

يَقُولُ الرَّسُولُ صَلَّى اللَّهُ عَلَيْهِ وَسَلَّمَ ، قَالَ : " مَنْ نَفَسَ عَنْ مُسْلِمٍ كُرْبَةً مِنْ كُرْبِ الدُّنْيَا نَفَسَ اللَّهُ عَنْهُ كُرْبَةً مِنْ كُرْبِ يَوْمِ الْقِيَامَةِ ،
وَمَنْ يَسَّرَ عَلَى مُسْلِمٍ يَسَّرَ اللَّهُ عَلَيْهِ فِي الدُّنْيَا وَالْآخِرَةِ ، وَمَنْ سَتَرَ مُسْلِمًا سَتَرَهُ اللَّهُ فِي الدُّنْيَا وَالْآخِرَةِ ،
وَاللَّهُ فِي عَوْنِ الْعَبْدِ مَا كَانَ الْعَبْدُ فِي عَوْنِ أَخِيهِ " .. أو كما قال عليه الصلاة والسلام ..

Pediatrics Case Study

Answered



Case Scenarios

All these Cases are
Mentioned in
**Tutorial Lectures
of the Department**

& Problem Solving

By

YAS'ER Project™ Team

- All these Cases are Mentioned in **Tutorial Lectures in the Department**
- These collection don't cover the whole subjects
- This summary is not official and may contain some wrongs ..
so, Please check it before adopt in the study .. & In case of wrongs, please let us know to correct it

がんばってね ! - Semoga Berjaya - ¡Buena suerte

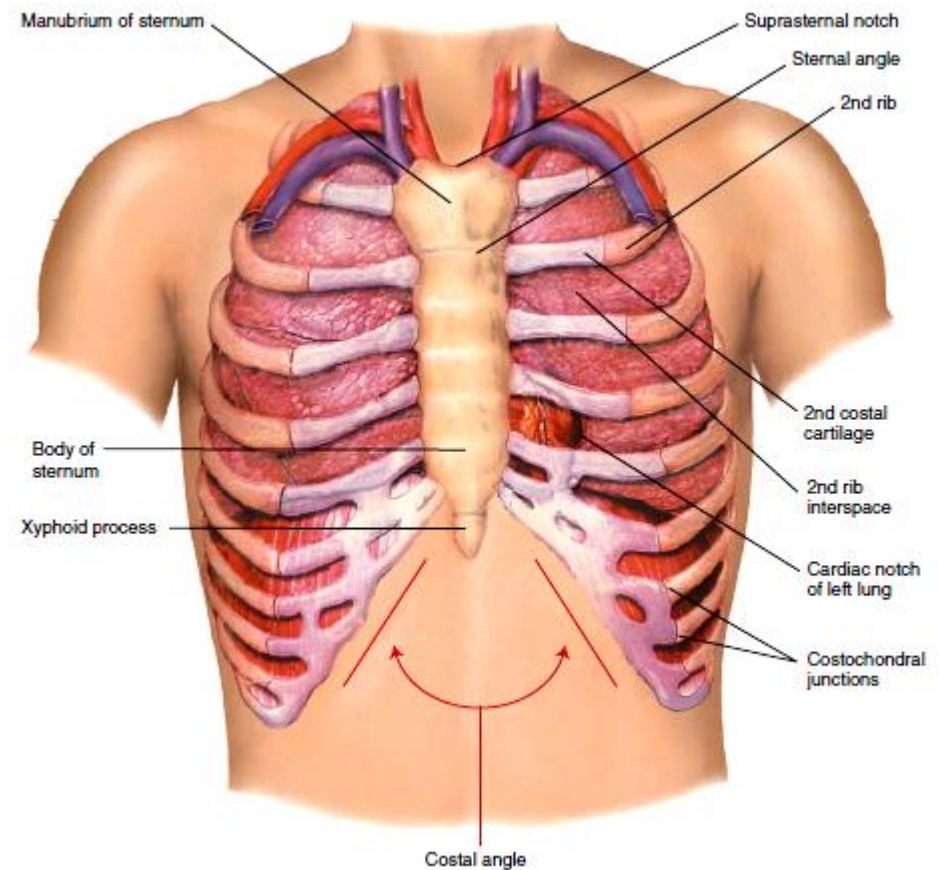
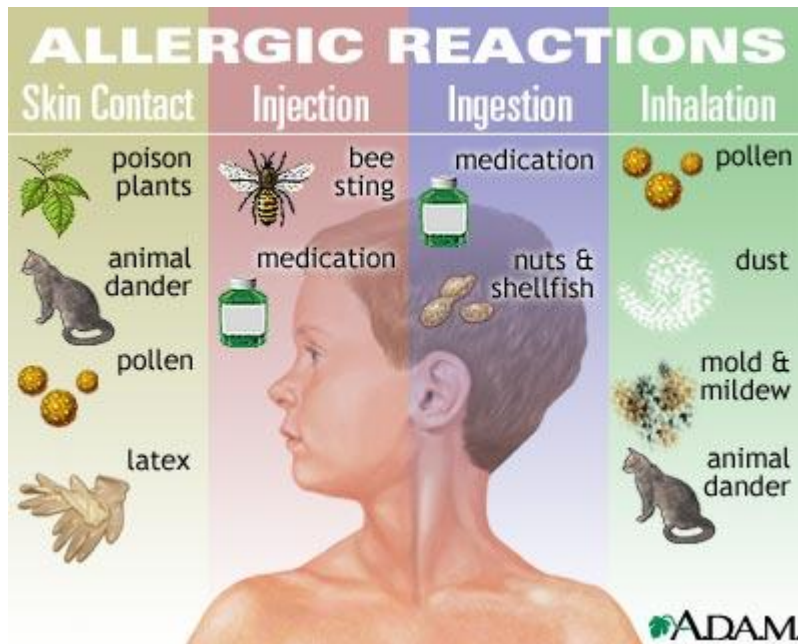
اللهم انفعنا بما علمتنا .. واهدنا إلى أحسن الأخلاق والأعمال ..
لا يهدي إلى أحسنها إلا أنت .. واصرف عنا سيئها لا يصرف سيئها إلا أنت ..



► Index :

Allergy & Pulmonology	Page 2
Cardiology	Page 33
Endocrinology	Page 47
Genetics	Page 53
Hematology	Page 59
Hepatology	Page 65
Malnutrition & Diarrhea	Page 68
Neonatology	Page 73
Nephrology	Page 77
Neurology	Page 83
Rheumatology	Page 91

Allergy & Pulmonology



► **Case (1) :**

A **3-year-old** boy presents with **high fever, stridor, and irritability**.
His RR is **45/min** and SaO₂ is **90%** in room air.

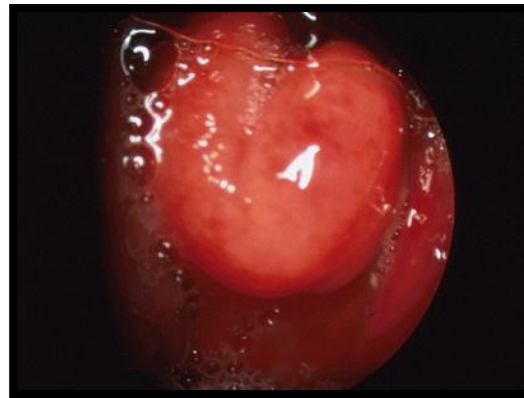
Which ONE of the following steps is most appropriate to do ?

- a- Oral steroids
- b- Examine his chest
- c- Keep him calm while arranging for intubation
- d- Oral paracetamol
- e- Inhaled adrenaline

Answer : c- Keep him calm while arranging for intubation

Comment :

- History and presentations are pointing towards likely diagnosis of **epiglottitis**.
- Once suspected, child should be **kept calm** as upsetting the child may precipitate resp. arrest.
- **No attempts** should be made to examine the child in detail before arranging for intubation by a skilled anesthetist.
- Child should be **nursed in parent's lap** and O₂ should be given via nasal cannula around the face by the parent.



[Acute Epiglottitis]

- Life-threatening emergency
- Caused by H. influenzae type b (Hib)
- Hib immunization led to a decrease in incidence.
- Age: all (most common 1-6 yrs of age).
- Onset is often very acute.
- High fever in an ill, toxic-looking child
- Intensely painful throat
(prevents the child from speaking or swallowing and drooling of saliva)
- Soft inspiratory stridor
- Rapidly increasing respiratory difficulty over hrs.
- Child sits immobile, upright, with an open mouth to optimize the airway.



**Examining the throat with a spatula
or performing a lat. neck X-ray may
lead to
➔ total airway obstruction & death
(Must NOT Be Done)**

➤ **Case (2) :**

• A **1-year-old** girl is brought to the Emergency Department by her upset parents.

She is the first child of the family. She is up to date with vaccinations.

• They tell you she was feeling **hot** for the last **few days**.

They are concerned about her breathing and the '**awful cough**' that she has.

• The observation chart reveals **mild pyrexia** with **good saturations** in air.

As you introduce yourself you can hear a **barking cough**.

Which ONE of the following is the most appropriate diagnosis ?

a- Acute laryngotracheobronchitis (viral croup)

b- Epiglottitis

c- Pneumonia

d- Bronchial asthma

e- Otitis media

Answer : a- Acute laryngotracheobronchitis (viral croup)

Comment :

• Acute laryngotracheobronchitis (croup) is caused by a number of viruses (parainfluenza, influenza, RSV & rhinovirus).

• Age: 6 mo to 6 yrs (peak; 2nd yr of life).

• Some patients can have recurrent attacks.

• Symptoms often start, and are worse, at night.

• Factors suggesting croup rather than Epiglottitis :

- a history of a few days' duration,

- low pyrexia without toxemia,

- barking cough, and absence of drooling.

[Croup Vs. Epiglottitis]

	Croup	Epiglottitis
Onset	Over days	Over hours
Preceding coryza	Yes	No
Fever	< 38.5°C	> 38.5°C
Appearance	Unwell	Toxic, very ill
Cough	Severe, barking	Absent or slight
Able to drink	Yes	No
Drooling of saliva	No	Yes
Stridor	Harsh, rasping	Soft, whispering
Voice and cry	Hoarseness	Muffled, reluctant to speak

► **Case (3) :**

- A **9 year-old** boy presents with a history of **headache** and persistent **green nasal discharge**.
- At **night** he has a **cough** and **snore**s loudly.
- Headache is exacerbated by **leaning forwards**.
- On examination he is afebrile, but has a persistent nasal obstruction and nasal speech.
- He is **tender** over the maxillae and forehead

What is the most likely diagnosis ?

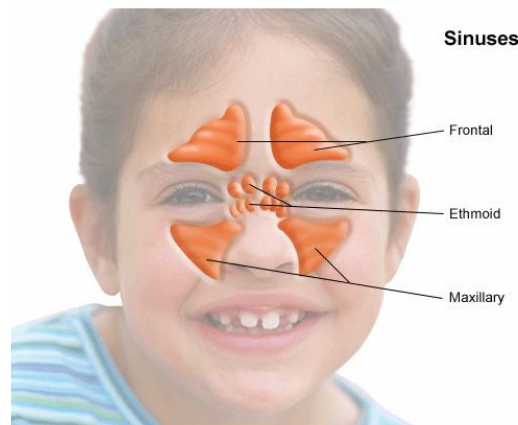
- a- Gastroesophageal reflux
- b- Allergic rhinitis
- c- Sinusitis
- d- Asthma
- e- Croup

Answer : c- Sinusitis

Comment :

Upper airways obstruction and **nasal discharge**, most likely due to sinusitis.

In this case maxillary & frontal sinuses are most likely to be involved.



► **Case (4) :**

- A **9 year-old** boy presents with **high fever** and **throat pain** and vomiting. His immunizations are up to date. There is no FH of note.
- On examination he has a fever to **40.1°C**, with respiratory rate of 14/min and pulse of 90/min.
- He has **bilateral tender** upper cervical lymph nodes. **Tonsils** are covered with grayish membrane, uvula is central.

What is the most likely diagnosis ?

- a- Epiglottitis
- b- Follicular tonsillitis
- c- Infectious mononucleosis
- d- Croup
- e- Bronchiolitis

Answer : b- Follicular tonsillitis

Comment :

- The history of high fever, pain and signs of tonsillitis suggest a bacterial cause, mainly **group A streptococci**.
- Penicillin for **10 days** is required



➤ **Case (5) :**

- A **5 yr-old** boy presents with **ear pain** and **fever**.
 - Every time he gets a cold he develops **ear pain** and receives **antibiotics** from the GP. He has about 6 episodes a year. He is fully immunized.
 - Parents are both **smokers**.
 - On examination :
 - His temp. is **38°C**, RR 15/min & pulse 90/min.
 - He has shotty slightly tender cervical nodes.
- His left eardrum is **red** and **bulging**.
- Throat examination is normal.

What is the most likely diagnosis ?

- a- Group A streptococcal pharyngitis
- b- Mastoiditis
- c- Otitis media with effusion
- d- Acute otitis media (OM)

Answer : d- Acute otitis media (OM)

Comment :

The history suggests recurrent acute OM with URTI.

If episodes are frequently recurring, immunodeficiency may be considered.

[Acute Otitis Media]

- Ear pain and fever.
- Child is irritable and may pull at the affected ear.
- Every child with a fever must have tympanic membranes examined.
- Occasionally, acute perforation of the eardrum with pus visible in the external canal.



➤ **Case (6) :**

- A **7 year-old** girl presents with **right ear pain** and **fever**, **worsening** over the past few days.
- Symptoms began **7 days** ago with severe ear pain and fever, which was treated with paracetamol.
- The following morning, the pain resolved and a **purulent discharge** mixed with blood was noted on her pillow.
- Four days later the pain is **throbbing** and her ear is **tender**.
- She is fully immunized & there is no FH of note.
- On examination she has a fever to **39.7°C**, and her right ear is **displaced** downwards and forwards
- She is **extremely tender** behind the right ear, and has a **purulent discharge** from the ear canal. Her pulse is **110/min** and respiratory rate is 19/min

What is the most likely diagnosis ?

- a- Tonsillitis
- b- Mastoiditis
- c- Otitis media with effusion
- d- Acute otitis media
- e- Otitis externa

Answer : b- Mastoiditis

Comment :

- The history suggests acute otitis media with perforation followed by acute Mastoiditis.

[Mastoiditis]

- Mastoiditis can be confirmed by CT scan of the mastoid.
- Streptococcus pneumoniae and Haemophilus influenzae are the most common pathogens.
- Some require surgical drainage.

➤ **Case (7) :**

- A **1 month old** child presents with **snuffliness** and difficulty in feeding. He has a **crusty** nose and **no** fever.
- His respiratory rate is 32/min.
- Symmetrical conducted secretion sounds are heard in the chest.

What is the most appropriate management ?

- a- Give him penicillin course
- b- Give normal saline nasal drops & reassurance
- c- Admit to hospital
- d- Give oral dexamethasone
- e- Give oral decongestant

Answer : b- Give normal saline nasal drops & reassurance

Comment :

- The history is suggestive of a viral upper respiratory tract infection.
 - Babies are often obligate nasal breathers in the first few months of life.
- Normal saline nose drops may assist feeds by thinning nasal secretions.

**Antibiotics,
steroids
& local decongestants
are NOT indicated.**

➤ **Case (8) :**

• A previously healthy **18-month-old** has been playing in a **separate** room from his family.

The family notices the **sudden** onset of coughing, stridor and cyanosis which resolves over a few minutes.

Subsequently, the patient appears to be normal except for **occasional cough**.

The most likely explanation for this toddler's condition is :

- a- Gastroesophageal reflux
- b- Foreign body in the airway
- c- Croup
- d- Epiglottitis
- e- Viral pneumonia

Answer : b- Foreign body in the airway

Comment :

- The sudden onset of stridor, cough & cyanosis in this toddler points to aspiration of foreign body in the airway that has descended to the lower airway tree explaining the brief resolution of symptoms.
- This can cause later complications (lung collapse, emphysema & pneumonia).
- A plain chest x-ray and bronchoscopy are warranted.



➤ **Case (9) :**

• A **previously well 3-yr-old** boy is brought by his parents with a **painful throat** of 2 days duration with refusal to eat. He has fever of 37.8°C

• Examination revealed **red throat** and runny nose.

What is the most likely diagnosis ?

- a- Streptococcal tonsillitis
- b- Viral nasopharyngitis
- c- Diphtheria
- d- Leukemia



Answer : b- Viral nasopharyngitis

In this child, what is the most appropriate treatment ?

- a- Amoxicillin
- b- Nose drops and paracetamol with ample fluids
- c- Oral decongestants
- d- Intramuscular cefotaxime

Answer : b- Nose drops and paracetamol with ample fluids

Comment :

- Viral infections of the nose and pharynx are very common in pediatric age group.
- Self-limited diseases that require **only** some rest, good hydration and symptomatic treatment.

Overuse of antibiotics is discouraged

**unless there are
signs suggesting a bacterial cause
like high fever, pus formation
or generalized ill health.**

► **Case (10) :**

- A **12-year-old** girl has had a “cold” for 14 days.
- In the 2 days prior to the visit to your office, she has developed a fever of **39°C**, **purulent** nasal discharge, **facial pain**, and a daytime cough.
- Examination of the nose after topical decongestants shows **pus** in the middle meatus.

The most likely explanation for this condition is :

- a- Brain abscess
- b- Maxillary sinusitis
- c- Streptococcal throat infection
- d- Tonsillitis
- e- Middle-ear infection

Answer : b- Maxillary sinusitis

Comment :

- A “cold” lasting >10 -14 days with **fever**, **headache** and **facial pain** is indicative of **sinusitis**.
- Examination of the nose can reveal pus draining from the middle meatus in maxillary, frontal, or anterior ethmoid sinusitis.
- Brain abscess is unlikely “**no neurologic symptoms**”
- Streptococcal throat infection is unlikely “**acute onset of sore throat without other signs and symptoms of upper respiratory infection**”
- Middle-ear infection is unlikely “**lack of ear pain**”

► **Case (11) :**

- A **2-year old** female presents to the ER at **3 AM** with **barking cough**, runny nose, **mild** chest recessions and fever of **37.7°C**.

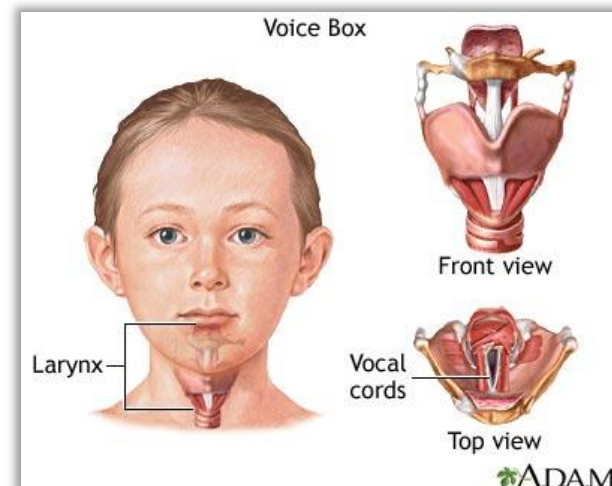
What is the most appropriate management ?

- a- Take her to resuscitation & ask for anesthetic to come immediately.
- b- Give an oral dose of dexamethasone.
- c- Request a lateral neck x ray.
- d- Steam inhalation, oral dexamethasone and observation for 4 hrs.
- e- Intravenous antibiotics.

Answer : d- Steam inhalation, oral dexamethasone and observation for 4 hrs

Comment :

- The child has infectious croup & mild resp. distress.
- The best management would be to give her steam inhalation and a dose of steroids and observe for few hours; most cases respond and can be safely managed later at home.
- No need for intubation for this child & antibiotics are not indicated.
- Investigations are usually not necessary.



➤ **Case (12) :**

• A **16-year-old** boy has been complaining of **shortness of breath** for the **last 2 days**.

• On examination **bronchial breathing** is heard over the **right lower lobe**.

What is the most likely diagnosis for this clinical finding ?

- a- Pneumothorax
- b- Pneumonia
- c- Asthma
- d- Emphysema

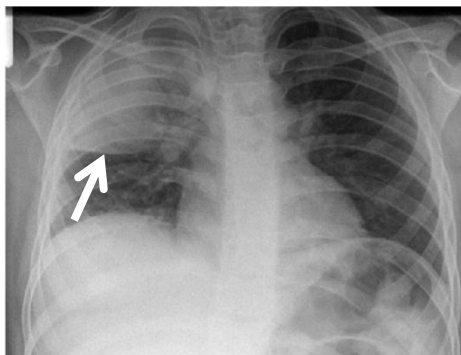
Answer : b- Pneumonia

Comment :

- Bronchial breathing is heard over an airless lung (consolidation, atelectasis or dense fibrosis).
- Bronchial breath sounds are classically heard throughout both inspiration and expiration.
- Very quiet breath sounds are heard :
 - over hyperinflated lungs (e.g. emphysema),
 - or when breath sounds are prevented from reaching the chest wall by a layer of air, fluid or fibrosis.
- There is some resemblance to the sounds heard over the normal trachea, but bronchial breathing is higher in pitch and more blowing in quality.
- It does not have to be loud.

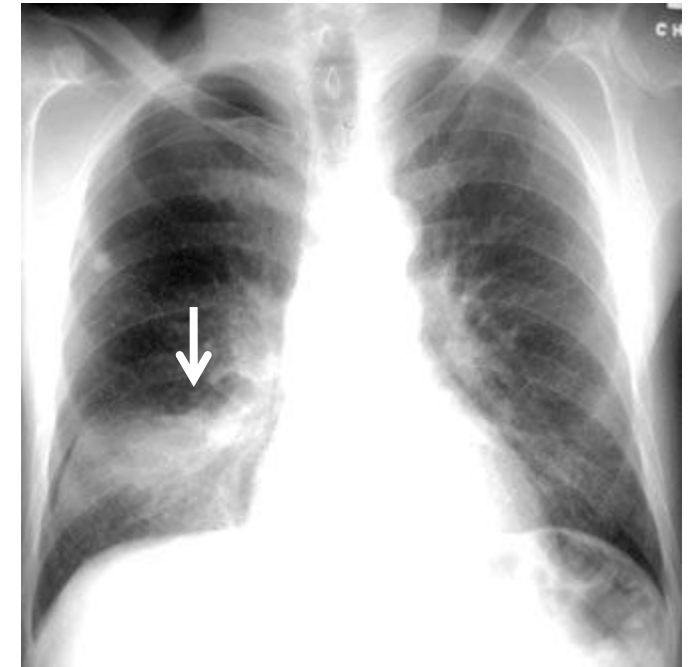
Consolidation of the right upper lobe.

Lobar consolidation is a feature of pneumococcal pneumonia



[Pneumonia]

- The classic presentation of pneumonia is of a **cough** and **fever** with the variable presence of **sputum** production, **dyspnea** and **pleurisy**.
- Most patients have **constitutional symptoms** such as malaise, fatigue and asthenia, and many also have gastrointestinal symptoms.
- Although patients with pneumonia usually possess these characteristic clinical features, there can be **major differences** in presentation based on **the host** and the **etiological agent**.



Right Lower Lobe Pneumonia

Alveolar space filled with inflammatory exudate, WBC, bacteria, plasma and debris

► **Case (13) :**

- A **10-month-old** infant has **poor** weight gain, **persistent** cough, and a history of **several** bouts of pneumonitis.
- The mother describes the child as having very large, foul-smelling **stools** for months.

Which of the following diagnostic maneuvers is likely to result in the correct diagnosis of this child ?

- a- CT of the chest
- b- Serum immunoglobulins
- c- TB skin test
- d- Inspiratory and expiratory chest x-ray
- e- Sweat chloride test

Answer : e- Sweat chloride test

Comment :

Clinical features of cystic fibrosis

Newborn

- Diagnosed through newborn screening.

Infancy

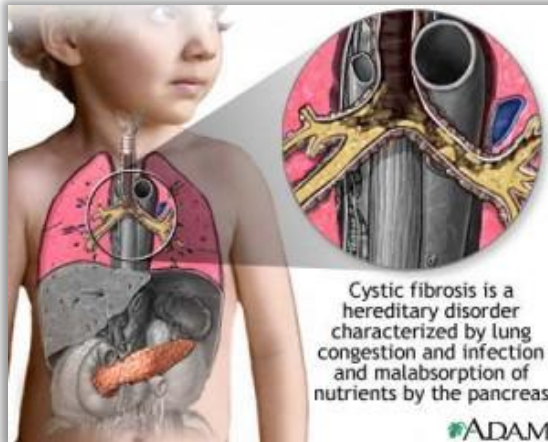
- Meconium ileus in newborn period
- Prolonged neonatal jaundice
- Failure to thrive
- Recurrent chest infections
- Malabsorption, steatorrhea

Young child

- Bronchiectasis
- Rectal prolapse
- Nasal polyp
- Sinusitis

Older child and adolescent

- Allergic bronchopulmonary aspergillosis (ABPA)
- Diabetes mellitus (often not insulin-dependent)
- Cirrhosis and portal hypertension
- Distal intestinal obstruction (DIOS, meconium ileus equivalent)
- Pneumothorax or recurrent haemoptysis
- Sterility in males
- Increasing psychological problems



[Cystic Fibrosis]

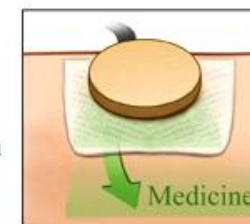
- Autosomal recessive.
- Defective gene is located on chromosome 7 codes for the protein called cystic fibrosis transmembrane regulator (CFTR) [cyclic AMP-dependent chloride channel blocker].
- Abnormal ion transport across the epithelial cells of the exocrine glands of the respiratory tract & pancreas → ↑↑ viscosity of secretions.
- Abnormal function of sweat glands → ↑↑↑ sodium & chloride concentrations in the sweat.

► **for Diagnosis of Cystic Fibrosis :**

- **Pilocarpine iontophoresis :**
 - 10-30 mmol/L in normal children
 - 30 - 59 mmol/L equivocal
 - **60-125 mmol/L in cystic fibrosis**



A mild electrical current pushes medicine into skin to cause sweating



Sweat is collected, and salt content is measured

© Healthwise, Incorporated

► **Case (14) :**

- A **previously** healthy **4 year old** boy presented with **difficult** breathing for 1 day.
- Three days prior, he had developed a runny nose, cough, and fever (**40°C**).
- On examination, he is awake, alert, in moderate distress, with **decreased** air entry over **right lower lobe** with **crackles**.
- His blood count shows **WBC 20,000/mm³**, 70% segs, **11% bands**.

What is the appropriate NEXT step in management ?

- a- To do a blood culture
- b- To do a chest x-ray
- c- To give an intramuscular antibiotic
- d- To give intravenous fluids
- e- To do another complete blood count

Answer : **b- To do a chest x-ray**



**Consolidation of the right lower lobe
with pleural effusion
(obliterated right costophrenic angle)**

► **Case (15) :**

- A **2-year-old** child with **poor growth** has seen the GP with **recurrent** chest infections.
- He is on inhaled sodium **cromoglycate** through a spacer with equivalent benefit.
- He was admitted at 9 months of age with bronchiolitis.
- In a recent referral to the ENT department they detected **nasal polyps** and prescribed intranasal steroids.

