgesics
  - resolves completely in the morning
- ❑ no associated systemic symptoms (e.g. fever)
- ❑ normal physical examination
- ❑ lab investigations not necessary if typical presentation

JUVENILE RHEUMATOID ARTHRITIS (JRA)

- ❑ a heterogenous group of conditions characterized by a persistent arthritis in childhood
- ❑ diagnosis
  - arthritis in at least one joint
  - lasts for at least 6 weeks
  - onset before the age of 16
  - other causes of arthritis excluded
- ❑ classification
  - defined by features/number of joints affected in the first 6 months of onset
  - systemic onset - fever at onset with arthritis appearing after
  - pauciarticular - 4 or less joints involved
  - polyarticular - 5 or more joints involved
- ❑ prognosis: ultimately good, 80% have good outcome, worst prognosis with systemic onset and polyarticular course

Table 26. Juvenile Arthritis Classification

	Systemic	Pauciarticular		Polyarticular	
		Type I	Type II	RF neg	RF pos
sex predominance	M=F	80% F	90% M	90% F	80% F
age of onset	any	< 5	> 8	< 5	> 8
Rheumatoid factor	neg	neg	neg	neg	100%
ANA	neg	60%	neg	25%	75%
HLA-B27	neg	neg	75%	neg	neg
eye involvement	neg	20%	neg	10-20%	neg
% of patients	20	30	15	25	10

Systemic (Still's Disease)

- ❑ high spiking fever (≥ 38.5˚C) for at least 2 weeks
- ❑ extra-articular features: erythematous “salmon-coloured” maculopapular rash, lymphadenopathy, hepatosplenomegaly, leukocytosis, thrombocytosis, anemia, serositis (pericarditis, pleuritis)
- ❑ arthritis may occur weeks to months later

Pauciarticular Type I

- ❑ most common subtype, peak age 2 years
- ❑ usually involves large joints: knee, ankle or elbow, rarely shoulder or hip
- ❑ often resolves without permanent sequelae
- ❑ prone to chronic iridocyclitis and uveitis, which, if untreated, may lead to permanent visual damage
- ❑ slit lamp exam should be done early in child presenting with joint swelling and then every 3 months if ANA positive

Pauciarticular Type II

- ❑ at onset, there is an asymmetrical peripheral arthritis usually confined to joints below the waist (hip, knees, ankles, feet)
- ❑ enthesitis (inflammation at tendon insertion sites) of Achilles tendon, patellar tendon, plantar fascia
- ❑ seronegative spondyloarthropathy may develop later in life
- ❑ family history of spondyloarthropathy, IBD or psoriasis

Polyarticular RF Negative
- ❑ often involves small joints of hands and feet, temporomandibular joint, sternoclavicular joint, distal interphalangeal joints, cervical spine
- ❑ patients who are ANA positive are prone to chronic uveitis

Polyarticular RF Positive
- ❑ similar to the aggressive form of adult rheumatoid arthritis
- ❑ severe, rapidly destructive, symmetrical arthritis of large and small joints
- ❑ associated with rheumatoid nodules at pressure points (elbows, knees)
- ❑ unremitting disease, persists into adulthood

Management
- ❑ children may complain very little about their pain and disability
- ❑ can develop contractures from guarding and disuse requiring night splints and aids
- ❑ exercise to maintain ROM and muscle strength
- ❑ multidisciplinary approach with OT/PT, social work, orthopedics, ophthalmology, rheumatology
- ❑ Ist line drug therapy: NSAIDs (naproxen, indomethacin available as suspensions)
- ❑ other options
    - methotrexate
    - corticosteroids - intra-articular, systemic, or topical eye drops
    - hydrochloroquine
    - IV gammaglobulin

HENOCH-SCHÖNLEIN PURPURA
- ❑ most common vasculitis of childhood
- ❑ peak incidence 4-10 years, M > F
- ❑ often have history of URTI 1-3 weeks before onset of symptoms
- ❑ features
    - skin: palpable, non-thrombocytopenic purpura in lower extremities and buttocks, edema, scrotal swelling
    - joints: arthritis/arthralgia involving large joints
    - GI: abdominal pain, GI bleeding, intussusception
    - renal: IgA nephropathy, hematuria, proteinuria, hypertension, acute renal failure in <5%, progressive renal failure in another 5%
- ❑ management
    - symptomatic, corticosteroids may relieve abdominal pain and edema
    - monitor for renal disease, may last a few years
- ❑ prognosis: self-limited disease in 90%

KAWASAKI DISEASE
- ❑ acute vasculitis of unknown etiology
- ❑ most common cause of acquired heart disease in children
- ❑ peak age < 5 years, Orientals > Blacks > Causasians, M > F

Diagnostic Criteria
- ❑ fever persisting 5 days or more and
- ❑ 4 of the following 5 features
    - bilateral nonpurulent conjunctivitis
    - red fissured lips, strawberry tongue, erythema of oropharynx
    - changes of the peripheral extremities
        - acute phase: erythema, edema of hands and feet, groin peeling
        - subacute phase: peeling from tips of fingers and toes
    - polymorphous rash
    - cervical lymphadenopathy > 1.5 cm in diameter
- ❑ exclusion of other diseases e.g. scarlet fever, measles
- ❑ atypical Kawasaki disease: less than 5 of 6 diagnostic features but coronary artery involvement

Associated Features
- ❑ anterior uveitis
- ❑ irritability, aseptic meningitis
- ❑ diarrhea, abdominal pain, mild hepatitis, gall bladder hydrops
- ❑ sterile pyuria
- ❑ arthritis, serous otitis media, pneumonia
- ❑ pericarditis, myocarditis, arrhythmias

Complications
- ❑ coronary artery vasculitis with aneurysm formation during subacute phase
- ❑ occurs in 20-25% of untreated children, 4-8% if receive IVGG within 10 days of fever onset
- ❑ risk factors for coronary disease: male, age < 1 or > 9 years, fever >10 days
- ❑ of those with aneuryms: 50% of aneurysms regress within 2 years, 20% develop stenosis with risk of MI
- ❑ children may have endothelial dysfunction with risk of early CAD

Management
- ❑ IV gammaglobulin (2 g/kg)
- ❑ high (antiinflammatory) dose of ASA while febrile
- ❑ low (antiplatelet) dose of ASA in subacute phase
- ❑ follow up with periodic 2D-echocardiograms

ENDOCRINOLOGY

DIABETES MELLITUS (see Endocrinology Notes)

Type I Diabetes
- ❑ insulin dependent, most common type in childhood
- ❑ prevalence: 1 in 500 children under 18 years of age
- ❑ etiology: genetic predisposition and environmental trigger leading to autoimmune destruction of the pancreas
- ❑ classic presentation: polyuria, polydipsia, polyphagia, weight loss; 25% present in diabetic ketoacidosis
- ❑ management
  - insulin, blood glucose monitoring
  - young children more susceptible to CNS damage with hypoglycemia with fewer benefits from tight control, hence target glucose range higher at 6-12 mmol/L
  - increasingly tighter control in older children, 4-8 mmol/L
  - diet, exercise
  - education, psychosocial support
- ❑ complications
  - hypoglycemia
    - cause: missed/delayed meals, excess insulin, increased exercise
    - complications: coma, seizures
  - hyperglycemia
    - cause: infection, stress, diet-to-insulin mismatch
    - complications: risk of diabetic ketoacidosis, long-term complications
  - diabetic ketoacidosis
    - cause: new-onset diabetes, missed insulin doses, infection
    - complications: dehydration, cerebral edema, decreased level of consciousness
  - long-term complications usually not seen in childhood
    - present 10-20 years after onset, related to metabolic control (HbA1c)
    - retinopathy, nephropathy, neuropathy

HYPOTHYROIDISM
- ❑ see also Endocrinology Notes

Congenital Hypothyroidism
- ❑ incidence: 1 in 4000 births
- ❑ usually caused by dysgenetic (agenesis or ectopic) malformation of the thyroid gland
- ❑ diagnosis through routine neonatal screening
- ❑ usually asymptomatic in neonatal period but may have:
  - prolonged jaundice
  - constipation
  - sluggish, coarse cry, lethargy, poor feeding
  - big tongue, coarse facial features, large fontenelle, umbilical hernia

- ❑ prognosis
  - excellent if treatment started within 1-2 months of birth
  - if treatment started after 3-6 months of age may result in developmental delay
- ❑ management: thyroxine replacement

Acquired Hypothyroidism
- ❑ most common: Hashimoto's thyroiditis (autoimmune destruction of the thyroid)
- ❑ signs and symptoms similar to hypothyroidism in adults, but also:
  - delayed bone age, decline in growth velocity, short stature
  - precocious puberty
  - does not cause permanent developmental delay

HYPERTHYROIDISM (see Endocrinology Notes)

Congenital Hyperthyroidism
- ❑ results from transplacental passage of maternal thyroid stimulating antibodies (mother with Grave's)
- ❑ clinical manifestations in the neonate may be masked by transplacental maternal antithyroid medication
- ❑ presents with tachycardia with CHF, irritability, craniosynostosis, poor feeding, FTT
- ❑ spontaneous resolution by 2-3 months of life as antibodies cleared
- ❑ management: propylthiouracil until antibodies cleared

Grave's Disease
- ❑ F:M = 5:1, peak incidence in adolescence
- ❑ results from thyroid stimulating antibodies as with adult Grave's
- ❑ may exhibit classic signs and symptoms of hyperthyroidism, but also:
  - personality changes
  - school difficulty
  - mood instability
- ❑ management similar to adults: anti-thyroid drugs (propylthiouracil), radioiodine reserved for older teens, surgical thyroidectomy

Clinical Pearl
- ❑ Children with a solitary thyroid nodule require prompt evaluation as 30-40% have carcinoma. Rest have adenoma, abscess, cyst or multinodular goiter

NORMAL SEXUAL DEVELOPMENT
- ❑ wide range of age of onset and development of puberty
- ❑ females
  - age 9 - 13
  - sequence begins with breast bud, mean age at menache = 12.8 years
- ❑ males
  - age 10 - 14
  - sequence begins with testicular enlargement