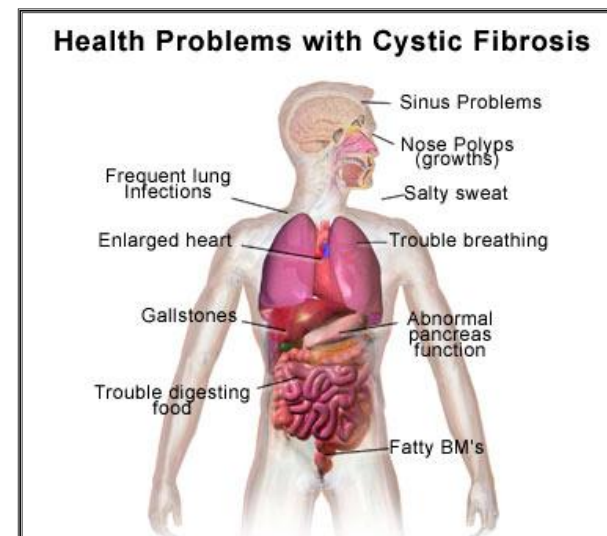
What is the best management for him?

- a- Chest physiotherapy
- b- Sweat chloride test
- c- 7 day course of oral amoxicillin
- d- Refer for a routine flexible bronchoscopy
- e- Prescribe 2 weeks of oral ciprofloxacin

Answer : **b- Sweat chloride test**

Comment :

- The child is displaying features suggestive of CF,
- A sweat test is the most appropriate thing to do.



➤ **Case (16) :**

• A **14-year-old** patient presents with **anorexia**, fever and hot flushes. The chest X-ray shows a 4-cm, large, left upper lobe cavity. **Active tuberculosis** is suspected.

What is the NEXT appropriate step to confirm the diagnosis ?

- a- Computed tomographic (CT) scanning
- b- Tuberculin test (Mantoux test)
- c- Blood cultures
- d- Sputum sample
- e- Serum inflammatory markers

Answer : d- Sputum sample

Comment :

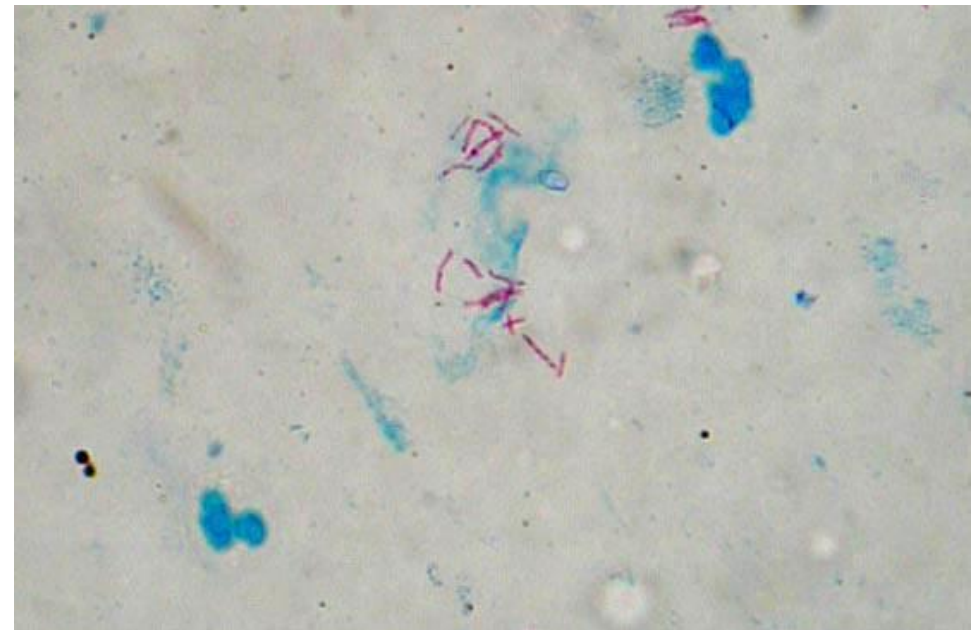
- The laboratory diagnosis of pulmonary TB relies on examination and culture of sputum or other respiratory tract specimens.
- The **definitive diagnosis** requires the **growth of Mycobacterium TB from respiratory secretions**.
- A diagnosis can be based on typical **clinical** and **chest X-ray** findings with either:
 - sputum positive for acid-fast bacilli or other specimens,
 - or typical histopathological findings on biopsy material.

Mycobacterium Tuberculosis

➤ Sputum examination :

- Direct smear and culture
- Direct smear examination is only positive when large numbers of bacilli begin to be excreted.

Acid-fast stain



Tubercle bacilli in sputum smear by Acid fast staining

► **Case (17) :**

- A **4-month-old** male infant presented during **winter** season with increased work of breathing, **not** related to feeding, having had two apneic episodes.
- He had **crackles** on auscultation of the chest.
- The chest X-ray showed **hyperinflation** with scattered areas of atelectasis.

What is the most likely diagnosis for this clinical finding ?

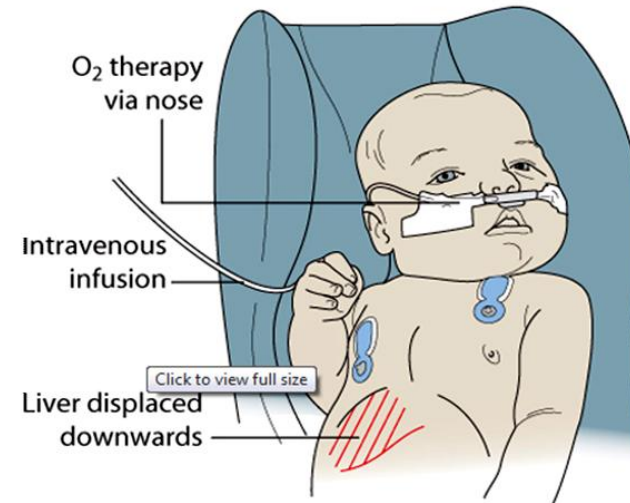
- a- Cystic fibrosis
- b- Pneumonia
- c- Bronchiolitis
- d- Immotile cilia syndrome
- e- Tuberculosis

Answer : c- Bronchiolitis

Comment :

- Bronchiolitis is most common in the first 6 months of life, during the winter months.
- It is usually secondary to respiratory syncytial virus (RSV) but many other microbes have been implicated in pathogenesis.
- Chest X-ray shows hyperinflation with scattered areas of atelectasis in about a third of patients.
- RSV can be demonstrated in nasopharyngeal secretions.
- Complications include feeding difficulty, apnea, respiratory failure and superimposed bacterial infections.
- Apnea is more common in early infancy or ex-pre-term infants or infants with neuromuscular weakness.
- Treatment is supportive.
- Up to half of the patients will have recurrent wheeze following bronchiolitis.

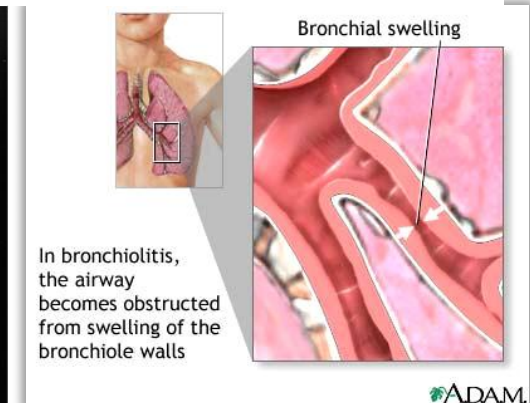
Bronchiolitis



- Apnoea in infants
<4 months
- Sharp, dry cough
- Cyanosis or pallor
- Hyperinflation of the chest:
- sternum prominent
 - liver displaced downwards
- Subcostal and intercostal recession
- Auscultation:
- fine end-inspiratory crackles
 - prolonged expiration



Chest X-ray shows **hyperinflation** of the lungs with **flattening** of the diaphragm, **horizontal ribs** and **increased hilar bronchial markings**



► **Case (18) :**

- A **15 year old** boy from **Sudan** presents with a **4 week** history of cough and weight loss.
- Initially the cough was dry, but recently he has coughed up some **blood**. He has **lost 8 kg in weight**. He has **previously been healthy**.
- Full term normal delivery, no neonatal problems.
- Immunizations up to date, including **BCG** at birth.
- No family or social history of note.
- On examination the temperature is **38.5°C**, and he has loose skin folds suggesting **recent** weight loss. He has **crackles** at the right base.
- A Mantoux (tuberclin) test gives **12 mm induration** after 48 hours.

What is the most likely diagnosis ?

- a- Active tuberculosis
- b- Latent tuberculosis
- c- Miliary tuberculosis
- d- Positive Mantoux due to BCG
- e- Tuberculous meningitis

Answer : a- Active tuberculosis

Comment :

- Chronic symptoms of fever, cough and weight loss, at risk background, and positive Mantoux test >10 mm make active TB likely.
- This can be confirmed by 3 x sputum for Acid Fast Bacilli (AFB).
- Treatment requires at least 2 mo of triple therapy and at least a further 4 mo of double therapy.
- It is preferable to monitor this directly (Directly Observed Therapy; DOT) to reduce the risk of relapse & development of resistant organisms.

► **Tuberculin Skin Test (TST) :**

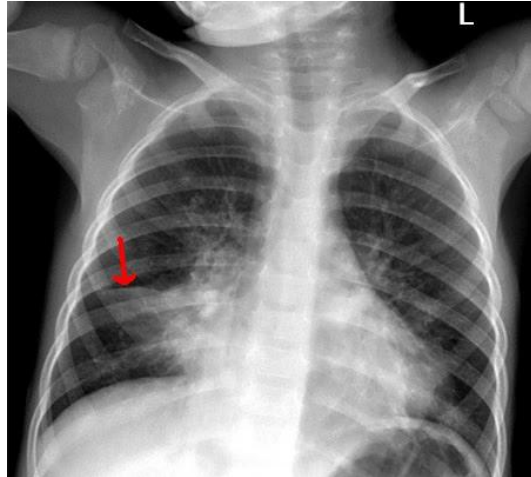


- A reaction of <5 mm is considered **negative**
- 5-9 mm is considered **positive (+)**
- 10-19 mm is considered **positive (++)**
- >20 mm is considered **positive (+++)**

N.B. A positive tuberculin skin test indicates tuberculous infection, with or without disease

► **Case (19) :**

- A **14 year old** boy from **Aswan** presents with a history of **cough** and **fever** of **2 month duration**.
- Recently he has begun to produce **rusty brown sputum** and has been hot and **sweaty at night in bed**.
- On examination he is on the 50th centile for height and the 3rd centile for weight. He has a RR of 20/min. with mild intercostal recession.
- He is **dull** to percussion at the right lung base with poor air entry there.
- He has a patch of bronchial breathing at the right mid-zone posteriorly.



What is the most likely diagnosis ?

- a- Allergic rhinitis
- b- Asthma
- c- Mycoplasma pneumonia
- d- Pneumococcal pneumonia
- e- Tuberculosis

Answer : e- Tuberculosis

Comment :

- The history is one suggestive of a **chronic** chest infection with **pleural effusion** and **weight loss**.

The **night sweats** and **ethnicity** make tuberculosis the prime suspect.

- Treatment requires prolonged therapy, and this is best given as DOT (Directly Observed Therapy) 3 times a week.

- Triple or Quadruple therapy (rifampicin, isoniazid, pyrazinamide, ethambutol) is the recommended initial combination.
- This is decreased to the two drugs rifampicin and isoniazid after 2 mo, and by this time antibiotic sensitivities are often known.

N.B. After puberty, pyridoxine should be given weekly to prevent the peripheral neuropathy associated with isoniazid therapy, a complication which does not occur in young children.

- Asymptomatic children who are Tuberclin –positive & therefore latently infected should also be treated (e.g. with rifampicin and isoniazid for 3 mo).

→ This will decrease the risk of reactivation of infection later in life.

➤ **Case (20) :**

- A **10-year-old** boy complains of persistent **clear** nasal discharge during the **spring** and **summer**.
- He constantly **rubs** his nose & eyes; sometimes he has an associated cough.
- The symptoms usually get better in the autumn.
- He was diagnosed with **asthma** at 2 yrs, but this has improved with age. He rarely requires beta agonist inhaler (bronchodilator inhaler).
- On examination he has a **clear** nasal discharge, **nasal speech** & **pink non-purulent conjunctivae**.

What is the most likely diagnosis ?

- a- Gastroesophageal reflux
- b- Allergic rhinitis
- c- Sinusitis
- d- Asthma
- e- Croup

Answer : **b- Allergic rhinitis**

[Allergic rhinitis]

- This atopic boy has classical symptoms of seasonal rhinitis.
- Skin prick testing can be helpful in identifying the causative antigen.
- May be associated with allergic conjunctivitis.
- **Topical intranasal corticosteroids should be used in children with allergic rhinitis that is resistant to antihistamine therapy.**
- Treatment of allergic rhinitis :
 - Antihistamines, Steroid nasal sprays, Cromoglycate drops, Leukotriene inhibitors & Specific immunotherapy

➤ **Case (21) :**

- A **12-year-old** boy presents in **Spring** with sneezing, clear rhinorrhea, and nasal itching associated with conjunctivitis. Physical examination reveals boggy, pale nasal edema with a clear discharge.

What is the most likely diagnosis ?

- a- Foreign body
- b- Vasomotor rhinitis
- c- Neutrophilic rhinitis
- d- Nasal mastocytosis
- e- Allergic rhinitis

Answer : **b- Allergic rhinitis**

Comment :

- Allergic rhinitis affects up to 20% of children.
- In addition to coryza and conjunctivitis, it can also present as 'cough-variant rhinitis' due to post-nasal drip.
- It may be associated with eczema, asthma, sinusitis and adenoidal hypertrophy.
- **Seasonal** allergic rhinitis is predominantly due to sensitization and exposure to airborne **pollens** and **spores**.
- **Perennial** allergic rhinitis (all the year) is usually due to sensitization and exposure to **house dust mite** or ongoing exposure to an allergen such as a **pet** to which the child is sensitized.

Which of the following is the recommended management?

- a- Institute strict measures to avoid outdoor allergen exposure.
- b- Begin seasonal use of topical sympatho-mimetic nasal drops.
- c- Begin seasonal use of topical intranasal corticosteroids.
- d- Give a 10-day course of amoxicillin.

Answer : **c- Begin seasonal use of topical intranasal corticosteroids**

➤ **Case (22) :**

- A **12-year-old** presents with **acute-onset urticaria** that has gradually worsened over the past 10 days.
- Detailed history reveals **no clues** to the possible etiology.
- Findings on physical exam. are normal except for **urticaria**.

Which of the following diagnostic options is recommended ?

- a- Systematic elimination diets to determine a possible ingestant cause
- b- Allergy skin testing
- c- Serum total IgE and specific IgE(RAST)
- d- Skin biopsy
- e- None of the above

Answer : b- Allergy skin testing

Comment :

- No laboratory test confirms or excludes the diagnosis of urticaria.
- Allergy skin testing can be helpful in sorting out causes of acute urticaria, especially when supported by historical evidence.
- Drugs and foods are the most common causes of acute urticaria.

Which of the following treatment options is recommended for this patient ?

- a- A bland diet
- b- Wearing cotton clothes
- c- Oral antihistamine
- d- Oral prednisone
- e- Topical corticosteroids

Answer : c- Oral antihistamine

Comment :

- Antihistamines are usually effective for treatment of urticaria.
- Diphenhydramine and hydroxyzine are effective but cause sedation.
- A non-sedating antihistamine (e.g. Loratadine) is often the preferred therapy for urticaria for school-aged children to minimize the effect on learning and school performance.

*** Causes of Urticaria :**

- **Infection**
- **IgE-mediated**
 - Specific food - cow's milk, nuts (especially peanuts), fish
 - Blood products
 - Drugs: penicillins, cephalosporins
- **Pharmacological**
 - Foods containing histamine-releasing substances (e.g. strawberries, egg white, cheese)
 - Aspirin and other nonsteroidal antiinflammatory agents
- **Physical agents**
 - Heat, cold, pressure

➤ **Case (23) :**

- A **9-year-old** girl with **repeated** episodes of streptococcal pharyngitis experiences another episode of sore throat.
- Oral **amoxicillin** is started, with the first dose given in the office. One hour later, she experiences a "**funny feeling**" and a **tingling** sensation around her mouth.
- Next she becomes **apprehensive**, has **difficulty swallowing**, and develops a **hoarse voice**.
- On arrival at the emergency department, she has giant **urticaria** and the following vital signs:
pulse **130**, RR 32/min, BP **70/30** mm Hg, and temperature 37.2°C.

What is the most likely diagnosis ?

- a- Streptococcal toxic shock
- b- Scarlet fever
- c- Stevens-Johnson syndrome
- d- Reye syndrome
- e- Anaphylaxis

Answer : e- Anaphylaxis

Comment :

- Anaphylaxis to penicillin usually occurs within 30-90 minutes of administration of this drug.
- Anaphylactic shock is often missed as a diagnosis unless a complete history is obtained and there is a high index of suspicion.

➤ **Drug Allergy :**

- Drug allergies do occur in children, especially to antibiotics, but **only a minority** who are labeled drug allergic are truly allergic.
- This is usually because viral illnesses, for which children are often prescribed antibiotics, themselves cause skin rashes.
- A **detailed history** is required of the nature & timing of the rash in relation to taking the antibiotics.

➤ **Case (24) :**

- The mother of an **8-year-old** boy with acute streptococcal tonsillitis calls to report that now, within **15 min** after the first dose of oral **penicillin V**, he is complaining of **itching**.

Which of the following should you recommend ?

- a- A dose of oral Benadryl, with instructions to call again if he has not improved within 30 min.
- b- Immediate return to the doctor or the nearest emergency department.
- c- Careful monitoring at home, with instructions to return to your office or the nearest emergency department if he develops shortness of breath or loss consciousness.
- d- Schedule a visit for a laboratory test to determine serum tryptase level.
- e- Substitution of erythromycin for penicillin.

Answer : b- Immediate return to the doctor or the nearest emergency department.

Comment :

- The urticaria reaction described may develop into anaphylaxis; the latter requires emergency treatment. In addition, the penicillin V should be **stopped** and a substitute non penicillin antibiotic chosen.

➤ **Case (25) :**

- A **6-month-old infant** develops protracted projectile **vomiting**, and **lethargy** about 2 hr after ingesting a **milk formula**.

The most likely diagnosis is :

- a- Generalized anaphylaxis
- b- Milk-induced enterocolitis syndrome
- c- Gastrointestinal anaphylaxis
- d- Allergic eosinophilic gastroenteritis

Answer : b- Milk-induced enterocolitis syndrome

Comment :

- Food protein-induced enterocolitis syndrome typically manifests in the first several months of life with irritability, protracted vomiting and diarrhea, not infrequently resulting in dehydration.
- Vomiting generally occurs 1-3 hour following feeding, and continued exposure may result in bloody diarrhea, anemia, abdominal distention, and failure to thrive.
- Symptoms are most commonly provoked by cow's milk- or soy protein-based formulas but occasionally result from food proteins passed in maternal breast milk.

➤ **Case (26) :**

- A **12-year-old** child with a history of allergy to insect bites is stung and immediately begins experiencing tightness in the chest and wheezing.

The drug of choice for management of this child is :

- a- Inhaled albuterol (bronchodilator)
- b- Subcutaneous epinephrine
- c- Intramuscular diphenhydramine
- d- Oral corticosteroids

Answer : b- Subcutaneous epinephrine

Comment :

- The principal treatment of choice of anaphylaxis is aqueous epinephrine, **1:1,000, 0.01 ml/kg** (maximum 0.3 ml for a child or 0.5 ml for an adult) by **IM injection**, which can achieve more rapid effective concentrations than obtainable with SC injection.

➤ **Treatment of Anaphylaxis :**

Additional measures may include :

- Systemic corticosteroids
- Supplemental O₂; airway maintenance
- IV fluids, vasopressor therapy
- Repeat epinephrine if Sx persist or increase after 10-15 minutes
- Repeat antihistamine ± H₂ blocker if Sx persist
- Observe for a minimum 4 hours
- Arrange follow-up care, provide EpiPen® Rx (self injectable adrenaline) and education

► **Case (27) :**

- A 5-year-old child presented with edema of the lips after ingesting eggs.

Which of the following is TRUE ?

- a- Hyposensitization therapy should be performed.
- b- Food allergy can be reliably diagnosed by skin testing.
- c- A history of apparent allergic reaction to eggs is usually adequate for a diagnosis.
- d- Food allergy may be IgE mediated or non-IgE mediated.
- e- The diagnostic test for food allergy is direct oral challenge in the hospital setting.

Answer : d- Food allergy may be IgE mediated or non-IgE mediated

Comment :

- Hyposensitization therapy can be dangerous and is not routinely recommended.
- IgE titers and skin tests are limited by false positive and false negative results.
- The history alone is not adequate for the diagnosis and the definitive diagnostic test for food allergy in the hospital setting is direct oral challenge.
- Food allergies may be IgE mediated or non-IgE mediated.

[Food Allergy]

- A food allergy occurs when a pathological immune response is mounted against a specific food protein.
- Food allergy may be IgE mediated or non-IgE mediated. If a non-immunological reaction to a specific food occurs this is called non-allergic food hypersensitivity.
- Presentation varies with the agent and the child's age: in infants the most common causes are milk, egg and peanut. In older children peanut, tree nut and fish.

► **Case (28) :**

- Because of a strong family history on both sides, the parents of a newborn baby ask for guidance about preventing their child from developing an allergy to peanuts.

Which of the following approaches is recommended ?

- a- Begin and extend breast-feeding until age 2 year, with exclusion of peanuts from the mother's diet.
- b- Begin and extend breast-feeding until age 2 year, with the mother ingesting gradually increasing amounts of peanut 18-24 mo of age.
- c- ~~Begin and continue breast-feeding as routinely recommended, with the mother regularly ingesting small amounts of peanuts but not introducing peanuts in the child's diet until age 1 year.~~
- d- Begin and continue breast-feeding as routinely recommended, excluding peanuts from the mother's diet while breast-feeding and from the child's diet until age 3 year.

Answer : d- Begin and continue breast-feeding as routinely recommended, excluding peanuts from the mother's diet while breast-feeding and from the child's diet until age 3 year.

Comment :

- There is no consensus on whether food allergies can be prevented. However, several authorities recommend delaying introduction of major food allergens to infants from atopic families.
- Recommendations include promotion of breast-feeding with maternal exclusion of peanut and nut products from the mother's diet and delay in introducing major allergenic foods: cow's milk until 1 yr of age; egg until 18-24 mo of age, and peanuts, tree nuts, and seafood until 3 yr of age

[Acute Asthmatic Attack] 1/2

* History :

- Adam a **13-year-old** boy is a known **asthmatic** who was referred to the emergency unit by his GP. This is his third attendance with an **acute wheeze** in 3 months.
- His mother reports that last time he was nearly transferred to the **pediatric ICU**. He has developed a **cold** and become **acutely breathless** and is using his salbutamol inhaler hourly **without** much relief.
- The accompanying letter says that he is prescribed :
 - Beclomethasone MDI 100 µg/ metered inhalation 2 puffs b.d. (inhaled steroid)
 - Salmeterol MDI 50 µg/metered inhalation 1puff b.d. (inhaled long acting bronchodilator)
 - and Salbutamol MDI 100 µg/metered inhalation p.r.n (short acting bronchodilator).

- This boy has **repeated** acute exacerbations of asthma.
- **Poor** adherence to home treatment is the most likely underlying cause.
- This is common in all age groups but particularly in **teenagers** with their growing independence and risk-taking behavior.

* Examination :

- Adam is sitting up in bed with a nebulizer in progress containing 5 mg salbutamol.
- His oxygen saturation on 5 L/min of O₂ on arrival is documented as **89%**.
- He is **quiet** but able to answer questions with **short sentences**.
- Chest is **hyperinflated** and is using his **accessory muscles** of respiration
- RR is **60 breaths/min** and he has marked tracheal **tug** with intercostal and subcostal **recession**.
- On auscultation there is **equal** but **poor** air entry with widespread **expiratory** wheeze.
- Temp. is **37.6°C**, and pulse is **180 beats/min** with **good** perfusion.

► Signs of impending respiratory failure:

- Cyanosis
- Pallor
- Hypoxia (O₂ ≤ 92% in air) despite high-flow humidified oxygen
- Restlessness
- Drowsiness
- Silent chest
- Tachycardia
- PEFR persistently 30% of predicted for height or personal best.

Assessment of the child with acute asthma

Determine the severity of the attack (see Fig 16.17)

- Mild
- Moderate
- Severe
- Life-threatening

Too breathless to talk or eat?

Increased work of breathing

- Tachypnoea – severe if >30 breaths/min
- Chest recession:
 - Moderate – some intercostal recession
 - Severe – use of accessory neck muscles
 - Life-threatening – poor respiratory effort
- Auscultation:
 - Wheeze
 - Silent chest – poor air entry in life-threatening

Pulse:

- Severe – >120 beats/min

Level of consciousness – altered in life-threatening
Exhaustion

Tongue:

- Cyanosis in life threatening

Peak flow (% predicted):

- Moderate >50%
- Severe <50%
- Life-threatening <33%

O₂ saturation:

- Moderate >92%
- Severe or life-threatening <92%

Is there a trigger for the attack?:

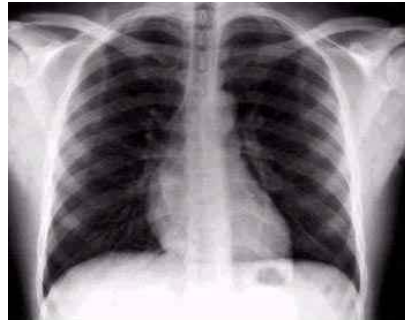
- URTI or other viral illness
- Pneumonia
- Allergen, e.g. animal dander
- Exercise
- Cold air



[Acute Asthmatic Attack] 2/2

* Investigations :

- Blood gases, and a chest X-ray may be required.
- Bilateral hyperinflation
- Depressed copulae of diaphragm
- May be atelectasis , pneumothorax



* Management :

- Acute management goals are:
 - to correct hypoxia,
 - to reverse airway obstruction
 - and to prevent progression
- Calm the patient
- Monitor the patient while giving high flow humidified O₂ via mask.
- Give inhaled β_2 -agonist (**salbutamol**) via a nebulizer.

Monitoring is needed as side-effects include irritability, tremor, tachycardia and hypokalemia.