Table 27. Tanner Staging

	female		male	
stage	breast	pubic hair	genitalia	pubic hair
1	–	–	–	–
2	bud	sparse labial hair	scrotal/testes enlargement	sparse hair at base of penis
3	single contour	hair over pubis	increase in length of penis	hair over pubis
4	nipple forms secondary mound	coarse adult hair	further increase in length and breadth of penis	coarse adult hair
5	adult size and shape	extends to medial thigh	adult size and shape	extends to medial thigh

PRECOCIOUS PUBERTY (see Gynecology Notes)

❑ secondary sexual development before 8 years in girls, 9 years in boys

True (Central) Precocious Puberty

❑ premature activation hypothalamic-pituitary-gonadal axis
❑ hypergonadotropic hypergonadism, hormone levels as in normal puberty
❑ nine times more common in females than males
❑ differential diagnosis
- idiopathic or constitutional (most common, especially females)
- CNS tumours, hamartomas, postmeningitis, increased ICP, radiotherapy
- neurofibromatosis, hypothyroidism

Peripheral Precocious Puberty

❑ hypogonadotropic hypergonadism
❑ differential diagnosis
- congenital adrenal hyperplasia, adrenal neoplasm
- testicular/ovarian tumour
- gonadotropin secreting tumour: hepatoblastoma, intracranial teratoma
- exogenous steroid administration

Evaluation

❑ history: symptoms of puberty, family history of puberty onset, medical illness
❑ physical exam: growth velocity, Tanner staging, neurological exam
❑ hormone levels: estradiol, testosterone, LH, FSH, TSH, GnRH test
❑ bone age
❑ consider CT or MRI of head, ultrasound of adrenals, pelvis

Management

❑ GnRH analogs, medroxyprogesterone
❑ treat underlying cause

Benign Premature Thelarche

❑ isolated breast tissue development in girls age 6 months to 3 years
❑ no other signs of puberty or excessive estrogen effect
❑ may be due to increased sensitivity to estrogen or temporary increase in estrogen levels
❑ normal bone age and adrenal androgens
❑ evaluate every 6-12 months to ensure no further signs of puberty

Isolated Premature Adrenarche

❑ appearance of secondary hair before age 8 in females, age 9 in males
❑ relatively common, caused by premature increase in adrenal androgens
❑ presence of other features of virilization (clitoral enlargement, advanced bone age) or other signs (acne, rapid growth, voice change) requires detailed investigation for pathologic cause
❑ reassurance, no treatment required

DELAYED PUBERTY

❑ see Gynecology section
❑ absence of pubertal development by age 13 in girls and age 14 in boys
❑ more common in males

Central Causes

❑ delay in activation of hypothalamic-pituitary-gonadal axis
❑ hypogonadotropic hypogonadism
❑ differential diagnosis
- constitutional (bone age delayed) – most common (> 90%)
- chronic disease, anorexia nervosa, malnutrition
- pituitary/hypothalamic failure (idiopathic or acquired)
- genetic (e.g. Kallman's symdrome)
- hypothyrodism

Peripheral Causes

❑ hypergonadotropic hypogonadism
❑ differential diagnosis
- genetic (e.g. Turner's, Kleinfelter's)
- gonadal damage – infection, radiation, trauma
- gonadal dysgenesis
- hormonal defect – androgen insensitivity, 5-reductase deficiency

Evaluation
- ❑ history: weight loss, short stature, family history of puberty onset, medical illness
- ❑ physical exam: growth velocity, Tanner staging, neurological exam, complete physical exam
- ❑ hormone levels: estradiol, testosterone, LH, FSH, TSH, GnRH test
- ❑ bone age
- ❑ consider CT or MRI of head, ultrasound of adrenals, pelvis
- ❑ karyotype in girls < 3rd percentile in height (rule out Turner's)

Management
- ❑ identify and treat underlying cause
- ❑ hormonal replacement: cyclic estradiol and progesterone for females, testosterone for males

GENITOURINARY

HEMATURIA

Asymptomatic Microscopic Hematuria
- ❑ 5% of school aged children on single test but < 1% on repeated testing
- ❑ usually found on routine screening
- ❑ 5-10 RBCs per hpf of centrifuged urine; dipsticks are very sensitive but have a high false positive rate
- ❑ benign recurrent hematuria in 2/3 of cases
  - sporadic or familial
  - no associated proteinuria

Gross Hematuria
- ❑ upper urinary tract source
  - cola/tea-coloured urine, casts, proteinuria, dysmorphic RBC's, associated symptoms (i.e. edema, azotemia, HTN)
- ❑ lower urinary tract source
  - bright red urine, initial and terminal stream hematuria, clots, normal RBC morphology, < 2+ proteinuria, no casts
- ❑ very large renal bleeding can look like a lower urinary tract bleed