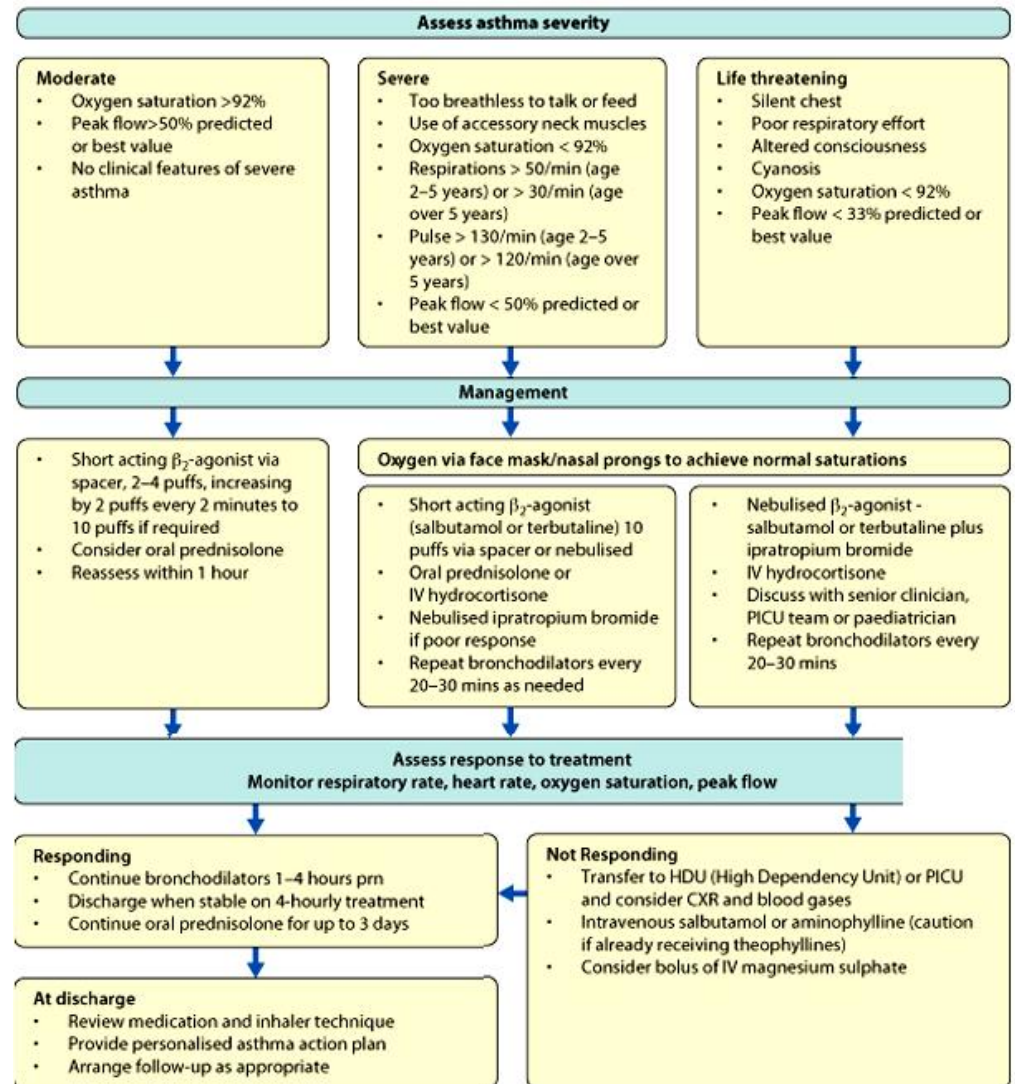
- Inhaled ipratropium bromide can be added.
- Oral prednisolone or IV hydrocortisone.
- Additional treatment if no improvement:
 - IV salbutamol,
 - IV magnesium sulphate
 - and IV aminophylline.

Antibiotics are unlikely to be beneficial

* Before Discharge :

- Review home management (controller therapy); consider changing to a combined steroid/long-acting β -agonist inhaler. This should improve adherence.
 - All asthmatics should have a written home management plan.
- Provide an asthma symptom diary and arrange hospital follow-up until control improves.
- Educate about allergen avoidance, ask about smoking – him and his family.
- Request adherence to treatment and parental guidance.

Assessment and management of acute asthma



[Bronchial Asthma (1)] 1/3

* History :

Mohamed an 8-year-old student born and raised in Helwan. He was referred for evaluation of bronchitis.

Over the last year, he received five courses of antibiotics for **episodes of cough , tight chest and audible wheeze precipitated by “colds” and aggravated by exercise or exposure to dust or smoke**. He had fever of three days duration at the onset of his first symptoms. He **used to cough at least two weeks after each episode** and his **symptoms are more prominent at night**.

Physical examination revealed no abnormality apart from **scattered expiratory ronchi** on chest auscultation.

He was prescribed amoxicillin 250 mg/6hrs, **salbutamol syrup/6hrs** and a cough syrup with **improvement of his symptoms**.

What is Your suggested diagnosis at this point ?

1. Immuno-deficiency
2. Bronchial asthma
3. Repeated bronchitis
4. Recurrent rhinopharyngitis

Answer : 2- Bronchial asthma

➤ When Should Cough/Wheeze be Called “Asthma”?

- When wheeze/cough becomes recurrent (with symptom free intervals).
- Airflow obstruction is at least partially reversible as shown by pulmonary function testing.
- When a number of known risk factors are present (atopy).
- When other wheeze/cough conditions have been excluded.
- When the child responds to anti-asthma therapy.

➤ Is it Asthma?

When Wheeze/Cough Becomes Recurrent (With Symptom Free Intervals)

- Recurrent episodes of wheezing.
- Troublesome cough at night.
- Cough or wheeze after exercise.
- Cough, wheeze or chest tightness after exposure to airborne allergens or pollutants.
- Colds “go to the chest” or take more than 10 days to clear.

➤ Airflow Obstruction Is At Least Partially Reversible (Pre - & Post - Bronchodilator)

Spirometry : Forced Expiratory Volume first second (FEV1

Peak Expiratory Flow Meter to measure Peak Expiratory Flow Rate (PEFr)



➤ When A Number Of Known Risk Factors Are Present (Atopy)

• Markers of atopy :

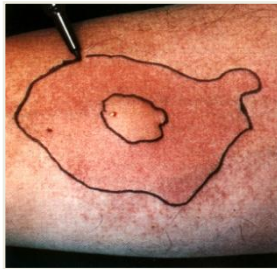
- Positive family history
- Other atopic manifestations
- Investigation :

- +ve Allergy Prick Skin Test (PST) - ↑ Serum total IgE
- ↑ specific IgE - Peripheral blood and tissue eosinophilia

[Bronchial Asthma (1)] 2/3

* Allergy Prick Skin Test :

- Number of allergens
- Quality of allergens (purification + standardization)
- Patient exposure (residence + occupation etc.)
- Quality control (positive + negative control)
- Interpretation + clinical correlation.



➤ When Other Wheeze/Cough Conditions Have Been Excluded.

(Differential diagnosis of asthma in children)

- Congenital malformation (laryngeal web, cyst, stenosis, TOF, vascular ring)
- Bronchiolitis
- Bronchiectasis
- Bronchopulmonary dysplasia
- Laryngotracheobronchitis
- Laryngotracheobronchomalacia
- Immunodeficiency syndromes
- Primary ciliary Dyskinesia
- Rhino-sinusitis
- Cystic fibrosis
- Foreign body
- Gastro-esophageal reflux
- Congenital heart disease
- Chronic respiratory infection
- Recurrent aspiration syndromes
- Vocal cord dysfunction

➤ Diagnosis of Bronchial Asthma is unlikely if :

• An atypical history may include:

- Onset of symptoms in the neonatal period
- History of ventilatory support in the neonatal period.
- Intractable wheezing that is unresponsive to bronchodilators.
- Wheezing associated with feeding; vomiting.
- The sudden onset of coughing or choking (FB).
- Stridor
- Steatorrhea (cystic fibrosis).

• Additional or Alternative Diagnosis:

- Failure thrive
- Clubbing
- A cardiac murmur
- Fixed monophonic/asymmetric wheeze
- No reversibility of airflow obstruction after administration of a bronchodilator
- A focal or persistent finding on chest radiograph.

➤ Exclude Other Conditions

- Structural problems: bronchoscopy
- Esophageal disease: Barium swallow, pH probes, scopes and gram
- Primary ciliary dyskinesia: nasal ciliary motility, Exhaled NO, EM, saccharine test
- TB: mantoux, induced sputum/ gastric lavage/ BAL = Culture, microscopy & PCR
- Bronchiectasis: HRCT scan, BAL
- CF: sweat test, nasal potentials, genotypes
- Systemic immune deficiency: Ig subtypes, lymphocytes & neutrophil function, HIV
- Cardiovascular disease: echo, angiography

[Bronchial Asthma (1)] 3/3

What would be the most helpful investigation at this point ?

1. Chest X-ray
2. Pulmonary function tests
3. Skin allergy test
4. Total and specific IgE

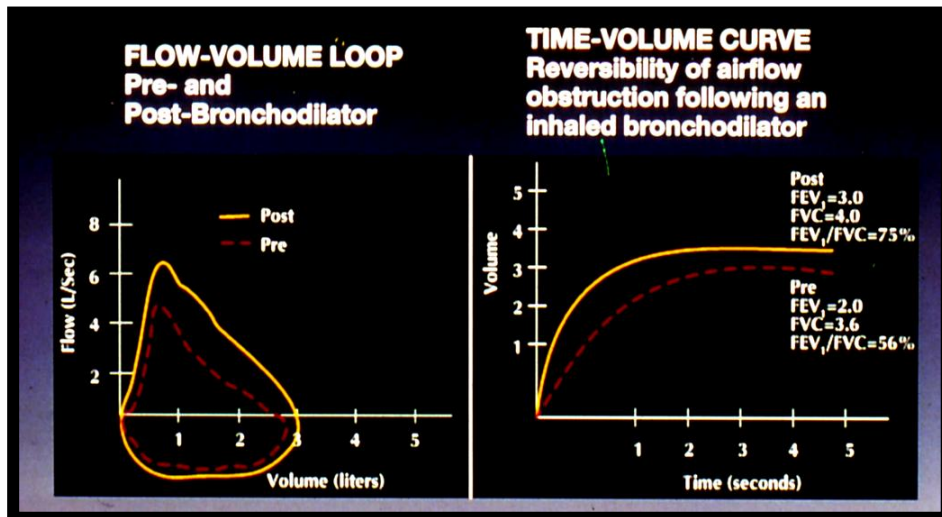
Answer : **2- Pulmonary function tests**

The most important lung function parameter to evaluate his condition is :

1. PEFR.
2. Spirometric evaluation pre & post bronchodilator.
3. Lung Diffusion .
4. Lung Capacities.

Answer : **2- Spirometric evaluation pre & post bronchodilator.**

Spirometry Pre & Post Bronchodilator
Forced Expiratory Volume first second (FEV1)



➤ Reversible and Variable Airflow Limitation :

• Reversible airway obstruction:

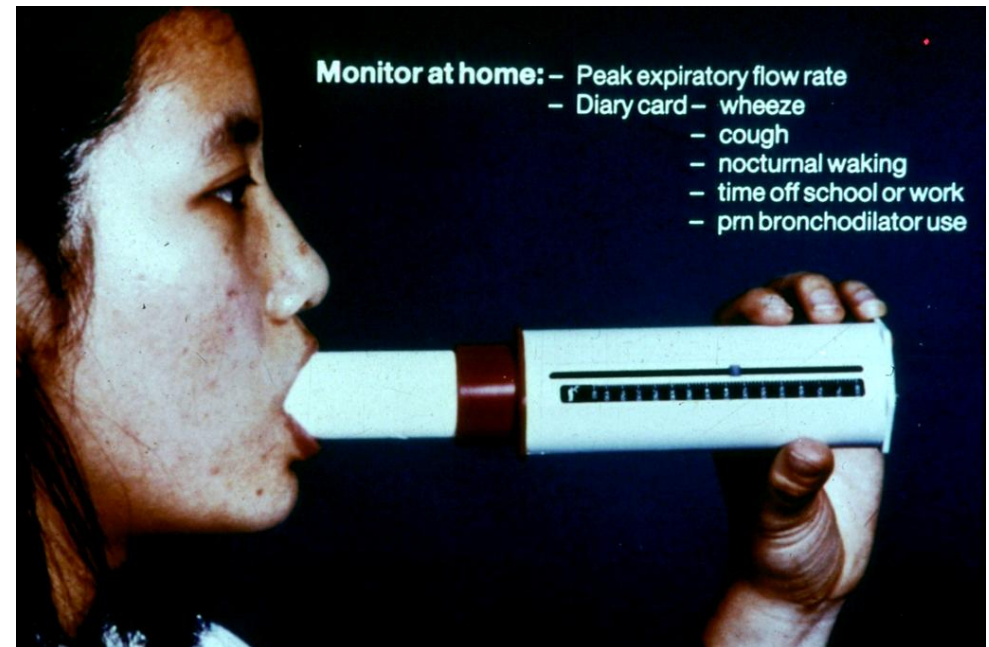
Increased FEV1 (PEFR) by 15% 15-20 after inhaling β 2-agonist.

• Variability of airflow obstruction:

> 20% variability in FEV1 (PEFR) between morning and evening values.

• Exercise-induced airway obstruction:

Decrease FEV1 (PEFR) \geq 15% after 6 minutes of exercise.



Important missing points that will help the diagnosis of asthma ?

1. Family history of atopy
2. History of atopic dermatitis or allergic rhinitis as a child
3. History of specific exposures
4. All the above

Answer : **4- All the above**

[Bronchial Asthma (2)]

* History :

Mohamed started to suffer frequent asthma symptoms and used Salbutamol inhaler (bronchodilator inhaler) on **daily** basis. He reported **nocturnal** symptoms waking him from sleep **2 to 3 nights** a week. Spirometry was done and FEV1 was **70%** of predicted.

In your opinion Mohamed is suffering from :

1. Mild intermittent asthma.
2. Mild persistent asthma.
3. Moderate persistent asthma.
4. Severe persistent asthma.

Answer : **3- Moderate persistent asthma.**



GINA (Global Initiative for Asthma) Classification of Severity

CLASSIFY SEVERITY Clinical Features Before Treatment			
	Symptoms	Nocturnal Symptoms	FEV ₁ or PEF
STEP 4 Severe Persistent	Continuous Limited physical activity	Frequent	≤ 60% predicted Variability > 30%
STEP 3 Moderate Persistent	Daily Attacks affect activity	> 1 time week	60 - 80% predicted Variability > 30%
STEP 2 Mild Persistent	> 1 time a week but < 1 time a day	> 2 times a month	≥ 80% predicted Variability 20 - 30%
STEP 1 Intermittent	< 1 time a week Asymptomatic & normal PEF between attacks	≤ 2 times a month	≥ 80% predicted Variability < 20%

The presence of one feature of severity is sufficient to place patients in that category.

Assessment of the child with chronic asthma

Clinical features to check

Growth and nutrition

Peak flow/spirometry

Chest for:

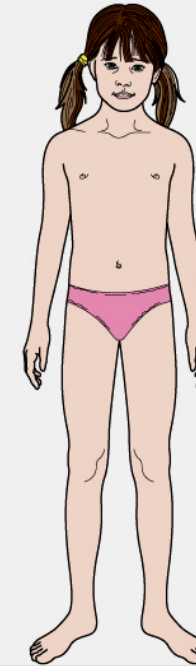
Hyperinflation
Harrison's sulcus
Wheeze

Are there other allergic disorders?

- Allergic rhinitis
- Eczema, etc.

If there is:

- Sputum
- Finger clubbing
- Growth failure
- If present, other causes should be sought



Monitor:

- Peak flow diary
- Severity and frequency of symptoms
- Exercise tolerance
- Interference with life, time off school
- Is sleep disturbed?
- Use of preventer and reliever medication – are they appropriate?
- Inhaler technique

Consider triggers:

- Allergic rhinitis needing treatment?
- Allergens - animal dander, etc.
- Stress

[Bronchial Asthma (3)] 1/2

* History :

Mohamed was prescribed Flexotide evohaler 125µg / puff BID (inhaled steroid) and to use Salbutamol inhaler (inhaled short acting bronchodilator) as needed.

He was also advised to keep a peak flow meter chart.

In a follow up visit 3 months later Mohamed was **still suffering** from **frequent nocturnal symptoms**, uses inhaled Salbutamol (inhaled bronchodilator) **>2 times/week** and his **PEF is 75%** of predicted.

In your opinion Mohamed's asthma is :

1. Partly controlled.
2. Uncontrolled.
3. Controlled.

Answer : 2- Uncontrolled.



Levels of Asthma Control

Characteristic	Controlled (All of the following)	Partly controlled (Any present in any week)	Uncontrolled
Daytime symptoms	None (2 or less / week)	More than twice / week	3 or more features of partly controlled asthma present in any week
Limitations of activities	None	Any	
Nocturnal symptoms / awakening	None	Any	
Need for rescue / "reliever" treatment	None (2 or less / week)	More than twice / week	
Lung function (PEF or FEV ₁)	Normal	<80% predicted or personal best (if known) on any day	
Exacerbation	None	One or more / year	1 in any week

The best therapeutic approach to consider at this point:

1. Double the dose of inhaled steroids.
2. Add on a long-acting beta agonist.
3. Add on a leukotriene modifier.
4. Options 1+2
5. Options 1+3

Answer : 4- Options 1+2

Table 16-2. Drugs in asthma

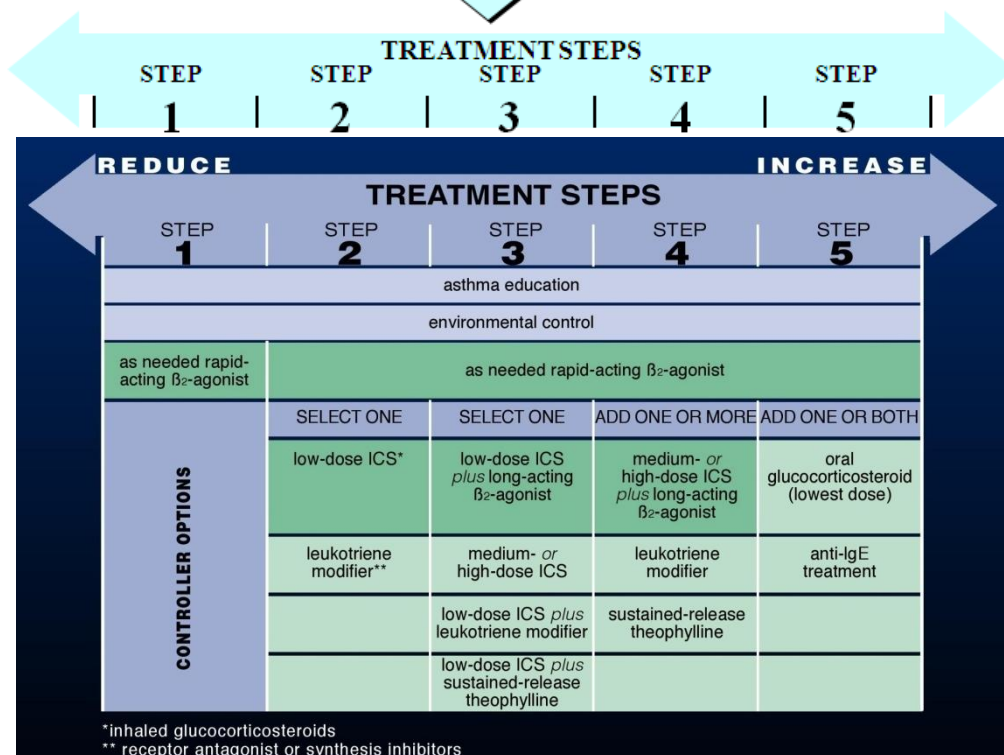
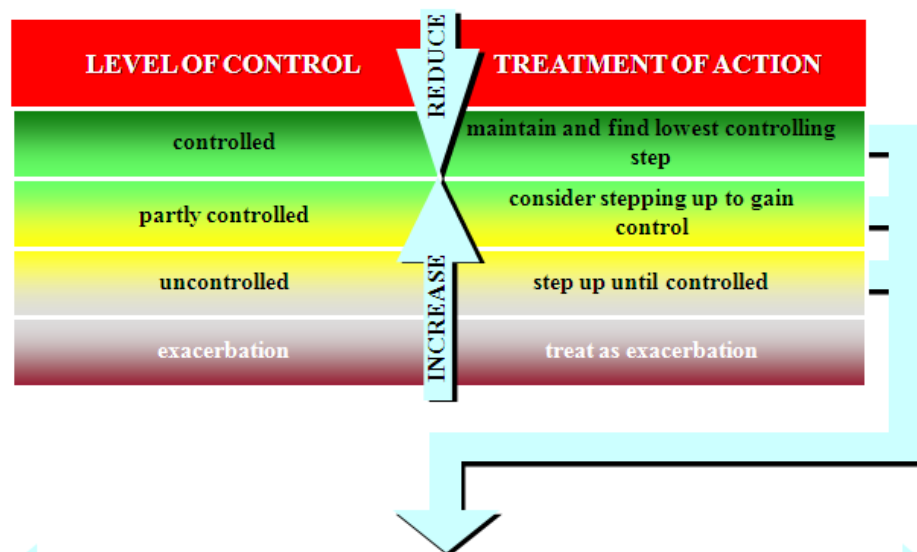
Type of drug	Drug
Bronchodilators	
β ₂ -agonists (relievers)	Salbutamol
	Terbutaline
Anticholinergic bronchodilator	Ipratropium bromide ^{Rx}
Preventative/prophylactic treatment	
Inhaled steroids	Budesonide ^{Rx}
	Beclometasone
	Fluticasone
	Mometasone
Long-acting β ₂ -bronchodilators	Salmeterol
	Formoterol
Methylxanthines	Theophylline ^{Rx}
Leukotriene inhibitors	Montelukast
Oral steroids	Prednisolone ^{Rx}

All are given by inhalation, except prednisolone^{Rx}, leukotriene modulators and theophylline^{Rx} preparations.

Controller Medications (Preventive / Prophylactic) :

- Inhaled glucocorticosteroids (ICS).
- Leukotriene modifiers - LTRA (montelukast).
- Long-acting inhaled β₂-agonists – LABA (Salmeterol- Formeterol).
- Long-acting theophylline.
- Cromones.
- Long-acting oral β₂-agonists.
- Systemic glucocorticosteroids.
- Anti-IgE.

[Bronchial Asthma (3)] 2/2



Mohamed's mother is now asking:

"For how long should he continue on medicines" ?

1. As soon as he gets better
2. Three months
3. Six months
4. As long as it takes

Answer : **2- Three months**

- Persistent asthma is most effectively controlled with daily long-term controller medications, especially topical anti inflammatory therapy.
- A stepwise approach to pharmacologic therapy is recommended to gain and maintain control of asthma.
- The amount and frequency of medication is dictated by asthma severity and degree of control.
- Therapy should be initiated at a higher level than the patient's step of severity at the onset to establish prompt control then stepped down.
- Continual monitoring is essential to ensure that asthma control is achieved . Step down therapy is essential to identify the minimum needed medication to maintain control
- Once control is achieved and sustained for several months (three months), a reduction in pharmacologic therapy is appropriate and helpful to identify the minimum therapy for maintaining control.
- Reduction in therapy should be gradual fearing from deterioration.

After establishing improvement, what is the next step in Mohamed's case management?

1. Decrease the dose of ICS
2. Stop LABA
3. Stop LTRA
4. Maintain the same treatment

Answer : **1- Decrease the dose of ICS**

[Bronchial Asthma (4)] 1/3

* History :

During an office visit with a new patient, the mother tells you that every time her one-year-old daughter Nada **has a cold, she has severe coughing and wheezing that lasts for two or three weeks.**

When she was prescribed **salbutamol syrup (bronchodilator)**, her symptoms got better, but she was fussy, couldn't sleep, and vomited. She **had four such colds in the last year**; the most recent occurred a month ago and **required admission in the ER for 24 hours**.

In the past year she has had 2 courses of systemic steroids. Nada does not have any symptoms now, but her mother is worried and asks you for help because **she doesn't want her daughter to suffer from asthma as she did till the age of 9 years.**

Physical examination revealed no abnormality apart from **scattered expiratory ronchi on chest auscultation.**

► When Should Cough/Wheeze be Called "Asthma"?

- When wheeze/cough becomes recurrent (with symptom free intervals).
- Airflow obstruction is at least partially reversible (FEV1- PEF).
- When a number of known risk factors are present (atopy).
- When other wheeze/cough conditions have been excluded.
- When the child responds to anti-asthma therapy.

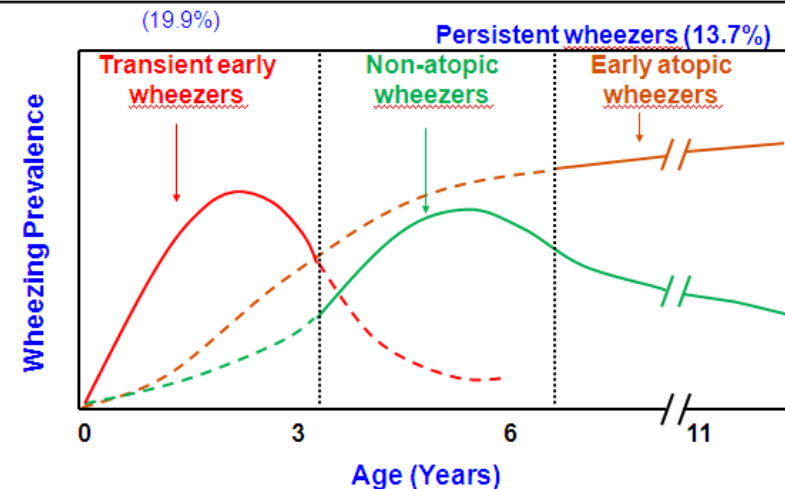
► Is it Asthma? When Wheeze/Cough Becomes Recurrent (With Symptom Free Intervals)

- Recurrent episodes of wheezing.
- Troublesome cough at night.
- Cough or wheeze after exercise.
- Cough, wheeze or chest tightness after exposure to airborne allergens or pollutants.
- Colds "go to the chest" or take more than 10 days to clear.

Causes of recurrent wheeze in infancy

- Transient early wheezing
- Non-atopic wheezing in the preschool child
- IgE-mediated wheezing (atopic asthma)
- Recurrent aspiration of feeds
- Cystic fibrosis
- Cow's milk protein intolerance
- Inhaled foreign body
- Congenital abnormality of lung, airway or heart
- Idiopathic

Wheezing Phenotypes In Infancy & Early Childhood



(51.5%) Never wheezed

Stein RT, et al Thorax 1997; 52: 946-952.

[Bronchial Asthma (4)] 2/3

➤ Early Atopic Wheezers

- ↑ airway responsiveness was related to a +ve FH of asthma & Other atopic manifestation.
- Normal initial level of lung function .
- Increased IgE levels .
- Tendency for persistence of symptoms.

➤ Transient Wheezers “Not Associated with Asthma Later in Life”

- Airflow obstruction prior to the first wheezy episode.
- Wheezing in the first 3 year of life.
- Associated with :

- Small airway caliber

- . Lung size
- . LBW
- . Male gender
- . Maternal smoking (Prenatal insults)

- Viral infections – environmental irritants

- . Lack of airway eosinophilia on BAL.
- . Poor response to inhaled corticosteroids.

• Early Wheezers: Predictive Index of Developing Asthma :

- Episodes of Wheezing +

• One Major Criteria :

[Parent with asthma / Atopic dermatitis / Aeroallergic sensitivity]

• or Two Minor Criteria :

[Food sensitivity / Peripheral eosinophilia / Wheezing not related to infection]



Levels of Asthma Control

Characteristic	Controlled (All of the following)	Partly controlled (Any present in any week)	Uncontrolled
Daytime symptoms	None (2 or less / week)	More than twice / week	3 or more features of partly controlled asthma present in any week
Limitations of activities	None	Any	
Nocturnal symptoms / awakening	None	Any	
Need for rescue / “reliever” treatment	None (2 or less / week)	More than twice / week	
Lung function (PEF or FEV ₁)	Normal	< 80% predicted or personal best (if known) on any day	1 in any week
Exacerbation	None	One or more / year	

* The challenging question is when to start controllers in infants & young children < 4 yrs ?