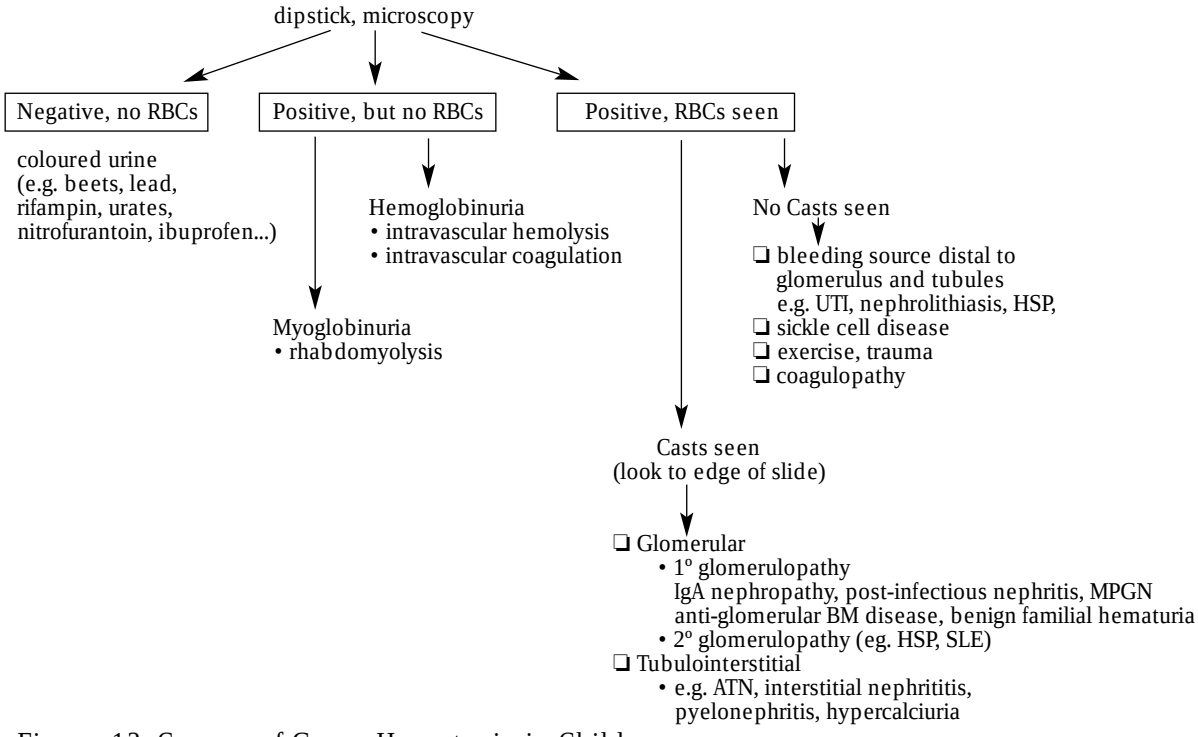


Figure 13. Causes of Gross Hematuria in Children

PROTEINURIA

- ❑ definition: qualitative: 1+ on dilute, 2+ on concentrated urine (specific gravity>1.015); quantitative: 4mg/kg/h on timed urine (>40 mg/kg/hr is nephrotic range)
- ❑ transient: due to fever, dehydration, exercise, seizures, stress
- ❑ persistent
  - orthostatic (more common in adolescents)
  - increased plasma protein concentration
  - glomerular (e.g. nephrotic syndrome, glomerulonephritis)
  - tubulointerstitial (e.g. Fanconi's syndrome, ATN)
  - structural abnormalities of urinary tract (e.g. hydronephrosis)

HEMOLYTIC UREMIC SYNDROME

- ❑ acquired renal insufficiency
- ❑ triad: nephropathy, thrombocytopenia, microangiopathic hemolytic anemia
- ❑ more common from 6 months to 4 years old
- ❑ etiology: E. *coli* toxin O157:H7 verotoxin or Shigella toxin ("hamburger disease") causes endothelial damage
- ❑ prodrome of bloody diarrhea 5-7 days before onset of renal insufficiency
- ❑ history – weakness, lethary, oliguria
- ❑ physical exam – pallor, jaundice (hemolysis), edema, petechiae, hepatosplenomegaly, hypertension
- ❑ investigations – CBC, platelets, reticulocytes, blood smear, Coombs, urinalysis, renal function
- ❑ prognosis: 5-10% mortality, 10-30% kidney damage
- ❑ supportive treatment, dialysis if severe; steroids not helpful

NEPHRITIC SYNDROME

- ❑ acute, subacute or chronic
  - hematuria with RBC casts, proteinuria (< 50 mg/kg/day, not nephrotic-range), hypertension,
  - renal failure (oliguria)
- ❑ post-streptococcal glomerulonephritis
  - most common in children, especially in 4-8 year olds, M > F
  - occurs 1-3 weeks following Group A hemolytic Strep infection (throat/impetigo)
  - diffuse, proliferative glomerulonephritis
  - diagnosed by elevated serum antibody titres against Strep antigens
  - 95% of children recover completely within 1-2 weeks
  - 5-10% have persistent hematuria

Table 28. Major Causes of Acute Glomerulonephritis

	↓ C3	Normal C3
Renal	Post-infectious GMN Membranoproliferative Type 1 (50-80%) Type 2 (> 80%)	IgA Nephropathy Idiopathic rapidly progressive GMN Anti GBM disease
Systemic	SLE SBE Shunt nephritis Cryoglobulinemia	Polyarteritis Wegener's Goodpasture's Henoch-Schonlein

NEPHROTIC SYNDROME

- ❑ severe proteinuria (> 50 mg/kg/day, or > 40 mg/m²/hr) hypoalbuminemia (< 25 g/L), edema, hyperlipidemia
- ❑ histopathology
  - minimal change disease (76%)
  - focal segmental glomerular sclerosis (7%)
  - membranous glomerulonephritis (8%)
  - membranoproliferative glomerulonephritis (5%)
- ❑ minimal change disease
  - peak occurrence between 2-6 years old
  - 90% are steroid-responsive

- ❑ treatment
  - salt and water restriction
  - diuretics may be required
  - prednisone for 8 weeks; if no response, renal biopsy may be required
  - frequent relapses or steroid resistance may require immunosuppressant cytotoxic agents
- ❑ children with nephrotic syndrome are at risk of
  - infections (peritonitis, cellulitis)
  - hypercoagulability (PE, renal vein thrombosis)
  - side effects of drugs (diuretics, steroids, immunosuppressants)
  - hypotension, shock, renal failure

URINARY TRACT OBSTRUCTION

Posterior Urethral Valves

- ❑ 1/50 000 most common obstructive urethral lesion in male infants
- ❑ mucosal folds at the distal prostatic urethra
- ❑ presents with obstructive symptoms, UTI, flank masses, urinary ascites if renal pelvis ruptures
- ❑ now detected antenatally: hydronephrosis, pulmonary hypoplasia
- ❑ diagnosis: U/S, VCUG
- ❑ treatment: destruction of valves

UPJ Obstruction

- ❑ most common ureteric abnormality in children
- ❑ usually in boys, on the left (10-15% bilateral)
- ❑ etiology: segment of ureter lacking peristaltic activity, congenital narrowing, muscular bands, external compression
- ❑ diagnosis: U/S, renal scan +/– furosemide
- ❑ surgical correction with good prognosis

VESICOURETERAL REFLUX (VR)

- ❑ urine flows back from the bladder into the ureter, kidney; common
- ❑ pathophysiology
  - most commonly due to short tunnel of ureter in wall of bladder
  - 30-50% of those with myelomeningoceles, by association with neurogenic bladder
  - secondary to bladder obstruction
- ❑ symptoms of
  - urinary tract infection, pyelonephritis
  - renal failure (FTT, uremia, hypertension) rare
- ❑ diagnosis with U/S, VCUG; tc-DMSA to assess renal scarring
- ❑ Staging by VCUG
  - I - ureters only fill
  - II - ureters and pelvis fill
  - III - ureters and pelvis fill, some dilatation
  - IV - ureters, pelvis and calices fill, significant dilatation
  - V - ureters, pelvis, and calices fill, major dilatation and tortuosity
- ❑ complications: pyelonephritis, recurrent UTI, reflux nephropathy, hypertension, end stage renal disease
- ❑ management: keep urine sterile to prevent renal damage
  - Stage I-III: more than 80% resolve with time
  - observe with repeat VCUG, U/S, urine cultures
  - monitor renal function
  - prophylactic antibiotics (TMP/SMZ, nitrofurantoin)
  - Stage IV and greater —> surgical intervention

GENITAL ABNORMALITIES

Hypospadias

- ❑ 1:500 newborns
- ❑ urethral meatus opens on the ventral side of the penis, proximal to the glans
- ❑ may be associated with chordee (ventral curvature of penile shaft), undescended testicles, inguinal hernia
- ❑ if severe, distinguish from ambiguous genitalia, and rule out other GU abnormalities
- ❑ do not circumcise; foreskin used for surgical repair

Epispadias
- ❑ urethral meatus opens on the dorsum of the penis, at points along the glans and shaft

Phimosis
- ❑ inability to retract prepuce (persistent > 3 years of age)
- ❑ may be congenital or a consequence of inflammation
- ❑ if it is severe, requires circumcision or surgical enlargement of opening

Cryptorchidism
- ❑ arrested descent of testicles in natural path to scrotum (prepubic > ext inguinal ring > inguinal canal > abdominal)
- ❑ common (30%) in premies, 3-4% of full term babies
- ❑ most descend by 3 months; no spontaneous descent at > 1 year old
- ❑ sequelae: trauma (inguinal testes), torsion, malignancy (40x risk), infertility
- ❑ differential: retractile, ectopic, atrophic testes, intersex state
- ❑ undescended testes: may palpate in inguinal canal but unable to milk down into scrotum
- ❑ retractile testes: parents may have seen them in scrotum, can milk them down with warm hands/warm room
- ❑ investigations
  - HCG stimulation test to induce descent, serum testosterone, U/S, CT, surgical exploration, karyotype
- ❑ treatment
  - orchidoplexy by age 2 years, HCG sometimes tried

RESPIROLOGY

UPPER RESPIRATORY TRACT DISEASES

STRIDOR

Common Causes of Stridor
- ❑ lumen: foreign body, hypertrophic tonsils or adenoids
- ❑ respiratory wall: croup, epiglottitis, bacterial tracheitis, post-intubation edema/trauma, tracheomalacia, subglottic stenosis
- ❑ surrounding structures: retropharyngeal or peritonsillar abscess, neoplasm, vascular ring

CROUP AND EPIGLOTTITIS (see Colour Atlas J3 and J4) (see Otolaryngology Notes)