NAEPP expert panel considers initiation of a controller therapy in those young children who:

- Require symptomatic treatment > 2 times a wk
- Have had viral induced wheezing > every 6 wks that requires an inhaled bronchodilator > 6hr over 24 hr.
- Have had > 3 wheezy episodes in the past year that lasted > 1 day and affected sleep and have risk factors for asthma (intermittent asthma with allergic sensitization).
- With intermittent asthma and severe exacerbation are managed as moderately severe asthma.

[Bronchial Asthma (4)] 3/3

What treatment plan would you recommend for Nada's asthma?

- a- Change the bronchodilator to Terbutaline or Aminophylline.
- b- Reassure the mother because all infants outgrow their asthma.
- c- LABA 1 puff bid .
- d- Montelukast 4mg chewable tablets.
- e- ICS and Montelukast

Answer : **e- ICS and Montelukast**

Children 5 Years of Age and Younger GINA Management Approach Based on Control¹

Asthma Education, Environmental Control, and As-Needed Rapid-Acting Beta ₂ -Agonists		
Controlled on as-needed rapid-acting beta ₂ -agonists	Partly controlled on as-needed rapid-acting beta ₂ -agonists	Uncontrolled or partly controlled on low-dose ICS ^a
↓	↓	↓
Controller Options		
Continue as-needed rapid-acting beta ₂ -agonists	Low-dose ICS	Double low-dose ICS
	Leukotriene modifier	Low-dose ICS + leukotriene modifier

^aOral corticosteroids should be used only for treatment of acute severe exacerbations of asthma.

Shaded boxes represent preferred treatment options.

ICS = inhaled corticosteroids.

1. Global Initiative for Asthma. *Pocket Guide for Asthma Management and Prevention in Children 5 Years and Younger. A Pocket Guide for Physicians and Nurses*. 2009. www.ginasthma.org. Accessed 5 May 2009.

• **Controller Medications :**

- Inhaled glucocorticosteroids.
- Leukotriene modifiers - LTRA (montelukast).
- Long-acting inhaled β₂-agonists – LABA (Salmeterol- Formeterol).
- Long-acting theophylline.
- Cromones.
- Long-acting oral β₂-agonists.
- Systemic glucocorticosteroids.
- Anti-IgE.

• **FDA Approved Therapies for Infants & Young Children :**

- ICS budesonide nebulizer solution.

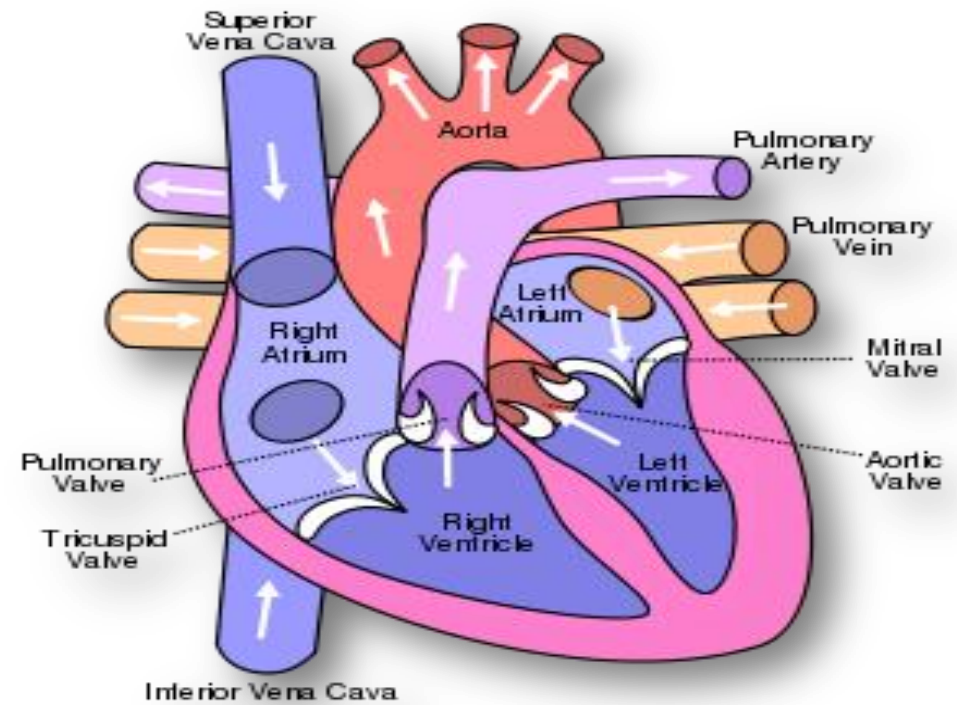
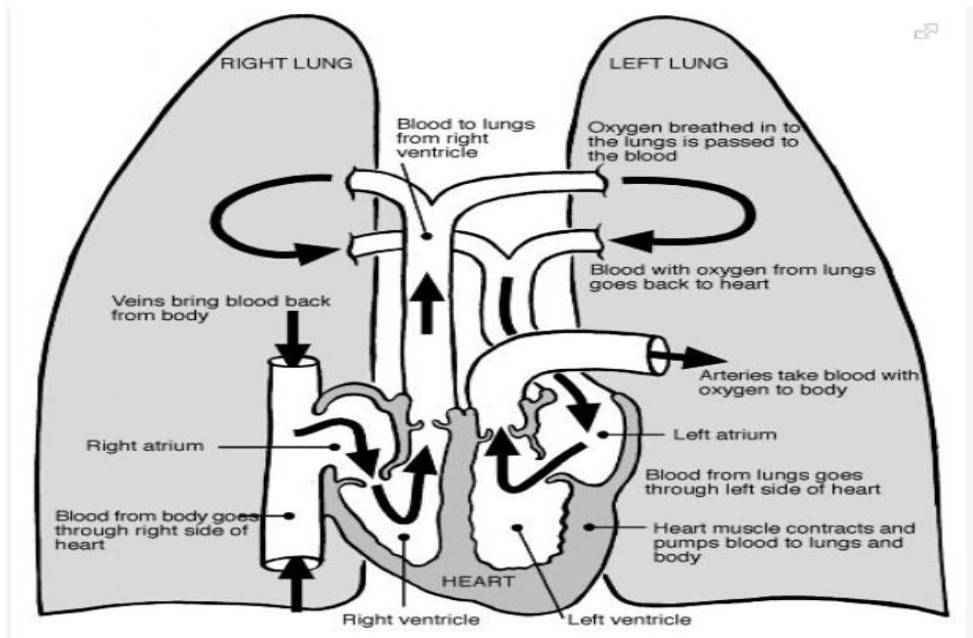
(The first and only approved nebulizer suspension 12 m and above by FDA and from 6 m outside USA)

- ICS fluticasone DPI (4 years of age and older).
- LABA and LABA/ICS combination DPI and MDI (4 years of age and older).
- Montelukast chewables (2-4 years), granules (down to 6 m of age).
- Cromolyn sodium nebulizer (2 years and older).

• **Factors predictive of progression from infantile wheezing to asthma in school age :**

- Allergen sensitization : - indoor allergens ≤ 3y
- early sensitivity (≤ 1y) to any allergen
- Family History : - parent with atopy or asthma
- Atopic dermatitis.
- Onset of wheezing after 2 years of age.
- RSV bronchiolitis requiring hospitalization in first year of life. (Rhinovirus-Coast study)
- Asthma predictive index.

Cardiology



Normal blood flow of heart and lungs

► Case (1) :

- An **11-year-old** boy presents with **fever** up to 39°C for the last 4 days, **joint pain** and **swelling**, along with **shortness of breath**.
- Two days ago, his **right knee** was painful and swollen, but today it has improved. The joints involved today include the **right ankle** and **left knee**. They are quite tender, painful and also swollen.

* History :

The shortness of breath occurs with walking, but he is now **unable to walk** because of the joint pain.

* Examination :

- Vital signs: **T 38.2°C**, **P 160/min**, **RR 32/min**, **BP 100/60 mmHg**, **O2 saturation 94%** in room air.
- He is tired appearing with tachypnea and tachycardia.
- HEENT: **Enlarged, erythematous tonsils with exudates**.
- Lungs are clear but with **tachypnea**.
- * HEENT = Head, Ears, Eyes, Nose and Throat examination
- Cardiac examination :
 - **Tachycardia**
 - Heart sounds are **distant** sounds
 - **Holosystolic murmur** 3/6 heard at apex with radiation to axilla.
 - No gallop
 - His point of maximal impulse (**PMI**) is prominent (size of silver dollar) at the **7th** intercostal space in the **mid-axillary line**.
 - His abdomen is soft with normoactive bowel sounds. Liver edge is **6 to 7** cm below the RCM.
 - His **left knee** is swollen and extremely **tender** with **warmth**. He has **difficulty with range of motion** but can flex his knee 30 degrees passively. His **right ankle** is very **swollen** and **warm**. He has limited subtalar motion.



Both his knee and ankle are very tender even to touch.

- Neurological exam: No abnormal movements of arms, hands, or tongue are noted. He is unable to walk due to pain.

* Clinical Course :

- The child is admitted to the hospital.
- Initial laboratory work includes an ESR of **110 mm/hr**, a CRP of **9.5 mg/L**
- A chest X-ray shows **cardiomegaly**.
- EKG reveals a **prolonged PR interval**.
- ASO titer is **754 units/ml** and streptozyme is **1:600**.



What is Your Clinical Diagnosis ?

[Acute Rheumatic Fever]

(**Arthritis + Carditis + Fever + Evidence of recent strept. Infection**)

• Evidence of recent Strept. Infection :

- ASOT • Anti-streptokinase • Antihyaluronidase • Throat swab

• Acute phase reactants

- ESR • CRP

Occasionally, in patients with acute arthritis, carditis may not be evident clinically, however, on Echo, there could be evidence of valvular affection. So, all patients with acute arthritis,

Echo is mandatory to rule out silent cardiac affection.

- Rheumatic carditis can be evident clinically as acute/subacute pancarditis or it may be silent and insidious to present later on as well established RHD with valvular affection.

*** Diagnosis :**

Final diagnosis should be:

- **Acute rheumatic fever**
- **Arthritis**
- **Carditis :**
 - Anatomical: MV disease (MR)
 - Functional: Decompensated HF
 - Complication: Not complicated by PHT or IE

Q1: What is the main difference between Rheumatic Heart Disease (RHD) and Acute Rheumatic Fever (ARF) ?

- a- In ARF there is an elevated ESR.
- b- In RHD there is a prolonged P-R interval.
- c- In ARF there is a history of arthralgias.
- d- In RHD there is evidence of chronic heart disease.
- e- In ARF there is evidence of erythema marginatum.

Answer : d- In RHD there is evidence of chronic heart disease

*** Management :**

- The patient is initially started on **salicylate** therapy at 75 mg/kg/day & his arthritis improves dramatically.
- However, the next day an echocardiogram confirms severe mitral insufficiency. Due to the significant cardiac disease with elements of congestive heart failure he is switched to **corticosteroids** and improves.
- Since the patient is presenting with carditis which was evident clinically, **steroids** should be initiated **immediately even if there is no evidence of heart failure.**

- Initiation of steroids should **NOT** be delayed until carditis is confirmed by echo.
- So, in that patient the initial management with salicylate was NOT proper.
- His heart size decreases over the next 2 weeks, and when it normalizes, he is switched back to **salicylates** for a total treatment duration of **8 weeks.**
- He does have a **persistent murmur** after this time however.
- He is started on **intramuscular benzathine penicillin**, which is given every 2-3 weeks for streptococcal prophylaxis.

Q2: All of the following are included in the revised Jones Major criteria EXCEPT :

- a- New murmur (carditis)
- b- Migrating polyarthritis
- c- Chorea
- d- Maculopapular rash
- e- Subcutaneous nodules

Answer : d- Maculopapular rash

Q3: A 7-year-old girl presents with a tender and swollen right knee as well as a more recently appearing swollen left wrist. She also has a fever. This patient fulfils which of the following revised Jones criteria ?

- a- 1 Major 1 minor
- b- 1 Major 2 minors
- c- 2 Majors
- d- 2 Minors
- e- 1 Major only

Answer : a- 1 Major 1 minor

Q4: Which of the following symptom lists of ARF are in the correct order of most common' least common ?

- a- Erythema marginatum, subcutaneous nodules, carditis, fever
- b- Arthritis, carditis, chorea, erythema marginatum
- c- Chorea, erythema marginatum, subcutaneous nodules, carditis, fever
- d- Arthritis, chorea, fever, carditis, subcutaneous nodules
- e- Fever, chorea, carditis, erythema marginatum

Answer : b- Arthritis, carditis, chorea, erythema marginatum

Q5: Salicylates are directed primarily at what symptom in ARF ?

- a- Rash
- b- Fever
- c- Arthritis
- d- Chorea
- e- Carditis

Answer : c- Arthritis

Q6: Corticosteroids are directed primarily at what symptom in ARF ?

- a- Rash
- b- Fever
- c- Arthritis
- d- Chorea
- e- Severe carditis

Answer : e- Severe carditis

What is the best antibiotic therapy for treatment of streptococcal infection ?

• **Penicillin :**

- Is the best ABO for strept. infection.
- One injection of LAP is sufficient.
- For clinical use, oral penicillins are commonly used (e.g. Oспен).
- Penicillins against β -lactamase activity are the best ABO (e.g. Augmentin, Unasyn , 2nd generation cephalosporins). They are better than oral penicillin Oспен as coorganisms are usually present which destroy penicillin ring by the β -lactamase E.

• **Erythromycin:** Orally if allergic to penicillin.

- **Sulfa drugs** (e.g. Bactrium and septrin) are contraindicated for treatment of streptococcal infection being bacteriostatic and not bacteriocidal, so they will not eradicate the infection nor prevent rheumatic fever.

For how long antibiotic therapy should be continued ?

• **10 Days**

Comment :

- ABO should be continued for 10 days to prevent development of rheumatic fever.
- For those with +ve streptococcal throat culture, you may prevent rheumatic fever, if proper ABO are given up to the 9th day following the culture.
- Streptococcal pharyngitis is highly suspected if associated with tender cervical lymphadenitis.

What does streptococcal carrier mean and how to manage ?

- **Streptococcal Carrier :**

- If throat culture is +ve for β .Strept.

ABO are given for 10 days.

- If throat culture is still +ve for β .Strept .

ABO are given for another 10 days

- If throat culture is still +ve for β .Strept.

NO ABO are to be given once more, as this patient is considered as a carrier.

► Case (2) :

- An **8-year-old** boy presents with **fever** of 2 weeks' duration, **shortness of breath, ankle edema, intermittent painful swelling of the wrists, elbows, and knees** unrelated to edema, and a **new systolic murmur**.
- One month ago, he had a **sore throat** that lasted for 5 days, which resolved spontaneously.

The most likely diagnosis is :

- a- Endocarditis
- b- Rheumatoid arthritis
- c- Meningococcal sepsis
- d- Glomerulonephritis
- e- Rheumatic fever

Answer : **e- Rheumatic fever**

Comment :

- When evaluating a child with acute onset arthritis, the differential diagnosis can be quite overwhelming.
- Certain elements of the history and physical can help lead to the correct diagnosis.
- In ARF: the **revised Jones criteria** are very helpful, but there are other findings which can also help confirm your suspicion of ARF.
- In ARF: the joints can be very painful, warm, sometimes erythematous, and very tender, yet the child is still fairly comfortable.
- **Juvenile Rheumatoid Arthritis ?** the joints swollen and without much tenderness, but very stiff in the morning.
- **Systemic Lupus Erythematosus ?** the effusions rather bland and non-tender lasting for a few days.
- **Septic joint ?** the joint so tender and swollen it can not be moved even a few degrees; the child usually has pain even at rest.

Infective Endocarditis

What is the most common microorganism found in pediatric infective endocarditis ?

- a- Staph. Aureus
- b- Strep. Viridians
- c- E. coli
- d - Pneumococci
- e- Strep. pyogenes

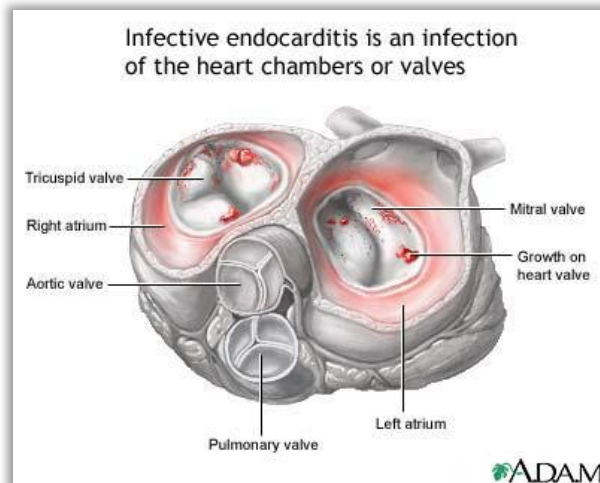
Answer : **b- Strep. Viridians**



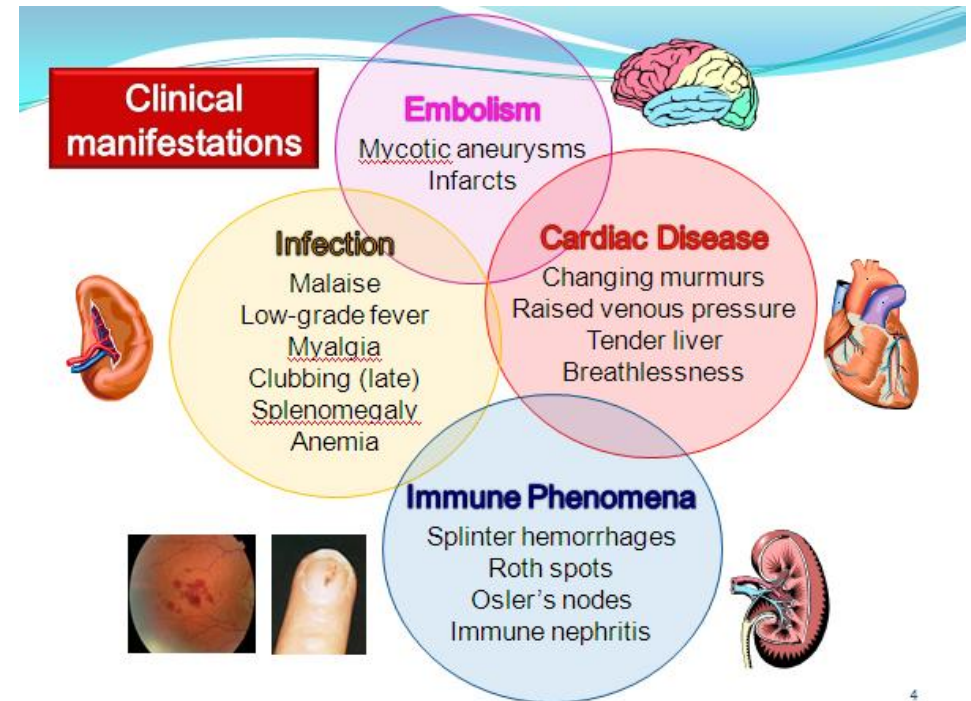
α -hemolysis

[Infective Endocarditis]

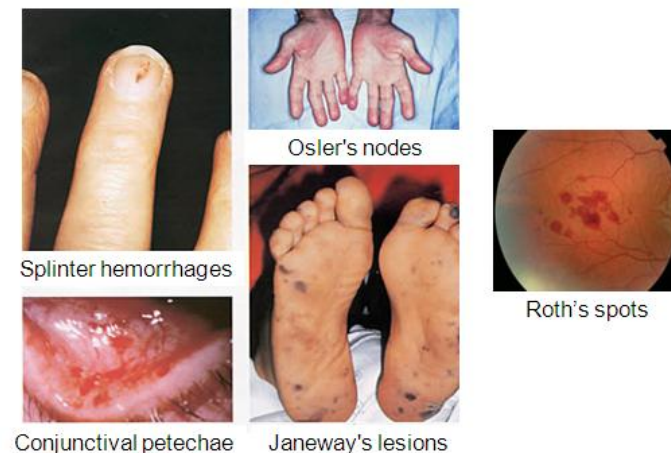
- All children of **any age** with congenital heart disease (**except secundum ASD**) are at risk of infective endocarditis.
- Infective endocarditis may occur on top of structurally normal heart.
- The most common causative organism is α -hemolytic streptococcus (**Strept. viridans**).



Cardiac Vegetations



Immune Manifestations



- Infective endocarditis should be suspected in any child or adult with a **sustained fever, malaise, raised ESR, unexplained anemia or hematuria.**
- The presence of the classical peripheral stigmata of infective endocarditis should **NOT** be relied upon.

What is the preferred antibiotic treatment for infective endocarditis caused by Strept. Viridians ?

- a- Penicillin G x 2 weeks
- b- Penicillin G x 6 weeks
- c- Oxacillin x 6 weeks

Answer : b- Penicillin G x 6 weeks

Comment :

- Penicillin G x 2 weeks → is too short course
- Oxacillin x 6 weeks → the preferred treatment for Staph. aureus infective endocarditis

What type of prophylactic antibiotic against infective endocarditis would you prescribe to a 9-year old female, with a past medical history only remarkable for an allergic reaction to penicillin, scheduled for a tooth extraction the next day ?

- a- Amoxicillin
- b- Ampicillin
- c- Clindamycin
- d- Cefazolin
- e- None

Answer : e- None

Comment :

- No antibiotics are needed, because this patient has no risk factors for infective endocarditis

Regarding infective endocarditis :

- a- The commonest causative organism is group A- β Streptococci. ()
- b- Amoxycillin is first choice for prophylaxis in procedures done under local anesthetic. ()
- c- Splenomegaly is common. ()
- d- Treatment consists prolonged antibiotic course; valve replacement may be required. ()

Answer : a- ✗ b- ✓ c- ✓ d- ✓

► Case (3) :

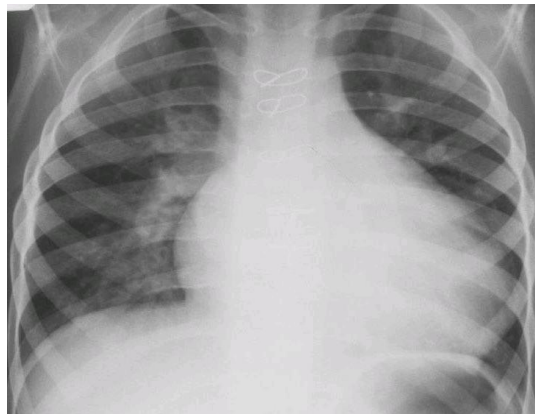
- A 6-week-old female infant is referred to hospital because of **wheezing**, **poor feeding** and **poor weight gain**, **excessive sweating** during the previous 2 weeks.
- Before this, she had been well.
- Her routine neonatal exam. had been normal.

* Examination :

- Vital signs: **RR 70/min**, **pulse 178/min** & BP 90/65 mmHg in the upper and lower extremities
- There is some sternal & **intercostal recession**.
- There are scattered **wheezes**.
- The liver is enlarged, palpable at **2 finger breadths** below the costal margin.
- A **thrill**, and a loud (grade 4) **pansystolic murmur** at the lower **left sternal edge** and a **mid-diastolic rumble** are noted.
- A slightly **accentuated** pulmonary component of the 2nd heart sound.

* Investigations :

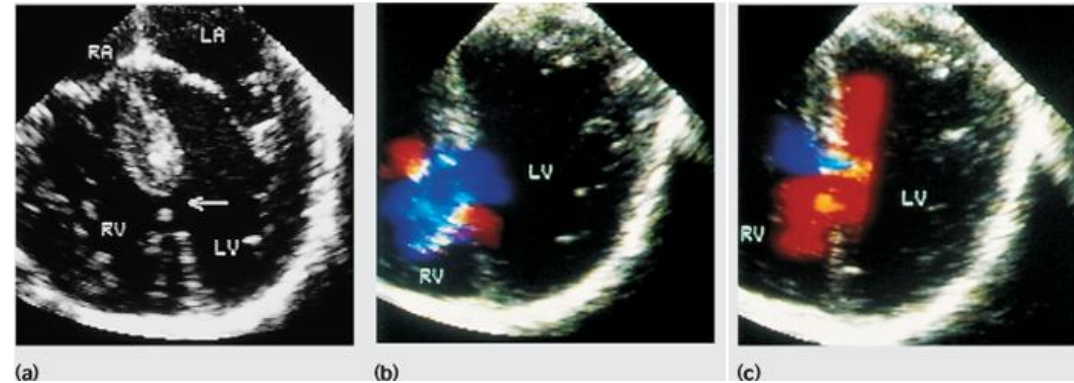
- Chest X-ray shows **cardiomegaly** & **increased pulmonary vascular markings**.
- ECG is normal.



What is the possible diagnosis ?

* Investigations :

- Echocardiogram shows a moderate-sized VSD.



- (a) Echocardiogram showing a medium-sized muscular VSD (arrow).
(b) Color Doppler shows a left-to-right shunt (blue) during systole.
(c) There is also a small right-to-left shunt (red) during diastole.

What is the proper management for this case ?

- a- Digoxin for HF. ()
b- Diuretics only. ()
c- Referral to surgery. ()
d- Reassurance for possible spontaneous closure. ()

Answer : a- ✗ b- ✗ c- ✗ d- ✗

Comment :

- Treatment with diuretics and captopril.

When to refer for intervention ?

Moderate to large defects with no response to medical treatment with failure to thrive

Explain how a child with an isolated VSD (acyanotic lesion) could become cyanotic ?

- 1- Congestive heart failure and pulmonary edema may cause hypoxia.
If the hypoxia is severe enough, visible cyanosis will result.
- 2- Long standing excessive pulmonary blood flow
→ Eisenmenger's syndrome.

Which of the following are true regarding a child with a large solitary VSD ?

- a- Cardiomegaly is common. ()
b- Second heart sounds will be normal. ()
c- CXR will show normal fields. ()
d- Symptoms are often not apparent until 3-6 wks of age. ()
e- Can present with failure to thrive. ()

Answer : a- ✓ b- ✗ c- ✗ d- ✓ e- ✓

Which of the following statements are true about VSD's ?

- a- Murmur characteristically radiates to the neck. ()
b- Patients are asymptomatic at birth. ()
c- Disappearance of the murmur is always reassuring, as it indicates that the defect has closed. ()
d- A diastolic murmur at the apex is due to increased flow through the defect. ()
e- Bacterial endocarditis is always a risk, even in hemodynamically insignificant defects. ()

Answer : a- ✗ b- ✓ c- ✗ d- ✗ e- ✓

In an infant, which of the following make cardiac failure unlikely ?

- a- Tachypnea ++
b- Tachycardia ++
c- An enlarged liver
d- Ascites
e- Excessive perspiration

Answer : d- Ascites

The earliest sign of congestive heart failure on a chest X-ray is :

- a- Increased heart size
b- Central pulmonary vascular congestion
c- Pulmonary edema
d- Pleural effusion

Answer : a- Increased heart size

➤ **Case (4) :**

• A **7-year-old** girl with **Down syndrome** has recurrent chest infections since birth and then she recently developed **central cyanosis** 1 year ago. Cardiac examination shows only a soft ESM at the upper left sternal border.

a- She has Fallot's tetralogy

b- Her hematocrit is likely to be 30%

c- Dental extraction should be covered by antibiotic prophylaxis

Answer : a- ✗ b- ✗ c- ✓

Comment :

- She most likely has **Eisenmenger's syndrome** secondary to a reversed VSD shunt.
- Most probably she will have **2ry polycythemia** and a raised hematocrit.
- She is a high risk for the development of **infective endocarditis**; therefore she should receive antibiotic prophylaxis.

➤ **Case (5) :**

• A **6-mo-old** boy is presented with **tachycardia**, **tachypnea**, and **poor feeding** for 3 months.

• Examination reveals a **continuous machinery murmur** and a **wide pulse pressure** with a prominent apical impulse.

The most likely diagnosis is:

a- Pulmonary Stenosis

b- Aortic Stenosis

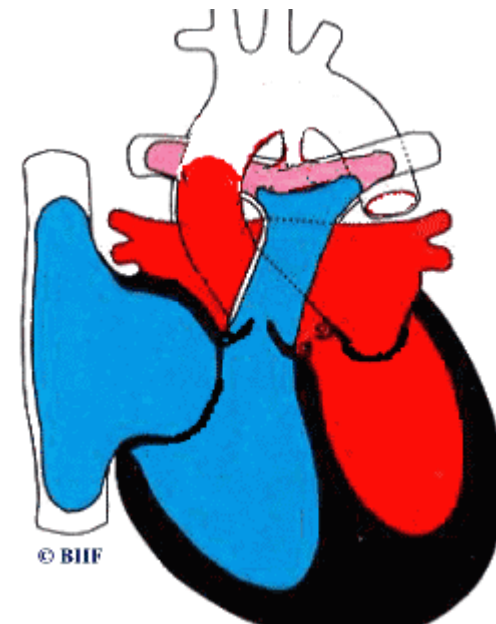
c- Ventricular septal defect

d- Patent ductus arteriosus

Answer : **d- Patent ductus arteriosus**

Comment :

- A PDA often presents like VSD except that there is a **continuous murmur** as well as the diastolic runoff, which produces **wide pulse pressure & bounding pulses**



► **Case (6) :**

- A **3-month-old** male infant who presents to the emergency department with a history of having episodes of **excessive crying** followed by **limpness, cyanosis** and **fainting**.
- He was born at 41 weeks of gestation by C-section because of failure to progress to a 23 year old mother. Apgar scores of 7 and 8 at 1 and 5 minutes, respectively.

*** History :**

- His cyanosis **increased** with crying and he had a grade 3/6 **ejection systolic murmur** along the **upper left sternal border**. His O₂ saturations were 95% and stable. He was discharged from the hospital and followed in the office until this episode.
- He is now being hospitalized.

Q1: Cyanosis is produced by the presence of deoxygenated hemoglobin of at least :

- a- 1-2 gm/dl
- b- 3-5 gm/dl
- c- 6-8 gm/dl
- d- 9-10 gm/dl

Answer : **b- 3-5 gm/dl**

*** Examination :**

- Vital signs T 37°C, P 164/min, RR 64/min, O₂ saturation 83% on oxygen by nasal prongs.
- Weight 50th percentile.
- He is alert and active in **mild** respiratory distress, with **visible** cyanosis.
- HEENT exam is negative.
- His heart rhythm is tachycardic.
- His lungs are clear.

- He has a mild **right** precordial heave with a grade **3/6 ejection murmur** at **upper left sternal border** and a **diminished 2nd heart sound**.
- Liver and spleen are not enlarged.
- He has normal peripheral pulses with cyanotic nail beds and mucous membranes.

*** Clinical Course :**

- An echo is obtained which identifies cyanotic CHD. This is confirmed at cardiac catheterization.
- He subsequently undergoes **palliative surgery** with improved oxygenation and appearance of a **continuous murmur**.
- He is discharged in stable condition to be followed on an outpatient basis and to undergo further corrective surgery at a later date.

Q2: The most likely underlying lesion is :

- a- Hypoplastic left heart
- b- Transposition of the Great Vessels
- c- Anomalous Pulmonary Venous Return
- d- Tetralogy of Fallot
- e- Aspiration with obstruction to air passages

Answer : **d- Tetralogy of Fallot**

Q3: Pulmonary vascularity is increased in all of the following except :

- a- TAPVR
- b- Tricuspid Atresia
- c- TGA
- d- Hypoplastic left heart

Answer : **b- Tricuspid Atresia**



Q4: Pulmonary vascularity is decreased in all of the following except :

- a- Tetralogy of Fallot
- b- Pulmonary Atresia
- c- TAPVR
- d- Tricuspid atresia

Answer : c- TAPVR

Q5: A cyanotic attack in tetralogy of Fallot is characterised by which of the following ?

- a- Cyanosis. ()
- b- Bradycardia. ()
- c- Increase in severity of systolic murmur. ()
- d- Apnea. ()
- e- Impaired consciousness. ()

Answer : a- ✓ b- ✗ c- ✗ d- ✗ e- ✓

Q6: A "blue" spell of tetralogy of Fallot is treated with all of the following EXCEPT :

- a- Knee-chest position
- b- Oxygen
- c- Adrenaline
- d- Morphine
- e- Sodium bicarbonate
- f- Propranolol

Answer : c- Adrenaline

Comment :

- Adrenaline is potentially dangerous

Q7: Recognized complications of tetralogy of Fallot include all of the following EXCEPT :

- a- Cerebral thrombosis
- b- Cerebral abscess
- c- Infective endocarditis
- d- Heart failure
- e- Hypoglycemia

Answer : e- Hypoglycemia

Comment :

- Cerebrovascular accidents result from a combination of polycythemia and the right-to-left shunting in patients with tetralogy of Fallot.
- Strokes are more common in children <2 year and in those with iron-deficiency anemia.
- Cerebral abscess formation is more common in children >2 year.

► Case (7) :

- A **5-hour-old** male newborn on the postnatal ward is noticed by the midwife because he looks **blue** around the **lips and tongue**.
- He is the first child of a 27-year-old mother with asthma who was taking inhaled steroids throughout pregnancy. Antenatal scans were **unremarkable**.

She went into spontaneous labor at **41 weeks** and there was thin meconium staining of the liquor when the membranes ruptured 1 hour before delivery.

* History :

- Cardiotocograph monitoring during labor revealed **normal variability** of fetal heart rate.
- The baby was born by normal vaginal delivery and weighed 3.3 kg. The Apgar scores were 7 at 1 min and **8** at 5 min.

* Examination :

- The baby is not dysmorphic.
- His temperature is 36.6°C and his central capillary refill time is **2 seconds**.
- His **lips, tongue & extremities** are **cyanosed**.
- He is crying **normally** and has **no signs** of increased respiratory effort.
- Heart rate is 160 beats/min, femoral pulses are palpable, heart sounds are normal and **no murmur** is audible.
- Oxygen saturation is **70%** in air and does **not rise** with facial oxygen, which has been administered by the midwife.
- There is no hepatosplenomegaly.

* Investigations :

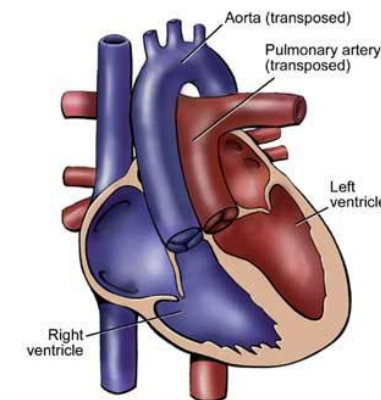
Arterial/blood gas		Normal
In air		
pH	7.25	7.35-7.42
PaO ₂	35.3 mmHg	69.7 – 99.7 mmHg
PaCO ₂	27.6 mmHg	35 - 45 mmHg
HCO ₃	11.5	22 - 26 mEq/L
After 10 min in high flow facemask oxygen		
pH	7.25	7.35-7.42
PaO ₂	39.9 mmHg	69.7 – 99.7 mmHg
PaCO ₂	28.1 mmHg	35 - 45 mmHg
HCO ₃	12.2	22 - 26 mEq/L

What is the interpretation of the patient's blood gases ?

There is evidence of tissue hypoxia in this case, as there is a **metabolic acidosis**.

What is the likely diagnosis & differential diagnosis ?

This baby is most likely to have transposition of the great arteries (**TGA**).



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➤ **Differential diagnosis of a cyanotic newborn :**

- Central
- Respiratory
- Cardiac
- PPHN

*** Hyperoxia Test :**

- It provides a means of diagnosing whether cyanosis is due to **cardiac** or **respiratory** disease.
- **Normally** arterial PaO₂ is **>67.5 mmHg** and rises to **>150 mmHg** after exposure to O₂ 90–100%.
- If the PaO₂ **fails to rise**, this is strongly suggestive of cyanotic HD.
- Persistent fetal circulation can also result in a lack of response.
- Persistent fetal circulation also results in cyanosis, usually with respiratory distress in the context of a newborn who has:
 - suffered a significant hypoxic, hypothermic or hypoglycemic insult,
 - pulmonary hypoplasia or sepsis,
 - or sometimes for unknown reasons.

• Diagnosis :

- In congenital cyanotic heart disease, definite clinical diagnosis is not required.
- Clinical diagnosis should be as such:

Congenital cyanotic heart disease with increased or decreased pulmonary blood flow as evident clinically by (history and clinical examination) for differential diagnosis.

• **Definite diagnosis will be achieved by :**

- Chest-X ray
- ECG
- Echocardiogram

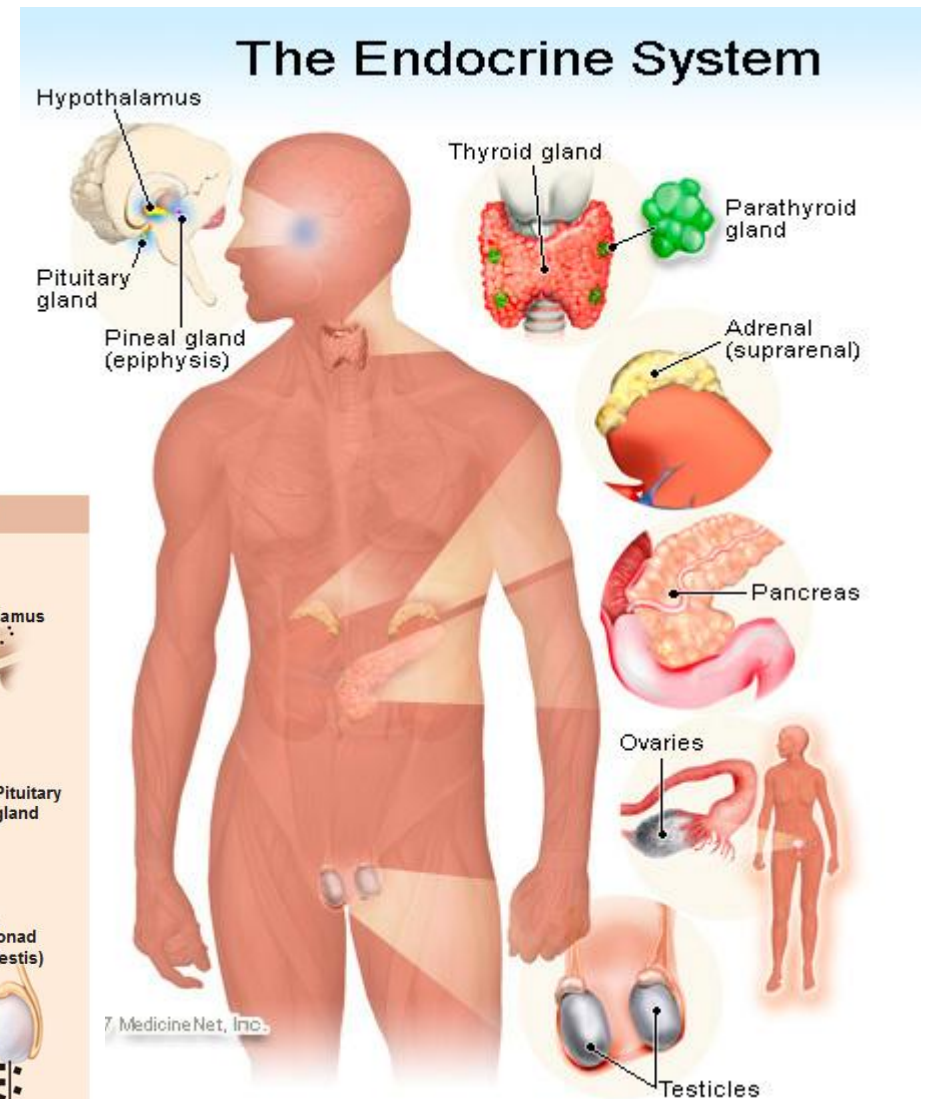
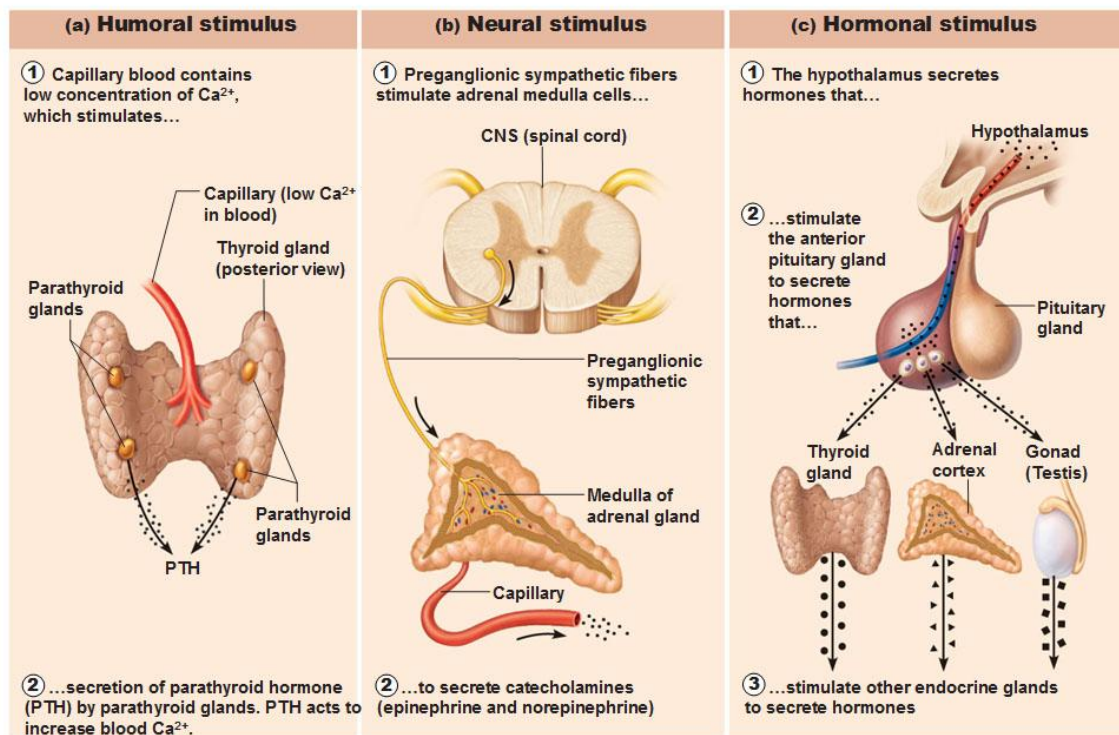
What is emergency management for this patient ?

Urgent echocardiography is mandatory

- **Prostaglandin infusion** to maintain patency of the ductus arteriosus.
- **Metabolic acidosis** correction.
- **Balloon atrial septostomy** to improve mixing of oxygenated and deoxygenated blood at the atrial level.
- **Arterial switch operation (ASO)**, in the first few days of life.

Endocrinology

Types of Endocrine Gland Stimuli



➤ Case (1) :

* History :

- Rasha is a 4-year-old girl brought to the GP by her mother, who is worried about her daughter's growth. She has noticed that her shoe size has not changed for almost 12 months and she is still in clothes for a 2- to 3-year-old. She was born at 38 weeks by normal delivery and weighed 2.1 kg. Her mother tried breast-feeding but she was never easy to feed, even with a bottle.
- She is generally healthy, apart from recurrent ear infections. She wears glasses for long-sightedness. Her development is normal, although nursery staff have reported that she seems to have poor concentration. The mother said that her hands and feet were puffy at birth.



* Examination :

- She is generally healthy and certainly well nourished. The GP notices wide-spaced nipples, neck webbing, wide carrying angle with no other detected abnormalities

What are the clinical signs that suggest a pathological cause for short stature ?

What is the most likely diagnosis ?

➤ The clinical signs that suggest a pathological cause for short stature :

- Extreme short stature – on or below 5th centile.
- Short for family size – outside target range for parents
- Short and relatively overweight – suggests an endocrinopathy
- Short and very underweight – suggests poor nutrition /malabsorption
- Growth failure – crossing the centiles downwards
- Dysmorphic features
- Skeletal disproportion – charts available for ratio between sitting height and leg length. Significant disproportion suggests a skeletal dysplasia, e.g. achondroplasia
- Signs of systemic disease, e.g. clubbing

➤ Does our girl have pathological short stature ?

- This girl is extremely short and, short for family
- She has dysmorphic features (wide spaced nipple, webbed neck, wide carrying angle).
- She is also relatively overweight for height
- She had low birth weight, difficulty to feed in infancy, middle ear disease, visual problems and poor concentration.

What is the most likely diagnosis ?

- | | |
|---------------------------|--------------------|
| a- Familial short stature | e- Turner syndrome |
| b- CDGP | f- Rickets |
| c- Achondroplasia | g- Malabsorption |
| d- Cushing syndrome | |

Answer : e- Hypoglycemia

How to prove the diagnosis of Turner syndrome ?

- Chromosomal analysis

➤ Case (2) :

* History :

- Omar is a 7-year-old boy who presents to pediatric outpatients because he's overweight.
- His father became concerned 1 year ago. His mother is overweight, hypertensive and has type 2 diabetes, but his father's weight is average.
- His father says the boy's diet is generally good but that his grandmother spoils him.
- He does sports twice a week at school. He is bullied at school about his weight.
- His birth weight was 3.8 kg and there were no problems in the neonatal period.
- He snores every night.
- He is on no medication.
- His development is normal.
- No other diseases run in the family.

* Examination :

- There are no dysmorphic features.
- There are some pink abdominal stretch marks.
- There is no acanthosis nigricans and no goitre.
- His blood pressure is 116/75 mmHg.
- His height is 125 cm (75th centile) and his weight is 38.7 kg (97th centile).
- His BMI = $\text{weight (kg)} / \text{height (M}^2\text{)} = 24.8$

* Investigations :

- Full blood count, urea and electrolytes, liver and thyroid function tests are normal.
- His fasting glucose, insulin, cholesterol and triglycerides are normal.

What is the most likely cause of this child's obesity ?

How should this child be treated ?

➤ Cause of obesity :

- **Simple obesity**
- **Genetic**, i.e. one or both parents is obese
- **Endocrine disease**, e.g. hypothyroidism, Cushing's syndrome, growth hormone deficiency, pseudohypoparathyroidism
- **Drugs**, e.g. steroids, sodium valproate
- **Syndromes**, e.g. Down's, Prader-Willi and Laurence-Moon-Biedl
- **Disorders associated with immobility**, e.g. cerebral palsy
- **Hypothalamic damage**, e.g. secondary to trauma or brain tumours
- Rarely, **single gene mutations**, such as those of the melanocortin-4 receptor and leptin

Most likely diagnosis of our boy ?

Simple obesity :

- Obesity due to caloric intake exceeding energy expenditure. It is the commonest cause of obesity and is not due to any underlying pathology.
- Children with simple obesity tend to be tall and overweight.
- Those who are short and overweight are more likely to have underlying pathology, such as an endocrine disorder

➤ Complications of obesity :

- Children with obesity should be assessed for **co-morbidities** such as:
 - hypertension
 - dyslipidaemia
 - breathlessness on exertion, obstructive sleep apnea,
 - hyperinsulinaemia and type 2 diabetes
 - knock-knees or bow legs,
 - polycystic ovary syndrome and psychosocial dysfunction.

➤ Treatment of simple obesity :

- Dietetic input is very important.
- Children should be encouraged to exercise for 60 min/day.
- Sedentary activities such as playing computer games should be discouraged.
- Obesity can lead to low self-esteem and, in our case, Omar is being bullied. He would therefore benefit from seeing a psychologist, to initiate behavioral therapies to help treat the obesity.

► Case (3) :

* History :

- Samira is a 4-year-old girl seen in the pediatric day unit with a 2-week history of polydipsia and polyuria. Having been dry at night for some time, she has also started wetting the bed. Her mother thinks that he has lost some weight.
- She has been less cheerful than usual and hasn't wanted to go to school – her mother has put this down to tiredness at the end of her first term. There is no significant past medical history and she is fully immunized. She has a 2-year-old brother who is well.

* Examination :

- Samira is playing happily in the playroom. Her weight is on the 10th centile and her height is on the 50th centile. Her trousers are a bit loose round her waist. She is a febrile. She is not dehydrated. Her pulse is 84 beats/min. Examination of the respiratory and abdominal systems is normal.

* Investigations :

		Normal
Haemoglobin	12.3 g/dL	11.5–15.5 g/dL
White cell count	$8.4 \times 10^9/L$	$6-17.5 \times 10^9/L$
Platelets	$365 \times 10^9/L$	$150-400 \times 10^9/L$
Sodium	138 mmol/L	138–146 mmol/L
Potassium	4.5 mmol/L	3.5–5.0 mmol/L
Urea	4.2 mmol/L	1.8–6.4 mmol/L
Creatinine	46 μ mol/L	27–62 μ mol/L
Glucose	22.4 mmol/L	3.3–5.5 mmol/L
Venous blood gas		
pH	7.35	7.35–7.45
PCO ₂	4.3 kPa	4.7–6.4 kPa
Bicarbonate	19 mmol/L	22–29 mmol/L
Urinalysis	Glucose + + +, ketones + + +	

What is the diagnosis ?

type 1 diabetes mellitus (T1DM), by far the commonest form of diabetes in childhood characterized by pancreatic β -cell dysfunction with insulin deficiency. The precise mechanism is not understood, but environmental factors probably 'trigger' a T-cell-mediated autoimmune process in those genetically susceptible.

- There may be a history of other autoimmune diseases. The incidence is rising. The incidence of type 2 is also rising alongside obesity.

What should happen next ?

Although this boy has ketonuria, he is not acidotic and does not have diabetic ketoacidosis (DKA) – a widely accepted biochemical definition being a pH less than 7.30 and/or a bicarbonate less than 15 mmol/L.

➔ **So if insulin treatment is not started within 24 hours, DKA will develop.**

- Current best practice states that insulin is best delivered by a 'basal bolus' regime – background 'basal' insulin given once/twice daily with rapid-acting 'bolus' insulin at mealtimes.

List the topics that need to be discussed with his family ?

- regular finger-prick blood testing – up to four times daily
- interpreting blood glucose results and altering insulin
- recognition and treatment of hypoglycemia – 'hypos' really worry parents and older patients, especially at night
- management during intercurrent illness, e.g. flu
- blood or urine ketone estimations
- who to contact in an emergency
- diet – matching insulin to carbohydrate in a 'basal-bolus' regime
- school – how to deal with it
- exercise
- the 'honeymoon' period
- long-term complications – discuss even at outset because many families know about and fear complications and need objective information
- using glycosylated hemoglobin (HbA1c) to measure overall control

► Follow up of patients with T1D :

- Growth assessment
- Blood pressure
- Site of insulin injections
- Self blood glucose monitoring readings
- Peripheral sensations and pulses
- Other autoimmune diseases
- Hb A1c every 3 months

Annual labs: Lipid profile, Albumin/creatinine ratio in urine,
Thyroid profile & Celiac screening

➤ **Case Scenario (4) :**

• **A mother came to you**, bringing her 1-month old infant, complaining of the following :

- Decreased activity
- Poor feeding
- Constipation
- Jaundice
- Hoarse cry

• **On examination**, you found the following :

- Full term infant
- BW: 2 kg
- Jaundice
- Opened posterior fontanel
- Hypotonia
- sluggish activity

• **You asked the mother if a sample was taken by heel stick for newborn screening in the first week or later, the mother denied**

• You thought of **hypothyroidism** as a cause and you ordered the following tests :

- FT3: low
- FT4: low
- TSH: high

• You ordered an Xray of the Knee joint, Thyroid ultrasound & Thyroid scan

➤ **Neonatal Thyroid screening :**

- Impregnating filter paper with a drop of blood obtained by a heel stick on day 5.
- A RIA determines TSH and may detect T4

➤ **Case Scenario (5) :**

• **A mother came to you**, bringing her 1-year-old infant , complaining of the following :

- Head lag: No head support
- Her infant cannot sit alone or even with support
- Her infant cannot recognize her as his own mother

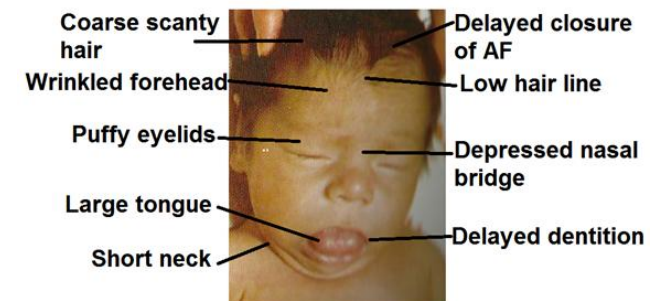
• **On examination**, you found the following :

- Decreased length: below 3rd centile
- Macroglossia
- Large head
- Coarse hair , low anterior hair line
- Swollen eye lids, depressed nasal bridge
- Mottled, cool, dry skin
- Umbilical hernia
- myxedema
- Goiter
- No head support,
- No social smile

• You thought of **hypothyroidism** as a cause and you ordered the following tests :

- FT3: low
- FT4: low
- TSH: high
- Bone age: 4 months
- Thyroid scan: enlarged thyroid
- Thyroid ultrasound: enlarged thyroid
- Dyshormonogenesis

Head & neck examination



► Case Scenario (6) :

• A 16- year-old child came **presenting** with the following :

- Short stature
- Delayed puberty
- School underachievement
- Laziness
- Constipation
- Cold intolerance

• On examination :

- Short stature: below 3rd percentile
- Delayed puberty: pre-pubertal
- Goiter
- Dry skin
- Cold peripheries
- Bradycardia
- Pale puffy eyes with loss of eyebrows

• Investigations :

- Serum free T4: < 5 ug/dl
- Serum TSH: 100 u unit/ml
- normal 2-10 u unit/ml (up to 20 in 1st week)
- Thyroid scan: ectopic thyroid
- Bone age: delayed: 6 years
- Thyroid ultrasound: ectopic neck mass, thyroid

► Diagnose Morbidity/complications :

- **Profound mental retardation** : most serious
- **Linear growth and bone maturation** : severe retardation, absolute arrest
- **Neurologic problems** such as spasticity and gait abnormalities, dysarthria or mutism, and autistic behavior, myopathy, neuropathy, motor inco-ordination, hypotonia, neurosensory hearing loss, stabisimus.