Table 29. Croup vs. Epiglottitis

	Croup	Epiglottitis
prevalence	very common	very rare (decreased since use of Hib vaccine)
common agents	Parainfluenza I, II, III RSV, enterovirus	*H.influenza* type b
age	3 months-3 years	3-7 years
onset	URI prodrome	rapid onset
physical exam	barking cough, stridor, non-toxic	quiet stridor, toxic, respiratory distress, 3D's: drooling, dysphagia, dysphonia
fever	< 39˚C	> 39˚C
WBC	normal	elevated
x-ray	steeple sign (tracheal narrowing)	thumbprint sign (swollen epiglottis)
treatment	humidified air oxygen if hypoxic racemic epinephrine dexamethasone	intubate/ventilate antibiotics: cefuroxime

FOREIGN BODY ASPIRATION

- ❑ acute: sudden onset of choking, stridor, wheezing, cough, respiratory distress
- ❑ chronic: persistent, localized atelectasis in lung; recurrent pneumonia

Diagnosis

- ❑ history: choking spell (recent or remote)
- ❑ chest x-ray: bilateral decubitus films may show air trapping, foreign body, or segmental collapse (see Colour Atlas J6)
- ❑ bronchoscopy: visualize obstruction

Management

- ❑ complete obstruction: Heimlich maneuver or alternating back blows and chest thrusts for infants < 1 year old
- ❑ if unable to expel foreign body: direct laryngoscopy and removal, intubation or emergency tracheotomy

LOWER RESPIRATORY TRACT DISEASES

WHEEZING

Differential Diagnosis of Wheezing

- ❑ asthma: recurrent wheezing episodes
- ❑ pneumonia: fever, cough, malaise
- ❑ bronchiolitis: first episode of wheezing (see Bronchiolitis Section)
- ❑ CF: prolonged wheezing unresponsive to therapy
- ❑ foreign body aspiration: sudden onset wheezing and coughing
- ❑ gastroesophageal reflux with aspiration: feeding difficulties
- ❑ congestive heart failure: associated FTT

BRONCHIOLITIS

- ❑ presents as first episode of wheezing associated with URI and signs of respiratory distress
- ❑ common, affects 15% of children in first 2 years of life
- ❑ peak incidence at 6 months, often in late fall and winter
- ❑ occurs in children prone to airway reactivity, i.e. increased incidence of asthma

Etiology

- ❑ RSV (75%)
- ❑ Parainfluenza, Influenza, Adenovirus

Clinical Features

- ❑ prodrome of URI with cough and fever
- ❑ feeding difficulties, irritability
- ❑ wheezing, respiratory distress, tachypnea, tachycardia
- ❑ children with chronic lung disease, severe CHD and immunodeficiency have a more severe course of the illness

Diagnosis

- ❑ chest x-ray
  - air trapping, peribronchial thickening, atelectasis, increased linear markings
- ❑ nasopharyngeal swab
  - direct detection of viral antigen (immunofluorescence)

Management

- ❑ mild distress
  - supportive: oral or IV hydration, antipyretics for fever
  - humidified oxygen and/or inhaled bronchodilator (Ventolin)
- ❑ moderate to severe distress
  - humidified oxygen
  - inhaled bronchodilator (Ventolin) or racemic epinephrine
  - continue only if effective
  - Atrovent and steroids are not effective
  - rarely intubation and ventilation
  - consider ribavirin in high risk groups: BPD, CHD, congenital lung disease, immunodeficient
  - case fatality rate < 1%

❑ indications for hospitalization
- hypoxia: oxygen saturation < 92%
- persistent resting tachypnea > 60/minute and retractions after several Ventolin masks
- past history of chronic lung disease, hemodynamically significant cardiac disease, neuromuscular problem, immunocompromise
- young infants < 3 months old (unless extremely mild)
- significant feeding problems
- social problem, i.e. inadequate care at home

PNEUMONIA

Clinical Features

❑ incidence is greatest in first year of life
❑ fever, cough, crackles
❑ tachypnea, tachycardia, respiratory distress
❑ bacterial cause has more acute onset, but viral cause is more common
❑ abnormal chest x-ray

Etiology

Table 30. Common Causes of Pneumonia at Different Ages

Age	Bacterial	Viral	Others
neonates	Group B streptococcus E. *Coli*	CMV Herpes virus	*Mycoplasma* *Ureaplasma*
1-3 months	S. *aureus* H. *influenzae* S. *pneumoniae*	CMV, RSV Influenza virus Parainfluenza virus	*Chlamydia trachomatis* *Ureaplasma*
3 months - 5 years	S. *pneumoniae* S. *aureus* H. *influenzae*	RSV Adenovirus Influenza virus	TB
> 5 years	S. *pneumoniae* H. *influenzae*	Influenza virus	*Mycoplasma pneumonia* (most common) *Chamydia pneumonia* TB

Management

❑ supportive treatment: hydration, antipyretics, humidified oxygen
❑ IV or PO antibiotics
- newborn
  - ampicillin and gentamicin +/– erythromycin
- 1-3 months
  - ampicillin +/– erythromycin
- 3 months - 5 years
  - sick: IV ampicillin
  - not sick: PO amoxicillin
- > 5 years
  - erythromycin

ASTHMA

❑ characterized by airway hyperreactivity, bronchospasm and inflammation, reversible small airway obstruction
❑ very common illness which presents most often in early childhood
❑ associated with other atopic diseases such as allergic rhinitis or eczema

Clinical Features

❑ episodic bouts of
- wheezing
- cough: at night, early morning, with activity
- tachypnea
- dyspnea
- tachycardia

Triggers
- ❑ URI (viral or *Mycoplasma*)
- ❑ weather (cold exposure, humidity changes)
- ❑ allergens (pets), irritants (smoke), cold dry air
- ❑ exercise, emotional stress
- ❑ drugs (aspirin, ß-blockers)

Classification
- ❑ mild asthma
  - occasional attacks of wheezing or coughing (< 2 per week)
  - symptoms respond quickly to inhalation therapy
- ❑ moderate asthma
  - more frequent episodes with symptoms persisting and chronic cough
  - decreased exercise tolerance
- ❑ severe asthma
  - daily and nocturnal symptoms
  - frequent ER visits and hospitalizations

Management
- ❑ acute
  - oxygen: to keep oxygen saturation > 92%
  - fluids: if dehydrated
  - ß2-agonists: salbutamol (Ventolin) 0.03cc/kg in 3cc NS q 20 minutes by mask until improvement, then masks q hourly
  - ipatropium bromide (Atrovent) if severe: 1 cc added to Ventolin mask
  - steroids: Prednisone 2mg/kg in ER, then 1 mg/kg po od x 4 days
    - in severe disease, give steroids immediately since onset of action is slow (4 hours)
- ❑ indications for hospitalization
  - initial oxygen saturation < 92%
  - past history of life-threatening asthma (ICU admission)
  - poor response to 5-6 frequent doses of Ventolin
  - concern over environmental issues or family's ability to cope
- ❑ chronic
  - education, emotional support, modification of environmental allergies or irritants (e.g. cigarette smoke)
  - exercise program (e.g. swimming)
  - monitoring if appreciation of symptoms is poor (e.g. peak flow meter)
  - PFTs > 6 years old
  - patients with moderate or severe asthma will need regular prophylaxis in addition to bronchodilators (e.g. inhaled steroids, sodium cromoglycate)

CYSTIC FIBROSIS

- ❑ autosomal recessive
- ❑ 1/3,000 live births, mostly Caucasians
- ❑ mutation in transmembrane conductance regulator of chloride
- ❑ CFTR gene found on chromosome 7 (F508 mutation in 70%)

Clinical Features
- ❑ neonatal
  - meconium ileus
  - prolonged jaundice
  - antenatal bowel perforation
- ❑ infancy
  - pancreatic insufficiency with steatorrhea and FTT (but voracious appetite)
- ❑ childhood
  - anemia, hypoproteinemia, hyponatremia
  - heat prostration
  - recurrent chest infections or wheezing (S. *aureus*, P. *aeruginosa*, *H. influenzae*)
  - hemoptysis
  - nasal polyps (associated with milder disease)
  - distal intestinal obstruction syndrome, rectal prolapse
  - clubbing of fingers

❑ older patients
- COPD
- infertility

Complications
❑ respiratory failure
❑ pneumothorax (poor prognostic sign)
❑ cor pulmonale (late)
❑ pancreatic fibrosis with diabetes mellitus
❑ gallstones
❑ cirrhosis with portal hypertension
❑ infertility
❑ early death (current median survival is 30 years)

Diagnosis
❑ sweat chloride test x 2 (> 60 meq/L)
- false positive tests: malnutrition, Celiac disease, adrenal insufficiency, anorexia nervosa, hypothyroidism, nephrogenic diabetes insipidus, nephrotic syndrome
- false negative tests: peripheral edema, cloxacillin, glycogen storage disease, hypoparathyroidism, atopic dermatitis, Klinefelter syndrome, hypogammaglobulinemia

❑ pancreatic dysfunction - determined by 3-day fecal fat collection
❑ genetics - useful where sweat chloride test is equivocal
❑ prenatal diagnosis for high risk families

Management
❑ nutritional counselling
- high calorie diet
- pancreatic enzyme replacements
- fat soluble vitamin supplements

❑ management of chest disease
- physiotherapy, postural drainage
- exercise
- bronchodilators
- antibiotics (depends on sputum C&S, e.g. cephalosporin, cloxacillin, ciprofloxacin, inhaled tobramycin)
- lung transplantation

❑ genetic counselling

ADOLESCENTS

HEALTH ISSUES

❑ growth and development
- physical growth
- sexual maturation and psychosexual issues
- skin problems

❑ nutritional concerns
- poor nutrition
- eating disorders
- obesity

❑ sexuality issues
- teen pregnancy
- sexual abuse
- STDs and HIV (incidence rising in adolescents)
- contraception
- sexual orientation

❑ substance abuse
- tobacco
- alcohol and drugs

❑ depression and mental health disorders
- suicide, homicide and accidents (70% of teen mortality)
- affective, behavior, adjustment, anxiety disorders
- self-esteem issues
- chronic illness (7-10%)

Notes

Clinical Pearl
- ❑ Injuries are the leading cause of death in adolescents, accounting for 80% of deaths in 15 to 19 year olds. Risk factors include: alcohol use, failure to use safety devices, access to firearms and athletic participation