► Exclude the following Health Hazards :

- **Hematologic** : anemia, hypercholesterolemia
- **Cardiac** : pericardial effusion
- **Hypothalamo-pituitary-axis** : delayed puberty, precocious puberty
- **Mental** : short attention span, memory loss

► Case Scenario (7) :

• A 12- year-old child came presenting with the following:

- School underachievement
- Laziness
- Constipation
- Cold intolerance
- The symptoms started only 6 months ago

• On examination :

- Pale puffy eyes
- Goiter
- Dry skin
- Cold peripheries
- Bradycardia

>> it's **Acquired Hypothyroidism**

- Chronic lymphocytic thyroiditis
- Thyroidectomy
- Irradiation
- Goitrogen drugs (iodides)
- Infiltrative disease
- With acquired hypopituitarism

► **Management of hypothyroidism :**

• **L-Thyroxine orally for life**

Neonates: 10-15 ug/kg (37.5-50 ug/24hr)

Infants: 6-8 ug/kg

Children: 4 ug/kg

Adults: 2 ug/kg

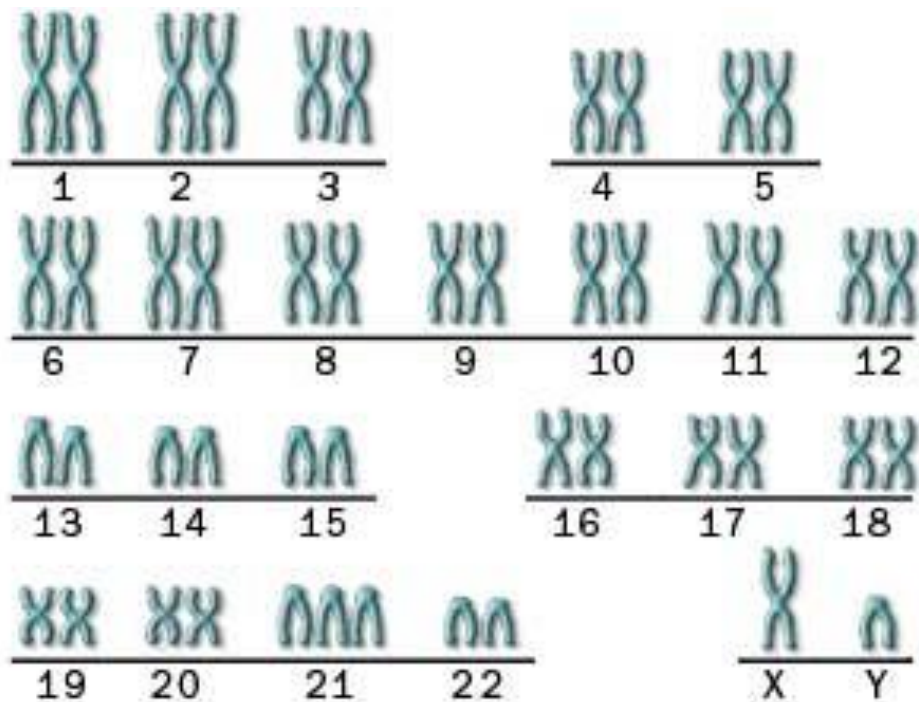
• **Adjustment of the dose :**

Clinical

Laboratory

• **Prognosis**

Genetics



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➤ **Case (1) :**

- You asked to see a baby on the postnatal ward as he is **floppy**.
- You find that he has a **flat mid-face** and a **flat back to the head, epicanthal folds, single palmar creases bilaterally**, a widened **gap** between his first and second toes and a **systolic heart murmur**. He is **feeding** normally.



What is the most likely diagnosis?

Comment :

- In a floppy baby without dysmorphic features, the most likely causes would be sepsis and hypoglycemia.
- The feature you noted indicate a **syndromic diagnosis** and are consistent with those of Down syndrome.
- Neonatal hypotonia is an almost universal feature of Down syndrome.
- Down syndrome, is associated with cardiac anomalies (40-50%).

➤ **Case (2) :**

- A couple come to see you with their second son, Sameh, who has Down syndrome. Their first son, Ahmed, is unaffected.
- They tell you that there is a **strong family history** of Down syndrome, with one of Sameh's cousins also being affected and his uncle is also being affected.

Which is the most likely cause of the Down syndrome in this family ?

The most likely cause here is **Robertsonian translocation involving Chromosome 21**.

- The previous mother got pregnant (8 weeks) and she is worried about this baby also to have Down syndrome

What tests would you do to this pregnant mother ?

- Measuring biochemical markers in maternal blood samples (**Triple test**).
↓ AFP ↓ UE3 ↑ hCG
- Fetal ultrasound to identify nuchal thickening

When an increased risk is identified :

→ Amniocentesis (16-20 wks' gestation) to check the fetal chromosome pattern.

→ Chorionic villus sampling (10-12 wks' gestation)

N.B. In patients with non-disjunction or mosaic Down syndrome, there is no need to investigate parents or offspring

► **Case (3) :**

- A newborn infant is noted to have **dysmorphic** features.
- The pregnancy was complicated by breech presentation, decreased fetal movements, and **polyhydramnios**.
- The baby demonstrates **hypotonia**, a **flat face**, flattened occiput, **epicanthal** folds, **brushfield spots** on the irides and **abdominal distention**.

To evaluate the abdominal distention, an x-ray study of the kidneys, ureters, and bladder is performed and reveals a "double-bubble" sign.

The best explanation for the neonate's abdominal distention is:

- a- Hirschsprung disease
- b- Meconium ileus
- c- Meconium plug
- d- Duodenal atresia

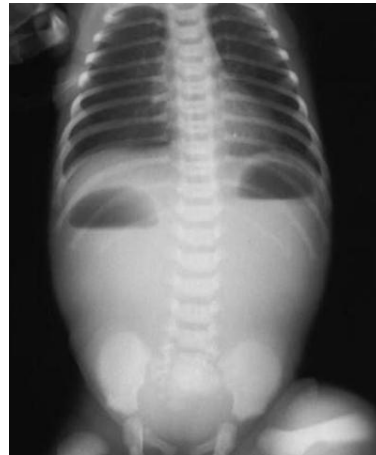
Answer : **d- Duodenal atresia**

Is it possible to find simian crease in a normal newborn ?

A single transverse palmar crease is present in 5% and bilateral palmar creases in 1% of normal newborns.

Are Brushfield spots pathognomonic for Down syndrome ?

They are seen in 75% of patients of Down syndrome.
Also in up to 7% of normal newborns.



Which of the following are true regarding Down syndrome ?

- a- It can cause hearing difficulties. ()
- b- It has an increased risk of leukemia. ()
- c- It is associated with an increased carrying angle. ()
- d- It is associated with hyperthyroidism. ()
- e- Atlanto-axial instability on cervical spine X-ray. ()
- f- Echocardiogram is mandatory even if no murmur. ()
- g- Cardiac anomalies are present in 60%. ()
- h- The commonest cardiac lesion is coarctation of the aorta. ()
- i- Tall stature is a feature. ()
- j- Hypertonia in the neonatal period is observed. ()

Answer : a- ✓ b- ✓ c- ✗ d- ✗ e- ✓
 f- ✓ g- ✗ h- ✗ i- ✗ j- ✗

Down syndrome shows an increased risk of occurrence compared to the general population of :

- a- Leukemia. ()
- b- Imperforate anus. ()
- c- Alzheimer's disease. ()
- d- Duodenal atresia. ()

Answer : a- ✓ b- ✗ c- ✓ d- ✓

Comment :

- Down syndrome is associated with an increased incidence of leukemia, Alzheimer's disease, duodenal atresia and atlanto-axial dislocation.
- It is also associated with an increased risk of CHD (about 40-50%). Defects include AV canal (most common) and VSD.
- They also are at risk of esophageal atresia, Hirschsprung's disease, **pulmonary hypertension** and cataracts.

➤ **Case (4) :**

- An 8 month-old girl presents with a history of **atrial septal defect** and **duplex kidneys**. A cranial ultrasound shows multiple choroid plexus cysts.
- On exam., you find a **small** baby with **hypertonia**, **micrognathia**, low-set ears and a prominent forehead and occiput. She has **overlapping fingers**. Her **rocker-bottom feet** show no evidence of lymphedema.
- Cardiac exam. reveals a short sternum with a 2/6 pansystolic murmur at the left sternal edge radiating to the apex.

Which is the possible diagnosis in this case ?

Edwards' syndrome (trisomy 18)

[Patau's Syndrome]

Characteristic features of Patau's syndrome, include which of the following ?

- a- Scalp defects. ()
- b- Polydactyly. ()
- c- Holoprosencephaly. ()
- d- Early death in infancy. ()

Answer : a- ✓ b- ✓ c- ✓ d- ✓

Clinical features of Edwards' syndrome (trisomy 18)

- Low birthweight
- Prominent occiput
- Small mouth and chin
- Short sternum
- Flexed, overlapping fingers
- Rocker-bottom feet
- Cardiac and renal malformations

Clinical features of Patau's syndrome (trisomy 13)

- Structural defect of brain
- Scalp defects
- Small eyes (microphthalmia) and other eye defects
- Cleft lip and palate
- Polydactyly
- Cardiac and renal malformations

➤ **Essentials for diagnosis of Patau's syndrome are:**
microcephaly, microphthalmia, clefting

N.B. Patients with trisomy 18 are hypertonic with scissoring, while patients with Down syndrome and trisomy 13 are hypotonic

➤ **Case (5) :**

- A **15-year-old girl** presents with **hypertension**.
- On examination she is **pre-pubertal**, has a **webbed neck**, and **widely-spaced nipples**.
- She has **weak femoral pulses**, and auscultation reveals a **systolic murmur** at the left upper sternal edge and infraclavicular region.

What is the most likely diagnosis for this clinical finding ?

Comment :

- The clinical features are suggestive of coarctation of the aorta.
- **Turner's syndrome** is associated with aortic coarctation.
- Other features include : short stature, widely-spaced nipples, webbed neck, streak ovaries.

Clinical features of Turner's syndrome

- Lymphoedema of hands and feet in neonate, which may persist
- Spoon-shaped nails
- Short stature - cardinal feature
- Neck webbing
- Wide carrying angle (cubitus valgus)
- Widely spaced nipples
- Congenital heart defects (particularly coarctation of the aorta)
- Delayed puberty
- Ovarian dysgenesis resulting in infertility
- Hypothyroidism
- Renal anomalies
- Pigmented moles
- Recurrent otitis media
- Normal intellectual function in most

Short stature may be the only clinical abnormality in children.

► Case (6) :

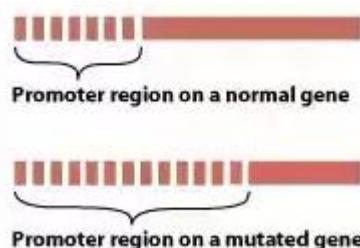
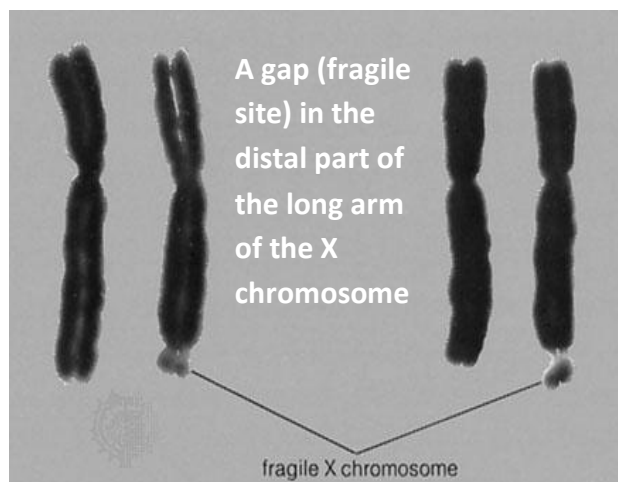
- You asked to investigate a **5-year-old** boy who has **autistic** features.
- His occipito frontal circumference (**OFC**) is on the **98th centil**, but height and weight are on the 50th centile. He also has **large ears**.
- Karyotype was **normal**.
- His **mother** suffered a **premature menopause** aged 36 years.

What is the most likely diagnosis ?

Fragile X Syndrome

- The normal copy of the gene contains **<50** copies of the CGG trinucleotide repeat sequence and is **stable** when transmitted to offspring.
- Pre-mutation: genes contain 55-199 copies of the repeat sequence; this expansion
 - Causes no intellectual disability
 - Is unstable and may become larger during transmission through females.
- Full mutation: genes contain **>200** copies of the repeat sequence. This affects gene function, causing the **clinical features** in all males and around half of the female carriers.

The full mutations always arise from expansion of pre-mutations, and never arise directly from normal genes.



- Diagnosis is now achieved by molecular analysis of the CGG trinucleotide repeat expansion in the relevant gene (**FMR1**)

N.B. Fragile X is NOT a single gene inheritance it is a molecular disease

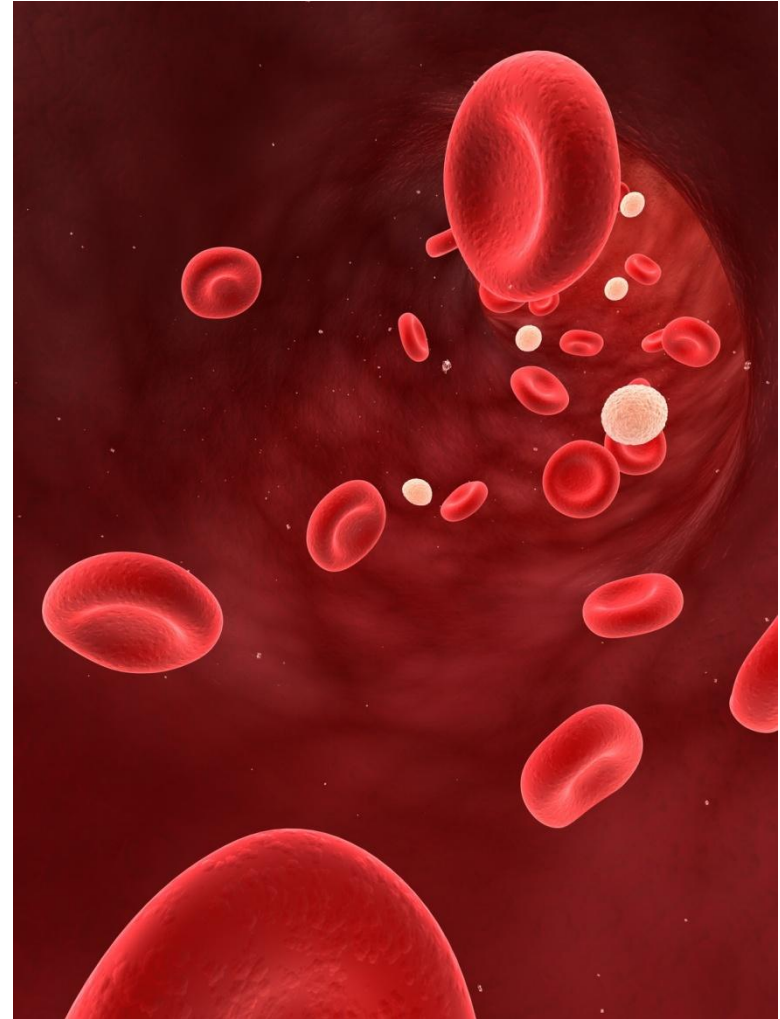
► Fragile X Syndrome in Males

- Mild, moderate, or severe mental retardation (according to the size of the mutation)
- Macrocephaly
- Macro-orchidism - **postpubertal**
- Characteristic facies: long face, **large everted ears**, prominent mandible (**mandibular prognathism**) and broad forehead, most evident in affected **adults**.
- Other features: mitral valve prolapse, joint laxity, scoliosis, **autism**, hyperactivity
- **Fragile X syndrome is the second most common genetic cause of mental retardation after Down syndrome**, it is often more serious than Down syndrome because once you diagnose a case, you will find other cases in the family.
- Fragile X syndrome should be suspected in any retarded male.

► Fragile X Syndrome in Females

- Females with mutated gene on one X chromosome are mostly asymptomatic carriers.
- Around 50% of females who are full mutation carriers may develop mental retardation.
- Female carries of premutations are at a higher risk of premature ovarian failure and learning difficulties in mathematics.

Hematology



► Case (1) :

* History :

- A **6 year** old girl, living in **Oasis**, who commonly presents to ER, presents with **pain** in her back and limbs.
- Her pain started this morning and has been worsening throughout the day, despite taking paracetamol and ibuprofen
- This was preceded by URI symptoms for 2 days, and **fever** reaching 38.9°C
- The URI symptoms consist of a stuffy nose, and a dry cough.
- Oral intake has been **decreased**, but adequate.
- She also takes **daily prophylactic amoxicillin**.
- The patient has been admitted to hospital three times in the last 3 months with **similar attacks**.
- The mother says that, at the age of 1.5yr, her child developed **swelling of the fingers and toes** that resolved within few days after analgesics in the form of NSAID.
- The patient has received **blood transfusion** twice over the last 3 years.
- Family history of **positive consanguinity** and a first degree relative with a **similar condition**.

What is the Differential Diagnosis ?

► Differential Diagnosis of Joint Pain :

- **Infections** : Bacterial, Viral, Reactive, Rheumatic fever
- **Inflammatory**
- **Vasculitis**
- **Hematological Disorders** : Sickle cell disease–Hemophilia
- **Malignant Diseases** : Leukemia-Neuroblastoma
- **Connective Tissue Disorders** : JIA-SLE

* Examination :

- Vital signs: Temperature:39.0°C, HR: 105 bpm, RR: 30, BP 98/55.
- Nontoxic appearing, mild pallor, slight scleral icterus, mild clear white nasal discharge and non-congested pharynx.
- She is in obvious pain, and is crying , however, there are no joint swellings, hotness, redness or tenderness and she has full range of motion of all her joints.
- Her lungs are clear to auscultation. Her heart is regular without murmurs.
- Her abdomen is soft and non-tender to palpation. Her spleen is not palpated below the left costal margin, and her liver is palpated 2 cm below the right costal margin.
- There are no rashes or skin lesions or lymphadenopathy.

Q1:What is the most probable diagnosis of this case ?

- The long duration and absence of physical findings makes malignant disorders unlikely
- The positive family history, recurrence of attacks and blood transfusion make infections unlikely
- The patient's age and pattern of joint affection make CT disorders unlikely.
- Considering the patient's history and findings by examination, **sickle cell anemia** is the most probable diagnosis.

Q2: What is the key investigation to diagnose the case ?

The diagnostic investigation is Hb electrophoresis :

- SS: HbS-HbF-No HbA
- S/β: HbS-HbF-elevated HbA2

Q3: What other investigations should be performed for any febrile sickle cell disease case ?

- Blood count, band count, and reticulocyte count
- Blood culture - Chest radiograph
- Urinalysis, culture, and sensitivity
- Mycoplasma titer
- Stool culture (if diarrhea present)
- Evaluation for osteomyelitis

All patients under age 5 with a documented temperature (by mouth) above 38.5°C should be admitted to the hospital

Q4: What is the best approach for a febrile child with sickle cell disease ?

- a- CBC, BC, oral hydration, IM or oral antibiotics if source of infection is noted on PE.
- b- CBC, BC, IM ceftriaxone
- c- CBC, BC, admit for IV hydration and IV antibiotics.
- d- CBC, BC, no oral antibiotics if no specific source of infection is noted on PE

Answer : c- CBC, BC, admit for IV hydration and IV antibiotics.

Q5: If CBC shows a hemoglobin of 7.9 g/dl and WBC 17.8, Appropriate initial management includes:

- a- IV hydration if oral intake is insufficient, IV or PO pain management as needed.
- b- IV hydration, IV analgesia by continuous infusion
- c- IV hydration, IM meperidine prn.
- d- IV hydration, transfusion of PRBC, IV narcotic q 4 hours prn.

Answer : b- IV hydration, IV analgesia by continuous infusion

► Case (2) :

*** History :**

- Six months later, the patient presented to ER with severe **chest pain**, difficulty to breath deeply and pain was **worse with inspiration**.
- PE: temperature was **38.8°C**, her heart rate was 120 bpm, BP 125/80 mmHg, respiratory rate 40 breaths/min, and oxygen saturation 91% in air.
- There were **bronchial breath sounds** at both lung bases. Heart sounds are normal.
- Her **chest radiograph** was:

Q6: What is the most likely cause of the patient's chest pain?

Acute sickle chest syndrome

- Thrombosis, infection and fat embolism to the lung produce a syndrome of pleuritic chest pain, shortness of breath and fever.
- It often evolves from the lung bases and produces consolidation, which may be clinically apparent before radiographic changes appear.
- Hypoxia is a frequent feature, and failure to recognize and manage this syndrome aggressively may lead to a rapid deterioration.
- Difficult to differentiate from pneumonia

Q7: How should this be managed ?

- Empiric antibiotics: add macrolides
- Analgesics
- Bronchodilator & chest physiotherapy
- Ventilatory support
- Hydration
- Oxygen therapy
- Exchange transfusion
- Hydroxyurea



➤ Case (3) :

- Salma is a 5 year old girl from aswan.
- Her mother came complaining of pallor with repeated blood transfusion since the age of 6 months
- Also she complained of jaundice and progressive abdominal distension
- NO history of purpura or bleeding from any orifice
- No history of prolonged fever or recurrent infections
- Normal nutritional history

• On examination :

- HR :120/min with large pulse volume - BP 95/45
- Height and weight below the 5th percentile
- Mongoloid facies with frontal bossing and prominent maxillae and widely separated central incisors
- Jaundice - Pallor - Short broad hands
- Short systolic murmur over Lt parasternal area
- Hepatosplenomegaly
- No purpura or generalized Lymphadenopathy

➤ Differential Diagnosis :

- Hemolytic anemia is the first possibility because of pallor , jaundice, HSM and mongoloid facies
- **Thalassemia** is the first possibility because it is the commonest hemolytic anemia and also because no history suggestive of sickle crisis
- Iron deficiency anemia unlikely because of normal nutritional history and repeated monthly blood transfusion
- Bone marrow aplasia unlikely because no other features of pancytopenia (purpura , fever or recurrent infections) throughout this period of 2.5 years.
- Malignancy unlikely because of longstanding history, no other features of pancytopenia, and no generalized lymphadenopathy

➤ Case (4) :

* History :

- A 3-years old girl presented to the emergency room with high fever, fatigue, petechial rash and gum bleeding.
- There was no remarkable past medical or family history.

* Physical examination :

- Blood pressure 60/40 • Heart rate 120
- Respiratory rate 32 • Temperature 40.5 C • Chest x-ray was normal
- Abdominal examination was normal with no organomegaly or Lymphadenopathy

* CBC :

- Hb – 8.9 g/dL • RBC – $3.0 \times 10^{12}/L$ • MCV – 103 fL
- Plt – $10 \times 10^9/L$ • WBC – $0.6 \times 10^9/L$
- Neuts – $0.4 \times 10^9/L$ • Lymphs – $2.3 \times 10^9/L$
- Reticulocytic count-1 %

Summary :

- Peripheral blood film : Pancytopenia, Macrocytosis & Reticulocytopenia
- Bone marrow biopsy
- Hypocellular with fat replacement

➤ Differential Diagnosis of Pancytopenia :

• Inherited :

- Fanconi's Anemia

• Acquired :

- Idiopathic (majority)-2/3rd of cases
- Drug : Acetazolamide, Carbamazepine, Gold, Hydantoin, Penicillin, Phenylbutazone.
- Chemical • Radiation exposure • Viral illness

➤ Other causes of Pancytopenia :

- Megaloblastic anemia
- Bone Marrow infiltration or Replacement: Lymphoma, Myeloma, Acute Leukemia, Secondaries
- Hypersplenism
- SLE • Disseminated TB • PNH • Sepsis

➤ Case (5) :

* History :

- A 7 year -old boy presents to the outpatient clinic complaining of shortness of breath and bruises on his arms. The bruises appeared 6 months previously and were attributed to minor trauma and the shortness of breath began 3 months earlier but worsened recently.
- In the last several days, gum bleeding was noticed on brushing his teeth.
- There have been no other accompanying symptoms.

* Clinical Examination :

- Blood pressure was 100/70 mmHg
- Heart rate 92 per min
- Respiratory rate 20 bpm
- Temperature 36.8C
- Height and weight were at the fifth percentile
- The heart and lung examination were normal
- There were no hepatosplenomegaly or Lymphadenopathy
- A faint tan to grey hyperpigmented eruption was noted over the upper trunk. Bruises on both upper extremities were noted along with some mild gum bleeding and abnormal thumbs.

* CBC :

- Hb 8.9 g/dL • RBC $3.0 \times 10^{12}/L$ • MCV 103 fL
- Plt $18 \times 10^9/L$ • WBC $2.2 \times 10^9/L$
- Neuts $0.2 \times 10^9/L$ • Lymphs $2.3 \times 10^9/L$
- Retix 0.5%

What is the investigation of choice ?

This is a case of **Pancytopenia**

➤ Case (6) :

* History :

- 1.5 years old male was referred to the hematology clinic with a chief complaint of easy bruising. He has not had epistaxis, oral bleeding, gross blood in urine or stools. He had history of palpable bruises and excessive post circumcision bleeding in the past.
- He has 1 older brother and 1 sister, neither of whom has had bleeding symptoms. But Family history is positive for frequent nosebleeds and menorrhagia.

What is the differential diagnosis of bleeding tendency in early infancy ?

➤ Bleeding Tendency in early infancy :

- Child physical abuse
- Haemophilia A
- Haemophilia B
- Idiopathic thrombocytopaenic purpura
- Congenital afibrinogenaemia
- Vitamin K deficiency
- Von Willebrands disease

* Examination :

- VS are normal. Height and weight are at the 50th percentile. He is a healthy appearing, cooperative male in no acute distress.
- HEENT exam demonstrates no signs of bleeding or bruising. Heart and lung examination are normal. His abdomen demonstrates no hepatosplenomegaly.
- palpable ecchymoses of varying ages were noted on his extremities. His neurologic examination demonstrates no deficits.

What is the diagnosis of this case ?

- Bruising with or without preceding trauma can be due to a defect in either primary or secondary hemostasis although deep palpable bruises are usually due to a clotting factor defect.
- The commonest causes of bleeding in early infancy are hemophilia A and B.
- Child's age, sex & the palpable bruises make **hemophilia** the most likely diagnosis.

➤ **Case (7) :**

- 4 year old female is referred to the Hematology department complaining of acute onset of easy bruising ,subconjunctival Hg and "rash" for 3 days.
- She had upper respiratory infection symptoms approximately 2 weeks ago.

*** History :**

- No history:
 - epistaxis, oral bleeding, gross blood in urine or stools.
 - hemarthrosis or deep muscle bleeds in the past.
- She has 2 older brothers, neither of whom have had bleeding symptoms.
- Family history is negative for any bleeding disorders.

What is the differential diagnosis of Purpura ?