Remember the HEEADSS Interview - assure confidentiality
- ❑ HOME: where, with whom? relations with family, recent moves, ever run away?
- ❑ EDUCATION: attending school? grades, doing OK?, failures, suspensions, future plans, goals
- ❑ EATING: habits, anorexia, anemia, obesity
- ❑ ACTIVITIES: extracurricular, work, sports, music, car, social clubs, gangs, best friend
- ❑ DRUGS: types used/tried, alcohol, smoking, with friends or alone?
- ❑ SEXUALITY: dating, active, preference, types of experiences, safe sex/contraception, pregnancies, STDs, sexual abuse
- ❑ SUICIDE: self harm thoughts, prior attempts, depression

NORMAL VARIATION IN PUBERTY

- ❑ breast asymmetry may occur as one breast may grow faster than the other; becomes less noticeable as maturation continues
- ❑ physiologic leukorrhea occurs prior to menarche; scant mucoid, clear to milky discharge not associated with pruritis or foul odour; occurs because of stimulation of endometrial glands by estrogen
- ❑ menses may be irregular in duration of period and/or time between periods; on average it takes 18 months to go through the first 12 periods; birth control pills should be avoided as treatment
- ❑ gynecomastia is a common self-limited condition seen in 50-60% of early male adolescents;1-3 cm round, mobile, sometimes tender, firm mass underneath areola; if discharge or fixed mass, should be investigated

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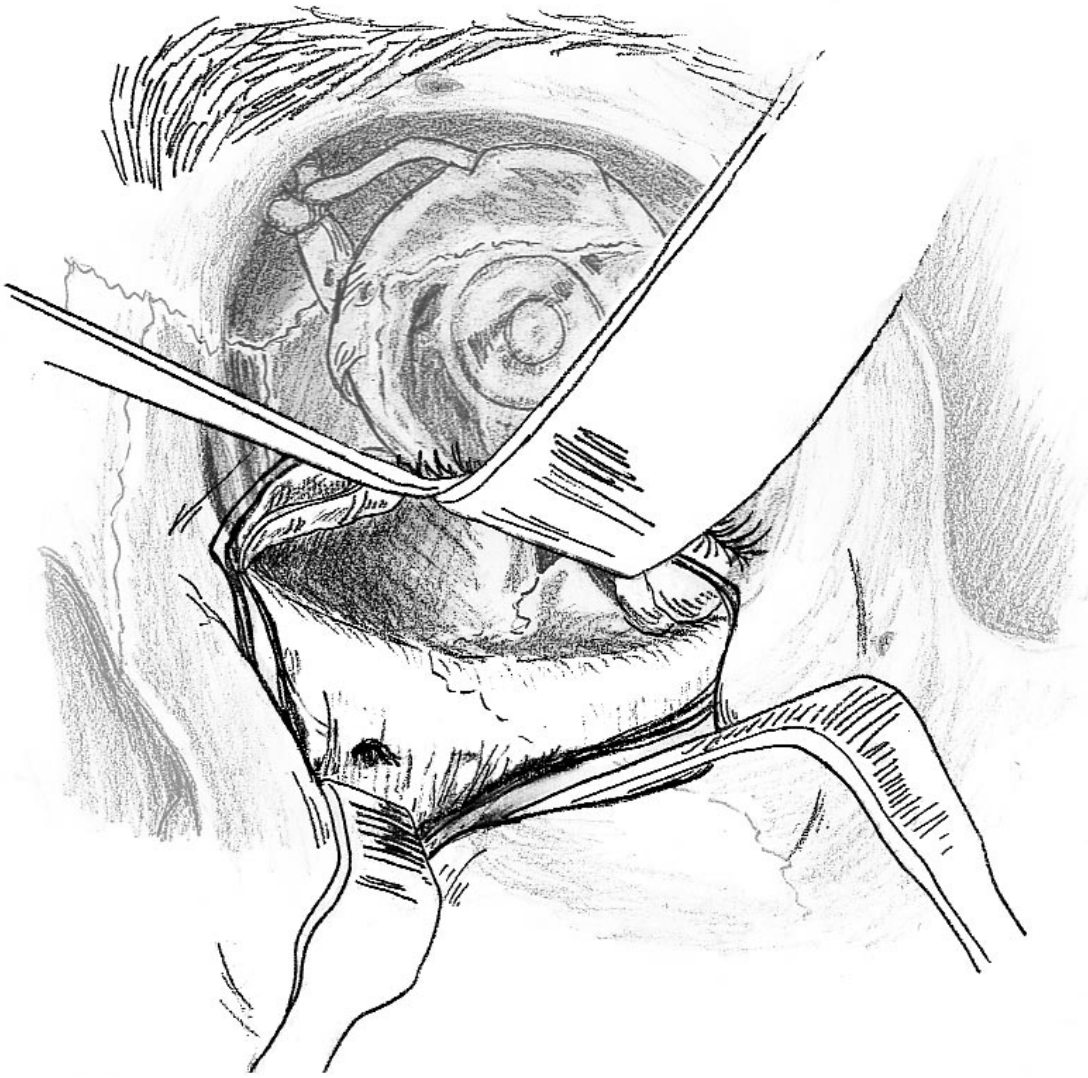

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