➤ **Purpura :**

• Thrombocytopenic :

- ITP - Bone marrow failure - DIC
- Immune disorders (primary ,secondary)

• Non-Thrombocytopenic :

- Vascular - Drugs
- Sepsis - Congenital

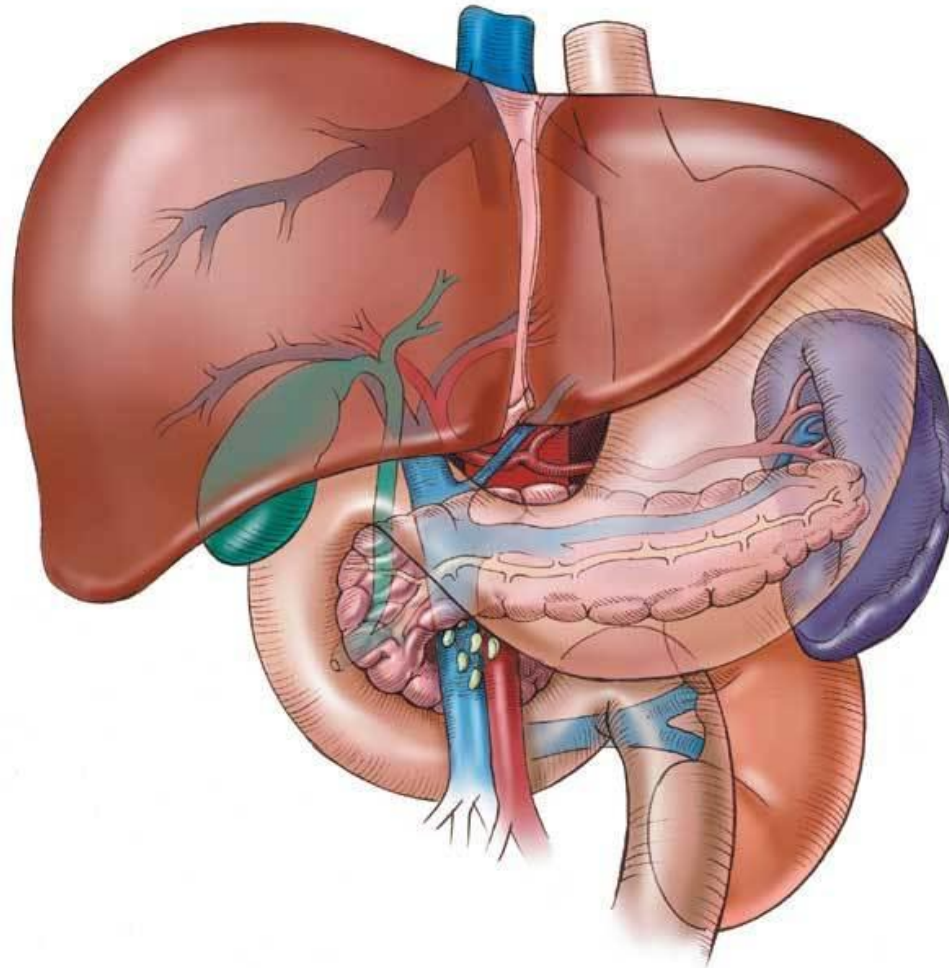
*** Examination :**

- She is a healthy appearing, cooperative girl in no acute distress.
- Height and weight are at the 50th percentile.
- HEENT exam demonstrates no signs of bleeding .
- Heart and lung exams are normal.
- Her neurological examination demonstrates no deficits.
- Her abdomen demonstrates no hepatosplenomegaly.
- A diffuse petechial rash is noted on her neck, trunk, extremities & groin.
- Nonpalpable ecchymosis of varying ages are present on chins, arms and trunk.
- Subconjunctival Hg. are seen.

What is the most likely diagnosis of this case ?

- Normal physical examination apart from **purpura** and **echymosis**.
- Make the most likely diagnosis is **Immune thrombocytopenic purpura**.

Hepatology



► Case (1) :

* History :

- Mahmoud is a **5-year-old** boy referred to the pediatric day unit by his GP.
- He was seen in the previous day with diarrhea and vomiting and seemed to have tummy ache.
- The vomit contained a small amount of **fresh blood**, but as he was otherwise well and cardiovascularly **stable**.
- Admission had been deferred because his mother is 36 weeks into her fourth pregnancy.
- Instead, later that day, the GP rang to check how he was and was reassured to hear that he seemed to have recovered and was tolerating drinks and some food.
- This morning he seems lethargic and a finger-prick blood glucose test performed by his GP was only 40 mg/dL.
- Mahmoud is the youngest of three children, the older two being 9 and 7 years old. His father is out of the country.

* Examination :

- He looks unwell.
- His airway is patent, his RR is 26 breaths/min and his **pulse rate is 180 beats/min** with a **capillary refill time of 5 seconds**. His **BP is 80/40 mmHg**.
- He is **jaundiced**.
- Both heart sounds are present and normal.
- Chest and abdominal examination are normal.
- He is **drowsy** but knows his mother and responds to her voice.
- He resists examination and withdraws to pain.
- There is **no** meningism and there are **no** focal neurological signs.

* Investigations :



INVESTIGATIONS

		Normal
Haemoglobin	12.3g/dL	11.5–15.5 g/dL
White cell count	$8.4 \times 10^9/L$	$6.0 - 17.5 \times 10^9/L$
Platelets	$140 \times 10^9/L$	$150 - 400 \times 10^9/L$
Prothrombin time	19 s	11–15 s
Partial thromboplastin time	32 s	25–35 s
Sodium	138 mmol/L	138–146 mmol/L
Potassium	3.6 mmol/L	3.5–5.0 mmol/L
Urea	8.2 mmol/L	1.8–6.4 mmol/L
Creatinine	33 μ mol/L	27–62 μ mol/L
Glucose	2.9 mmol/L	3.3–5.5 mmol/L
Bilirubin	85 mmol/L	2–26 mmol/L
Alanine aminotransferase (ALT)	1875 IU/L	5–45 IU/L
Alkaline phosphatase	2624 IU/L	145–420 IU/L
Albumin	32 g/L	39–50 g/L
C-reactive protein	<6 mg/L	<6 mg/L
Lactate	3.2	0.8–1.5 mmol/L
Venous blood gas on 15 L/min of oxygen		
pH	7.28	7.35–7.45
PCO ₂	3.8 kPa	4.5–6.0 kPa
Bicarbonate	17 mmol/L	22–29 mmol/L

What pathological processes are evident from his clinical signs and investigations ?

What is the most likely cause ?

How is this condition managed ?

Discussion :

- First: **Acute liver dysfunction**; evidenced by:
 - Prolonged PT, hypoglycemia and low albumin
 - Liver inflammation with $\uparrow\uparrow$ ALT & Bilirubin
- Second: **Tissue underperfusion** and anaerobic metabolism; evidenced by:
 - Tachycardia, borderline hypotension and a prolonged capillary refill time.
 - Metabolic acidosis and a high lactate with a compensatory reduction in the PCO₂.

- Third: **Mild encephalopathy**, although his drowsiness could be due to hypoglycemia.
 - Finally: **↑ Blood urea** (could be due to dehydration but his diarrhea and vomiting were resolving and the creatinine is normal). It is more likely to be due to the digestion of blood following upper GIT bleeding → ask about melena stools.
 - ➔ The most likely cause is a **fulminant hepatitis**.
- Fulminant hepatitis A in endemic countries and non-A to G in non endemic.
- This history is also classical for some form of poisoning.
 - Hepatotoxicity due to many drugs can cause fulminant liver failure.

Malnutrition & Diarrhea



► Case (1) :

* History :

- Ahmed is a **7-month-old** infant who is referred to the pediatric referral clinic with a 2-day history of **diarrhea** with **blood & mucus** in the stool.
- His mother says that he has periods of **inconsolable crying** which are getting **worse** and more **frequent**.
- In the last few hours, he had started to **vomit** and the last vomit was **bile-stained**.
- No history of:
 - Contact with gastroenteritis, Travel, or Bleeding disorders
 - He had neonatal meningitis & subsequently developed epilepsy (treated with sodium valproate). He has had no fits in the past month.
 - His mother is in good health but his father has type 1 diabetes.

Q1: What is the differential diagnosis of rectal bleeding ?

- | | |
|-------------------------|------------------------------|
| • Gastroenteritis | • Anal fissure |
| • Intussusception | • Cow's milk protein allergy |
| • Meckel's Diverticulum | • IBD |
| • Polyp | • Clotting abnormality |
| • Sexual abuse | |

* Examination :

- His temp. is **37.9°C**, pulse rate is **186 beats/min**, BP is 80/44 mmHg & **capillary refill is 4 sec**.

He is **difficult to examine** due to frequent **crying**.

When examined during a quiet period, a **mass** is felt on the **right** side of the abdomen.

The anus appears normal and no other signs.

Q2: What is the diagnosis of this case ?

- The commonest causes of passing blood per rectum are gastroenteritis and an anal fissure.
- Gastroenteritis is a possibility, but somewhat less likely in this case
- Anal fissure unlikely
- Child's age, rectal bleeding & the mass make an **intussusception** the most likely diagnosis.

* Investigations of Our Case :

- Hemoglobin 12.8 g/dl (N: 10.5–13.5 g/dl)
- White cell count $7 \times 10^3/\mu\text{L}$ (N: 4.0–11.0 $\times 10^3/\mu\text{L}$)
- **Platelets $457 \times 10^3/\mu\text{L}$** (N: 150–400 $\times 10^3/\mu\text{L}$)
- Sodium 138 mEq/L (N: 135–145 mEq/L)
- Potassium 3.9 mEq/L (N: 3.5–5.0 mEq/L)
- **Urea 25 mmol/L** (N: 5–17 mmol/L)
- Creatinine 0.6 $\mu\text{mol/L}$ (N: 0.2–0.9 $\mu\text{mol/L}$)
- C-reactive protein 12 mg/L (N: 6 mg/L)

Q3: What is the key investigation ?

The diagnostic investigation is an **ultrasound**.

Q4: How to treat this case ?

- Initial treatment:
 - IV fluid resuscitation
 - IV antibiotics (penicillin, gentamicin & flagyl)
- Nasogastric tube insertion & stomach emptying.
- Reduction by air enema (rectal air insufflation) (success rate 75%).
- If fails, or if the child is unstable or has signs of peritonitis or a perforation
→ surgery.

Q5: What is the recurrence rate ?

- Recurrence in <5%
- More frequent after rectal air insufflations

➤ Case (2) :

* History :

• A mother brings her 1-year old daughter because she is **always crying**, she observed that her child is **wasted** & looks **skin over bone**.

Q1: What is the differential diagnosis of wasted child ?

- Nutritional marasmus
- Congenital anomalies
- CHD & renal anomalies
- CHPS (in young infants)
- Chronic infection: empyema, TB & UTI
- Chronic illness: DM, cystic fibrosis, cerebral palsy & mental retardation
- Malignancy

• On asking the mother she says that she works as a servant, she is so **busy** and she leaves the baby with her older "**8 years old**" sister to feed her.

• Her sister says that the infant takes **all her food** in spite of that the baby is **always hungry**, and if the mother gives her any food, the baby **takes it all**.

Q2: What is the information you have got from the previous sentence ?

- His older sister (8 years old) usually eats his food
- ➔ The **infant receives a very small amount of food**

* Examination :

- The child is **miserable** and **cries** most of the time.
- She is very thin with **lost subcutaneous fat** over her abdomen, buttocks and thighs.
- Lips are **pale** with **macerated** mouth angles.
- Her weight is **6.5 kg**, and her temp. is **36.4° C**.
- She can sit but **cannot crawl or stand**.
- All her muscles (e.g. deltoid, biceps and triceps) are extremely **wasted**.

Q3: What is the most likely diagnosis ?

It is a case of **nutritional marasmus**

Q4: What is the degree of that marasmic child ?

Second degree marasmus

Q5: What are the possible complications of marasmus ?

- Infections
- Gastroenteritis & dehydration
- Chest infection
- Hypothermia
- Hypoglycemia

Q6: What are the possible lab. investigations for a case of marasmus ?

1) Findings seen in nutritional marasmus :

- Plasma proteins: within normal
- Blood glucose: low
- Blood urea nitrogen: low
- CBC : Anemia (iron deficiency) , Leucocytosis (infection)

2) For the etiology of non-nutritional (2ry) marasmus :

- Stool examination for:
 - Parasitic infestations
 - pH and disaccharides (lactase deficiency)
- Urine examination for:
 - Ketonuria (starvation)
 - Pus cells if UTI
 - Sugar for diabetes
- Plasma amino acid pattern: aminoacidopathies
- X- ray for: heart, chest & kidney

► Case (3) :

* History :

- A mother brings her 1-year old **plumpy edematous** child because he is not well.
- Lately, she observed that her infant **lost his appetite**, became **disinterested** in playing, and commonly cries without a known cause.

Q1: What is the differential diagnosis of generalized edema in young children ?

► Edema in a Young Child :

- Nutritional (KWO)
- Cardiac (heart failure)
- Renal (nephrotic syndrome)
- Protein losing enteropathy
- Allergic reaction

- On asking the mother she says that she have got **another baby** 3 weeks ago.
- In the last 2 months, she **stopped breastfeeding** of her older infant and started to give him food in the form of **mashed potatoes**, and **cooked rice** (usually with some added sugar because her infant refuses to take them with salt). The infant was also interested in eating bread & **boiled macaroni**, but these days he **refuses** most of that food.

Q2: What is meant by maternal deprivation syndrome?

The psychological effect of maternal neglect on the young infant

- ➔ This participates in the mentality changes which occur in such child

* Examination :

- The child is miserable & disinterested on playing
- Plumpy with sparse light colored hair
- Lips are pale with macerated mouth angles
- His weight is 8.5 kg and his temp. is 36.48°C.
- He can set but he cannot crawl or stand.
- Edema of both feet and dorsum of his hands.
- Abdominal examination :
 - Hepatomegaly
 - A reasonable amount of fat in the abdomen is observed.
- Skin is **dispigmented**, with cracks & maceration over the buttocks, groin & lower limbs.
- Deltoid, biceps & triceps muscles are extremely **wasted**.

Q3: What are the constant features of KWO ?

- **Edema** (Albumin – ↑ ADH – ↑ Aldosterone)
- **Mental changes** (apathy & misery)
- **Growth failure & growth retardation**
- **Disturbed muscle to fat ratio**

Q4: What are the variable features of KWO ?

- Hair changes
- Skin changes
- Hepatomegaly
- Anorexia, vomiting & diarrhea
- Anemia (dyshemopoietic - hemorrhagic)
- Vitamin deficiency (Vit. A, K, B complex)
- Mineral deficiency: iron, copper & zinc
- Infections (acquired immunodeficiency)

Q5: What are the possible complications of KWO ?

- Hypoglycemia
- Hypothermia
- Water and electrolyte imbalance
- Serious infections

Q6: What are the possible lab. findings in KWO?

- CBC: anemia, leucocytosis (infection)
- Plasma proteins
 - ↓ Total proteins
 - ↓ Albumin
 - ↓ α & β globulins but ↑ γ globulins
- Hypoglycemia
- ↓ K⁺, dilutional ↓ Na⁺ (but ↑ total body Na⁺), ↓ Mg

* Investigations of Our Case :

- Hb% 9.8 g /dl, total WBCs count 13500 with 62% PNLs & 10% of them is staph.
- Random blood glucose was 68 mg/dl,
- Serum albumen was 2 g/dl & total serum protein was 4.5 g/dl
- Serum sodium was 127 mEq/L while serum potassium was 3 mEq/L

Q7: How to manage a case of KWO ?

- Prevention of PEM
 - Breastfeeding promotion
 - Nutrition education of mothers
 - Nutrition assessment of infants and toddlers every visit to the health care unit

➤ Case (4) :

* History :

- A mother brings her one & half year old boy because he is **not able to walk**.

Q1: What are the causes of delayed walking ?

➤ Delayed Walking :

- Rickets • PEM (marasmus & KWO)
- Bone abnormalities e.g. osteogenesis imperfecta
- Muscular disorders e.g. Duchenne muscular dystrophy
- Simple lack of training • Cerebral palsy
- Mental retardation • Hydrocephalus
- Down syndrome • Cretinism

- She also observed that her child has **frequent chest infection**.

Q2: Causes of recurrent chest infection in young children ?

➤ Recurrent Chest Infections :

- CHD with ↑↑ pulmonary blood flow • Bronchial asthma
- Foreign body inhalation • Cystic fibrosis
- Immunodeficiency • Frequent aspiration (e.g. GER & hiatus hernia)
- Hypotonia of respiratory muscles & chest wall deformity e.g. rickets

- On asking the mother she says that she was breastfeeding her child until the age of **9 mo**, then she started weaning.
- The mother states that her child was **not exposed to the sun**, as the sun never enter their house.

Q3: What are the predisposing factors for rickets ?

- Prolonged breastfeeding without vit. D supplementation
- Diet poor in vit. D (e.g. milk, fruits & vegetables)
- Lack of exposure to sunlight

Neonatology



► Case (1) :

*** History :**

- You are called to the operating room to manage an infant recently born by emergency cesarean delivery due to late fetal decelerations.
- The mother, an 18-year-old with one previous healthy child, received no prenatal care and arrived at the hospital approximately 1 hour prior to delivery.

*** Clinical Symptoms & Signs :**

- At delivery you found a large (4500 g), grayish-colored infant with poor tone, no spontaneous respirations, and a pulse of 100 beats/minute.
- His Apgar scores were 3 and 7 at 1' and 5' respectively. After 10' his Apgar score was 8.
- His anthropometric measures were all above the 90th percentile.
- The baby had hepatomegaly 4 cm below the costal margin.
- He had poor Moro and grasp reflexes.
- He was transferred to the NICU for further management

*** Investigations :**

- First random blood sugar (RBS) immediately after birth was 60 mg/dl.
- At 30 minutes postnatal RBS, was 35 mg/dl.
- His CBC showed a Hb of 19 gm/dl and a Ht of 70%.

Q1: What is the most likely diagnosis ?

Q2: What is the first step in the evaluation of this baby ?

Q3: What is the management of this baby?

[Infant of Diabetic Mother]

Considerations :

- Fetal hyperinsulinism is a response to **poorly controlled maternal hyperglycemia** resulting in fetal macrosomia and increased fetal oxygen requirements.
- These two factors can make the birth process difficult and result in neonatal distress.
- High infant insulin levels cause him to become hypoglycemic when he is removed from the high-sugar in utero environment and must be managed immediately to prevent further complications.
- A blood glucose level of 25 to 40 mg/dl requires immediate feeding. A level less than 25 mg/dl (or higher levels in symptomatic infants) is treated with intravenous glucose.
- Polycythemia, hypocalcemia, and hyper-bilirubinemia are other sequelae of gestational diabetes that may require management.

► Revise from Theoretical Book

➤ Case (2) :

*** History :**

- A mother is concerned that her 4-day-old son's face and chest are turning yellow.
- This infant was delivered vaginally after an uncomplicated term pregnancy.
- The family history is unremarkable.
- His physical examination is normal, with the exception of a large cephalhematoma.
- He is breast-feeding well and shows no signs of illness.

*** Examination :**

- The general and systemic examination revealed no abnormalities except for a yellowish discoloration of skin and mucous membranes involving the face, trunk, as well as the upper and the lower limbs.
- There is a moderate size cephalhematoma in the temporal region.

*** Investigations :**

- Serum bilirubin:
 - Total: 16 mg/dl. Direct: 0.2 mg/dl
- Hb=10gm/dl.
- Mother's blood group is O+
- Baby's blood group is A+
- Retic = 1 %

Q1: What is the most likely diagnosis ?

Q2: What are the investigations you would like to order ?

Q3: What is the best therapy ?

[Neonatal Jaundice]

Considerations :

- Neonatal hyperbilirubinemia results from higher rates of bilirubin production and a limited ability to excrete it.
- It includes physiologic jaundice and non physiologic jaundice.

Risk factors for development of severe hyperbilirubinemia in infants of 35 or more weeks' gestation (in approximate order of importance)

➤ Major Risk Factors :

- Pre-discharge TSB or TcB levels in the high-risk zone
- Jaundice observed in the first 24 hrs
- Blood group incompatibility with positive direct anti-globulin test, other known hemolytic disease (G6PD deficiency) elevated ETCOc
- Gestational age 35-36 weeks
- Previous sibling received phototherapy
- Cephalohematoma or significant bruising
- Exclusive breastfeeding, particularly if nursing is not going well and weight loss is excessive
- East Asian race

➤ Minor Risk Factors :

- Pre discharge TSB or TcB levels in the high intermediate-risk zone
- Gestational age 37-38 weeks
- Jaundice observed before discharge
- Previous sibling with jaundice
- Macrosomic infant of a diabetic mother
- Maternal age ≥25 years
- Male gender

➤ Revise from Theoretical Book

➤ **Case (3) :**

*** History :**

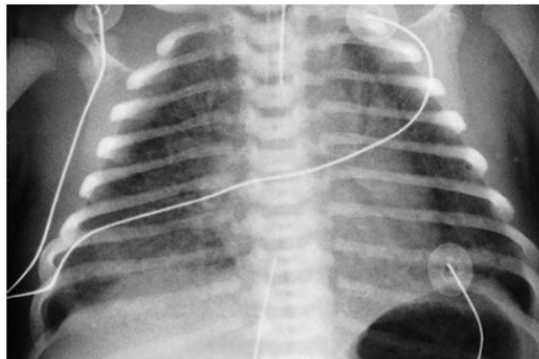
- A 2800-g male is born at 36-weeks' gestation to a 19-year-old mother via vaginal delivery.
- Delivery occurred 19 hours after membrane rupture.
- The mother's pregnancy was uncomplicated, but her prenatal records are not available at delivery.

*** Clinical Symptoms & Signs :**

- At 6 hours of age he is "breathing hard" and refusing to breast-feed.
- His respiratory rate is 60 breaths/min with "grunting".
- His temperature is 96.5°F (35.8°C), and his blood pressure is lower than normal.
- Upon arrival you confirm that he is in respiratory distress & that his perfusion is poor.

*** Investigations :**

- The CBC demonstrates a white blood cell (WBC) count of 2500 cells/mm³ with 80% bands.



Chest X-ray

Q1: What is the most likely diagnosis ?

Q2: What other investigations you would like to order ?

Q3: What is the best therapy ?

[Neonatal Sepsis]

Considerations :

- The rapid symptom onset, the low WBC count with left shift, and the chest x-ray findings are typical for GBS pneumonia.
- At this point, management would include rapid application of the ABCs of resuscitation (maintain Airway, control Breathing, and ensure adequate Circulation), followed by rapid institution of appropriate antibiotics once cultures are obtained.

N.B. Despite these measures, mortality from this infection is high.

➤ **Revise from Theoretical Book**

Nephrology



N.B. you have to check (UTI Case Scenario PowerPoint) ..

► **Case (1) :**

*** History :**

- A **7 year old male** presents to his primary care physician with the chief complaint of **dark "cola colored" urine, facial puffiness** and abdominal pain for the past 2 days.
- He had been in his usual state of good health until 14 days ago when he had a **sore throat** and **fever**. His sore throat and fever resolved. He was not seen by a physician at that time.
- There is no nausea or vomiting. His urine is dark brown and he **has not been voiding as much as usual**, only 2 times in the past 24 hrs.
- There is **no urinary frequency, urgency, dysuria or foul smell to the urine**.
- His appetite has been poor although he is still drinking fluids well.
- He is also complaining of **some back pain** in the flank area that he describes as a dull pain that comes and goes and does not seem to be related to activity.
- His **energy level is down** and he has not felt up to going to school for the past 2 days.
- He is also complaining of a dull generalized **headache** that has not been relieved with acetaminophen.
- **Review of systems is negative** for recent skin infection, skin rash, cough, rhinorrhea, seizure activity, fever, arthralgia or weight loss.
- His **past medical history, family history and social history are unremarkable**.

*** Examination :**

- VS T 37, P 100, RR 20, BP 120/75, oxygen saturation 100% in RA.
- Height and weight at 50th %tile.
- He is tired appearing but in no acute distress.

- Pupils are equal and reactive. Optic disc margins are sharp. Sclera are white and conjunctiva are clear. Mild periorbital is edema noted.
- TMs are normal.
- Throat, oral mucosa and nose are normal.
- Neck is supple without lymphadenopathy.
- Heart is regular without murmurs.
- Lungs are clear.
- Abdomen is diffusely tender (mild), without guarding or rebound. Bowel sounds are normal. No organomegaly is noted.
- His extremities are warm, with strong pulses. Capillary refill is less than 2 seconds.
- **No edema** is noted in his legs, feet or hands.
- **No skin rashes or impetigo** scars are noted.
- His genitalia are normal. **No scrotal edema** is present.
- Neurologic exam is normal.

*** Investigations :**

- His **urine** is tea colored. UA shows an increased specific gravity. A **dipstick** is positive for a large amount of blood and moderate protein. **RBCs are too numerous to count**. 5-10 WBCs per HPF. **RBC casts** are present.
- **CBC with diff is normal**.
- **Throat swab** is sent for culture.
- **ASO titer** is elevated.
- Serum complement **C3** level is low.
- Serum **electrolytes** are normal.
- **BUN 23 and Cr 0.8**.

* Clinical Course :

- He is diagnosed with **acute poststreptococcal glomerulonephritis**.
- He is initially **hospitalized** for treatment of oliguria/volume overload with furosemide, and monitoring of his modest hypertension.
- He has a **good urine output with the furosemide**, however he later requires a **calcium channel blocker** to control worsening hypertension.
- He is placed on a **fluid and sodium restricted diet**.
- His **throat culture later returns positive** for group A beta hemolytic streptococci (GABHS), so he is given a course of penicillin.
- He is **discharged** after 3 days of hospitalization.
- His **hypertension resolves** over the next 2 weeks.
- He is **followed closely** by his primary physician and his **proteinuria and gross hematuria resolve early**.
- His **C3 level normalizes** two months later.
- **Microscopic hematuria is expected to persist for months**.
- He **does not develop any long term complications**.

Q1: When does the C3 level return to normal in APSGN ?

C3 levels return to normal within a 6-8 week period in APSGN. Persistently low C3 levels suggest a cause other than APSGN.

Q2: What is the significance of finding red cell casts in the urine ?

The presence of red cell casts on urinalysis almost always indicates the presence of glomerulonephritis. They can also be seen after strenuous exercise and renal trauma.

Q3: What is the significance of finding white cell casts in the urine ?

The presence of white cell casts on urinalysis can be seen in APSGN, interstitial nephritis and pyelonephritis.

Q4: How long does hematuria persist in APSGN ?

Gross hematuria resolves within days to weeks. Microhematuria may persist for months.

Q5: Describe some indications for hospitalization of patients with APSGN:

An uncertain diagnosis, significant hypertension, anticipated poor follow-up, cardiovascular or cerebrovascular compromise, etc.

Q6: What are the clinical elements of the nephritic syndrome ?

Gross hematuria, oliguria, hypertension, edema (usually mild).

Q7: What are classic causes of the nephritic syndrome ?

APSGN and Goodpasture's. Other causes of nephritis include SLE nephritis, MPGN, RPGN, Alport's, etc.

Q8: A 5 year old boy has a screening urinalysis as part of a general physical exam. The UA shows microscopic hematuria. History suggests that he has impetigo periodically.

What is a likely cause for the microscopic hematuria ?

Convalescing APSGN.



➤ Case (2) :

* History :

- A **previously well 5 year old male** presented with the chief complaint of **facial puffiness**.
- His mother noticed this a few days ago and it seems to be worsening.
- He has no other symptoms, but about two weeks ago **had "a bad cold"**.

* Examination :

- VS T 37, HR 90, RR 20, BP 92/55.
- He is alert and cooperative with the examination.
- His face shows **moderate periorbital edema**.
- His eyes are non-injected, his conjunctiva are not edematous and his throat is not red.
- His heart is regular without murmurs. Heart sounds are normal.
- His lung exam shows good aeration, with no crackles or rhonchi.
- Abdomen is soft, non-tender, non-distended and without masses or shifting dullness. No hepatosplenomegaly.
- The **dorsal surfaces of his hands and feet have mild pitting edema**.
- He has brisk capillary refill.
- No rashes are noted.
- He has normal male genitalia with **no scrotal edema**.

* Investigations :

- Urinalysis shows 4+ protein, and a specific gravity of 1.030.
- His chemistry panel is remarkable for protein of 2 g/dL, serum albumin of 1.4 g/dL and cholesterol of 350 mg/dL.
- BUN and creatinine are normal.

* Management :

- He is not ill enough to require hospitalization.
- He is started on **oral prednisone TID**.
- He is followed as an **outpatient clinically** and by daily **urine dipsticks**.
- His **edema and proteinuria gradually resolve**.
- His **corticosteroids are tapered** off and he remains stable.

Q1: The most common cause of primary idiopathic nephrotic syndrome is:

- a- Focal segmental glomerular sclerosis
- b- Membranoproliferative glomerulonephritis
- c- Membranous glomerulopathy
- d- Minimal change disease

Answer : **d- Minimal change disease**

Comment :

- Minimal change disease or "nil disease" accounts for 80-85% of cases of primary idiopathic nephrotic syndrome in childhood.

Q2: Common causes of mortality in primary nephrotic syndrome is/are :

- a- Acute renal failure
- b- Thromboembolism
- c- Congestive heart failure
- d- Peritonitis
- e- Seizure

Answer : **b- Thromboembolism**
& d- Peritonitis

Comment :

- Infection, especially peritonitis and thrombosis account for the majority to nephrotic syndrome mortality.

Q3: True/False: A renal biopsy is necessary to confirm the diagnosis of primary idiopathic nephrotic syndrome.

Answer : False

Comment :

• The decision to perform a renal biopsy is usually deferred until the initial course of corticosteroid is initiated, unless there are specific risk factors such as age below one or above 10, hypertension on presentation or decreased complement on presentation.

Q4: The inheritance pattern of primary idiopathic nephrotic syndrome is/are :

- a- Autosomal recessive
- b- X-linked recessive
- c- Autosomal dominant
- d- Sporadic

Answer : d- Sporadic

Comment :

• Primary nephrotic syndrome is sporadic in nature. Congenital nephrotic syndrome is passed in an autosomal recessive manner.

Q5: Reasons for biopsy in a patient with nephrotic syndrome include :

- a- Continued proteinuria after a week of prednisone therapy.
- b- Age at onset of 10 months.
- c- Relapse 1 year after initial course of therapy.
- d- Cholesterol level greater than 400 mg/dL.
- e- A patient who has a history of systemic lupus erythematosus.

Answer : b- Age at onset of 10 months.

& e- A patient who has a history of systemic lupus erythematosus

Comment :

• Nephrotic syndrome in a child less than 1 year old may indicate congenital nephrotic syndrome and renal biopsy is often performed. In a patient with SLE, the nephrotic syndrome is likely secondary and a renal biopsy is indicated.

► Case (3) :

* History :

- This is a **4.5 year old** male who presents to the office with his mother with a chief complaint of **bed-wetting twice a week**.
- Essentially he is **healthy** except for an occasional cough and fever that the mother attributes to exposure to other children with colds.
- Involuntary urinary discharge occurs **at night only** and he therefore has to wear diapers to bed.
- His mother is worried since his **brothers and sisters were all toilet trained** by this age.
- There is **no history of dysuria, intermittent daytime wetness, polyuria, or polydipsia**.
- His past medical history is unremarkable.
- Family history is significant for his **father being a bed-wetter**. His child development is normal.

* Examination :

- VS T 37, P 110, R 20, BP 107/64, Ht 102 cm (25th percentile), Wt 16.2 kg (25th percentile).
- He is alert and active, in no distress. His appearance is non-toxic.
- HEENT and neck exams are negative.
- His lungs are clear bilaterally. His heart has a normal rate and rhythm, normal S1 and S2, and no murmurs or rubs.
- No masses, organomegaly, or tenderness are appreciated on exam of his abdomen. Bowel sounds are present. He has no inguinal hernias.
- He has a circumcised penis of normal size. The meatus is normally placed, without discharge. No phimosis is present. His testes are descended bilaterally and are of normal size (Tanner stage 1).
- His back is straight with normal posture with no scoliosis or tenderness, or midline defects.
- His extremities and muscle tone are normal. His gait is normal. He is able to hop, skip, and stand on each foot for 5 seconds, copy a square and get dressed without help.
- His speech and behavior are age appropriate.

* Management :

- You reassure his mother that bladder **control is usually attained between the ages of 1 and 5 years** and bed-wetting becomes less frequent with each passing year.
- You recommend that she **be supportive** of her son's dry nights and avoid criticism of wet nights.
- You also recommend **avoiding excessive fluid intake** two hours before bedtime and emptying his bladder at bedtime.

Q1: At what age do parents usually become concerned about bed-wetting ?

Typically at age 5 or 6 years.

Q2: True/False: Most nocturnal enuresis is due to organic causes.

False.

Q3: Which drug for nocturnal enuresis is cardiotoxic ?

Imipramine.

Q4: What laboratory test should be done to evaluate a child with enuresis ?

Urinalysis with specific gravity, glucose, protein, blood and white cells.

Q5: What is the bladder capacity of children ?

Most adults have a bladder capacity between 250-400 ml, but the average bladder capacity in children can be approximated by the formula: volume = (2 + age in years) x 28

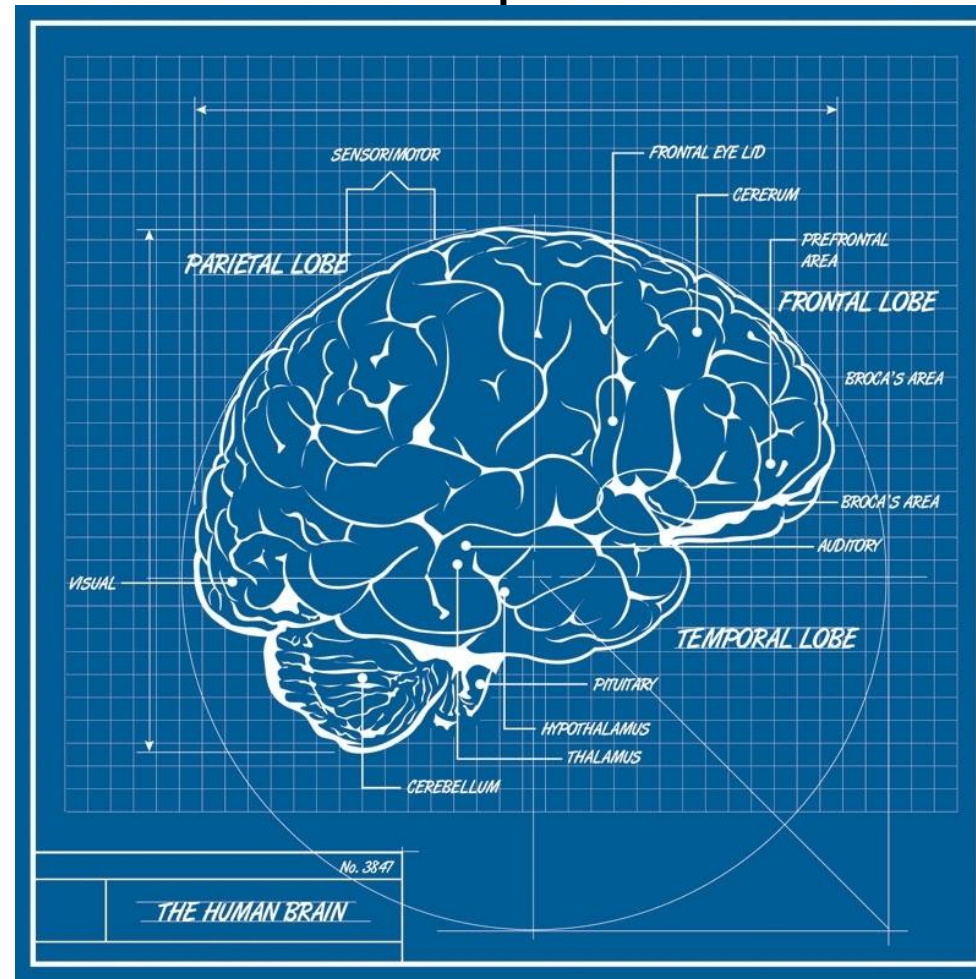
Q6: In evaluating a chronic bed-wetting child, what should you look for in an abdominal exam ?

The abdominal exam should assess for masses secondary to enlarged urinary organs (bladder, kidney) and for evidence of palpable stool in the colon suggesting fecal impaction.

Q7: True/False: Enuresis alarms produce excellent results if the child wakes up spontaneously when the alarm sounds.

True.

Neurology



➤ **Case (1) :**

*** History :**

- A **2-year-old** boy complained of fever and sore throat followed 2 weeks later by sudden refusal to stand or walk and by screaming on **touch**.
- First the **lower limbs** were affected followed by inability to **sit** from a supine position **paradoxical respiration** and **inability to swallow** with **hoarseness of voice**.

*** Examination :**

- Examination revealed a fully **conscious** patient generalized **hypotonia** and **hyporeflexia** involving both the trunkal muscles and the limbs .
- No bulbar palsy, ptosis or squint, no facial asymmetry.

The most probable diagnosis is :

- a- Myasthenia gravis
- b- Duchenne muscular dystrophy
- c- Acute post-infectious polyneuropathy
- d- Werdnig Hoffman disease

Answer : c- Acute post-infectious polyneuropathy

[Guillain-Barré syndrome]

- Acute post-infectious polyneuropathy
- Presentation is typically 2-3 weeks after an upper respiratory tract infection or campylobacter gastroenteritis.
- Fleeting abnormal sensory symptoms in legs (may be)
- **Ascending symmetrical** weakness with loss of reflexes and autonomic involvement.
- Sensory symptoms, usually in the distal limbs, are **less striking** than the paresis but can be **unpleasant**.
- Involvement of bulbar muscles → difficulty with chewing and swallowing & ↑ risk of aspiration.
- Respiratory depression may require MV.
- The maximum muscle weakness may occur only 2-4 weeks after the onset of illness.

N.B. Although full recovery may be expected in 95% of cases, this may take up to 2 years.

➤ **Causes of acute paralysis ?**

- **Spinal cord:** - Transverse myelitis (symmetric paraplegia) -Trauma to spinal cord.
- **Anterior horn cells:** Poliomyelitis (asymmetric, ascending).
- **Peripheral Nerves:** - Guillain Barre syndrome (symmetric and ascending) - Post-diphtheritic paralysis (symmetric and descending).
- **Neuromuscular:** Botulism (symmetric, descending).

1- Poliomyelitis :

- Acute paralysis :
- Acute, **asymmetric**, flaccid, massive, and purely motor. It starts in LL (one more than the other). It may extend to bulbar and/or respiratory muscles.
- Gradual :
- Incomplete recovery with residual weakness follows after few weeks.

2- Guillain-Barre syndrome :

- Bilateral and **symmetrical** paralysis in LL then **ascending** to UL.
- **Sensory** loss may be present but rare
- Course is usually prolonged.
- CSF: cytoalbuminous dissociation.
- EMG & NCV:
 - Demyelinating neuropathy mainly
 - Mixture of axonal & demyelinating neuropathy

3- Post - diphtheritic paralysis :

- History suggestive of diphtheria.
- Ocular and bulbar paralysis precedes limb paralysis.
- Not observed nowadays due to regular compulsory vaccinations.

4- Transverse myelitis :

- History Symmetric and flaccid paralysis of LL (paraplegia) with sensory loss (with a level)
- Later: features of pyramidal lesion.
- Upper limb is free.
- Incontinence to urine and stool is common.

5- Pseudoparalysis :

- Postinjectional, unrecognized trauma, osteomyelitis, myositis, toxic synovitis

Q1: What is the most reliable test(s) for diagnosis ?

- a- EMG and NCV
- b- CSF analysis
- c- Brain CT scan
- d- a & b

Answer : **d- a & b**

Q2: The CSF analysis will show :

- a- Normal cell number and increased protein
- b- Increased cells and increased protein
- c- Normal cell number and normal protein
- d- RBCs in the CSF

Answer : **a- Normal cell number and increased protein**

* Investigations of Our Case :

- CSF showed: - Cells: 2 cells/lymphocytes - Protein: 100mg/dl (20-40 mg)
 - Sugar: 60 mg/dl (concomitant blood sugar 90 mg/dl)
 - Culture: no growth
- EMG/NCV: delayed NCV, picture of severe demyelinating neuropathy
- The CSF protein is characteristically markedly raised, but this may not be seen until the second week of illness. The CSF white cell count is not raised (cytoalbuminous dissociation).
- Nerve conduction velocities are reduced.

Q3: The most appropriate treatment for the patient is :

- a- Corticosteroid
- b- Physiotherapy
- c- Intravenous immunoglobulin
- d- Bed rest

Answer : **c- Intravenous immunoglobulin**

► **Case (2) :**

*** History :**

- A 4-year-old male boy, the first child of nonconsanguineous parents, was referred to his pediatrician because the parents noticed that :
 - he walked later than older siblings
 - Difficulty running and riding tricycle
 - Always climbs up stairs on all fours
 - Frequent falling
- He was examined and reassured by his pediatrician and given some vitamins.
- At the age of 5 years, parents sought medical advice again as they noticed that their child has difficulty in rising from the sitting position and marked difficulty in climbing stairs and that the abdomen became protuberant during walking with somewhat waddling gait
- Chewing, swallowing, hearing and vision were all normal.
- Mentality and speech was normal.

*** Examination :**

- A child with average body built ,intelligent and interactive with the physician and well oriented
- Cranial nerve examination did not reveal ptosis or external ophthalmoplegia, facial asymmetry,
- Uvula was central and normal gag reflex,
- Tongue central without any observed fasciculations or atrophy.
- **Motor examination :**
 - By inspection: hypertrophy of the calf muscles, quadriceps, deltoid, infra and supraspinatus was observed in a bilateral and symmetrical fashion.
 - No abnormal movements were noticed during examination.
 - No muscular fasciculation either spontaneous or induced by stimulation.
 - No trophic ulcers observed at the heel or the tip of toes.

- Muscle power: weakness of both upper and lower limbs, proximal more than distal, noticed mainly in glutei
- Generalized hypotonia
- Hyporeflexia
- Positive Gower sign
- Gait : waddling gait
- Tests for coordination were all normal

Q1: What is the most likely diagnosis ?

- a- Spinal muscular atrophy
- b- Guillain barre syndrome
- c- Duchenne muscular dystrophy
- d- Becker muscular dystrophy

Answer : c- Duchenne muscular dystrophy

[Duchenne Muscular Dystrophy]

- Duchenne muscular dystrophy is the most common hereditary neuromuscular disease.
- Progressive muscle weakness.
- The disease process usually starts before 5 years.
- Achievement of the motor developmental milestones may be normal or slightly delayed.
- There is difficulty in climbing stairs and difficulty in rising from sitting position.
- Pseudohypertrophy of the calf muscles
- Fasciculation do not occur
- Mainly in a wheelchair by early teens
- Respiratory muscles eventually involved
- Death usually in late teens, early twenties from cardiorespiratory failure

Q2: What is the form of inheritance ?

- a- Autosomal dominant
 - b- Autosomal recessive
 - c- X linked recessive
-

Answer : c- X linked recessive

Comment :

- X linked recessive condition, hence males affected and females are carriers

Q3: What is the most diagnostic investigation ?

- a- EMG & NCV
 - b- CPK
 - c- Muscle biopsy
 - d- Molecular analysis for dystrophine gene deletion
-

Answer : c- Muscle biopsy

Comment :

- Muscle biopsy with immunohistochemical staining

► **Case (3) :**

- A 9-year-old boy is sent by his pediatrician for neurologic evaluation because of episodes of "confusion".
- The parents report that over the past year the child has experienced episodes during which he develops a fearful look on his face and responds slowly or not at all to their questions.
- Initially, the parents and pediatrician dismissed these as anxiety attacks
- Recently, the parents became more concerned when he developed chewing and swallowing movements during the attacks.
- The overt episodes last approximately 1-2 minutes. However, it appears to take several minutes before the boy recovers from the episodes.
- By the time the child is referred for neurologic work-up, he is suffering several attacks per week.
- The boy has little memory of these episodes, although he says he becomes quite frightened during the events.
- He reports that following the attacks he often has a headache and is fatigued. Although he is a good third-grade student,
- During the past few months he says he's had to work harder in his classes to maintain his grades.
- The boy's medical history reveals that at age 13 months he experienced 2 febrile seizures. These seizures, which were separated by 4 months, occurred early in the course of a viral-type illness with temperatures reaching 39°C and lasted approximately 2 minutes.
- The child was not prescribed any AEDs at that time.
- Family history is noncontributory for epilepsy or other neurologic conditions.

*** Examination :**

- His vital signs and general examination are unremarkable.
- His neurologic examination is likewise unremarkable:

- His cranial nerves are intact with no facial asymmetry or sensory deficit;
- Strength and reflexes are normal and symmetric, with bilateral flexor plantar response;
- Primary/cortical sensory modalities are intact; and coordination/gait are intact without truncal or appendicular dystaxia.

Q1: What is the most likely diagnosis in this patient ?

- a- Complex partial seizures
- b- Nonepileptic seizure (pseudoseizure)
- c- Absence seizures

Answer : a- Complex partial seizures

Comment :

- This child had discrete, paroxysmal events characterized by :
 - a change of facial expression,
 - impaired consciousness,
 - automatisms (chewing and swallowing), followed by postictal confusion.
- This makes **complex partial seizures** the most likely diagnosis.

Explanation :

- The term **partial seizures** refers to seizures that begin in 1 area or location of the brain.
- Partial seizures can be either **simple** (without impairment of consciousness) or **complex** (with impaired or altered consciousness).
- This child had discrete, paroxysmal events characterized by a change of facial expression, impaired (but not a total loss of) consciousness, and automatisms (chewing and swallowing), followed by postictal confusion.
- Automatisms, which refer to nonpurposeful motor activity, are common in complex partial seizures. The fear the child experiences likely is an aura that precedes the impairment of consciousness.

- **Nonepileptic seizures**, previously termed pseudoseizures or hysterical seizures, are unusual in children and typically mimic generalized tonic-clonic rather than complex partial seizures.
- The stereotyped nature of the attacks, the lack of precipitating stressful events, and the postictal period would make it unlikely this child has nonepileptic seizures.
- Absence seizures typically last less than 15 seconds, have a sudden onset and offset, and are not associated with an aura or postictal confusion.
- Automatisms can occur in both complex partial and absence seizures.
- Untreated children with absence seizures typically have many per day, and it would be unusual for the child to have only a few per week.

[Complex Partial Seizures]

- The consulting neurologist suspects these episodes are complex partial seizures.
- Complex partial (formerly called temporal lobe or psychomotor) seizures impair consciousness and occur in all age groups.
- Typically, staring is accompanied by impaired consciousness and recall. If asked a question, the patient might respond with an inappropriate or unintelligible answer, regardless of the question's content.
- Automatic movements (automatisms) are associated with most complex partial seizures and involve the mouth and face (lip smacking, chewing, tasting, and swallowing movements).
- Upper extremities (fumbling, picking, tapping, or claspings movements).
- Vocal apparatus (grunts or repetition of words or phrases), or
- More complex acts (such as walking or mixing foods in a bowl).
- Less commonly, automatisms include screaming, running, shouting, bizarre and sometimes "sexual"-appearing movements (pelvic thrusting or masturbation), and disrobing.

- Complex partial seizures usually last from 30 seconds to 3 minutes. Auras (simple partial seizures) typically precede the impairment of consciousness by seconds.
- The aura of fear, as seen in this child, frequently occurs. After the seizure, lethargy and confusion are common, but usually last less than 15 minutes.
- Patients typically have a normal neurologic examination, although subtle lateralizing signs, such as an asymmetric smile, may be present.

Q2: Is the history of febrile seizures of importance in this patient ?

a- Yes

b- No

Answer : **b- No**

Comment :

- The risk of developing epilepsy is only slightly higher among children who have suffered 1 or more febrile seizures as compared with those who have not.
- However, the link can be more significant depending on specific characteristics of the febrile seizure and subsequent chronic seizure type.
- Prolonged febrile seizures have been implicated as a major predisposing factor for the development of temporal-lobe epilepsy and mesial temporal sclerosis.

Q3: Which test would be most helpful in supporting the diagnosis of seizures (choose all that are correct) ?

a- Electroencephalogram (EEG)

b- Electrocardiogram (ECG)

c- Magnetic resonance imaging (MRI)

d- Computed tomography (CT)

Answer : **a- Electroencephalogram (EEG)**

Comment :

- Electroencephalogram (EEG) would be the most helpful first test to be ordered.
- MRI will also help to rule out structural causes for the seizure.
- In certain circumstances, ECG and CT scan might also be helpful in the later work-up of this patient.

• Initial Assessment :

- The EEG would be the most helpful test in supporting the diagnosis of seizures. The finding of epileptiform activity, defined as any paroxysmal discharges containing spikes or sharp waves, would support the diagnosis of seizures.

• Electroencephalogram (EEG) :

- Epileptiform activity on the EEG is rarely totally diagnostic of epilepsy. Only if the child has a seizure during the EEG could one conclude that the EEG confirmed the diagnosis of epilepsy.
- EEG abnormalities are not uncommon in normal children. In large studies of nonepileptic children, up to 9% will have epileptiform activity on EEG. Therefore, the EEG must be interpreted in conjunction with the clinical history.
- Likewise, a normal EEG does not rule out the possibility of epilepsy because patients with well-documented seizures may have normal EEGs between their seizures. This is particularly important to remember when dealing with partial seizures.

• Electrocardiogram (ECG) :

- An ECG would be useful if the clinician suspected a cardiac arrhythmia was responsible for the impaired consciousness.
- Disorders such as the prolonged QT syndrome would be more likely to mimic generalized tonic-clonic than complex partial seizures.

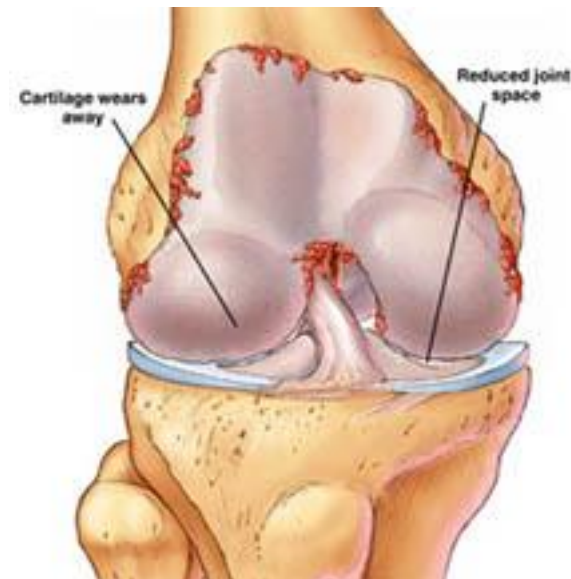
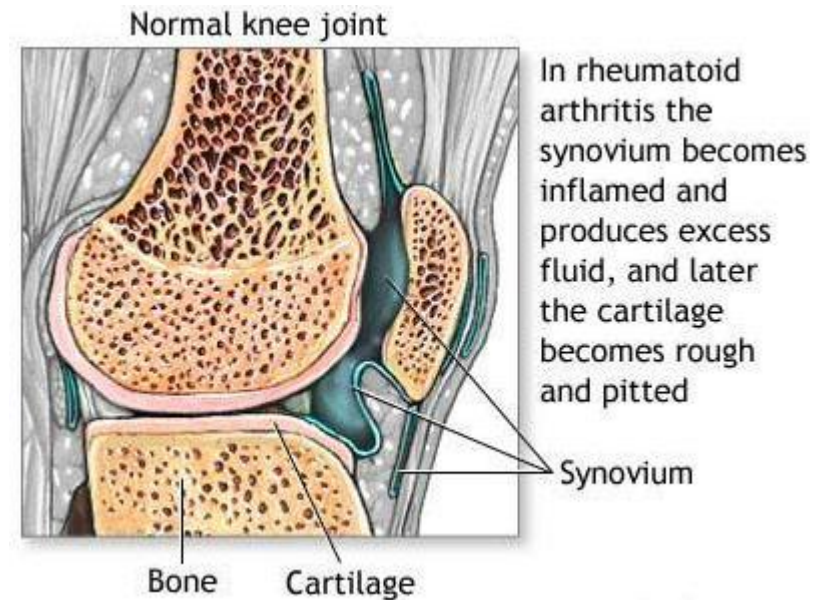
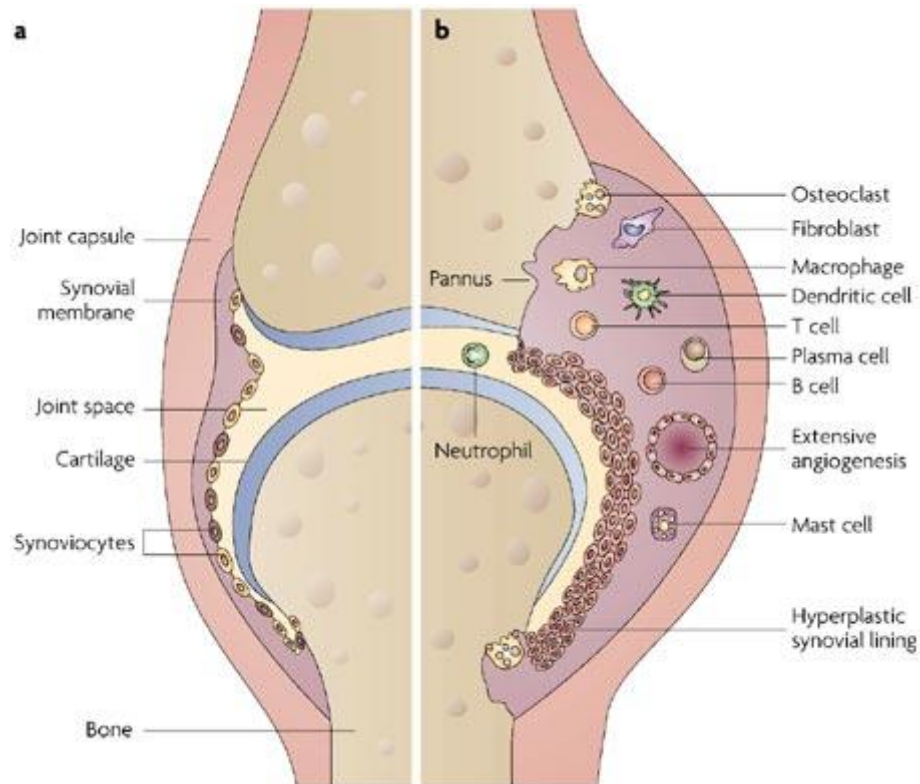
• MRI – CT Scan :

- An **MRI is recommended in all patients with complex partial seizures.** This test would be useful in looking for the etiology of the seizures rather than supporting the diagnosis.
- **CT scan** is useful in ruling out serious intracranial pathology acutely, such as hemorrhage or tumor following the first seizure.
- However, the MRI is the preferred neuroimaging technique when evaluating a patient with partial seizures. The resolution of the MRI is superior to CT in detecting subtle abnormalities, such as mesial temporal sclerosis or cortical brain malformation.
- The consulting neurologist orders an EEG and MRI study.

* Drugs used in seizures :

Narrow spectrum AED	Type of seizures	Broad spectrum AED	Type of seizures
Ethosuximide	Absence	Lamotrigine	Partial/generalized
Carbamazepine	Partial	Levetiracetam	Partial/generalized
Gabapentin	Partial	Phenobarbiton	Partial/generalized
Oxcarbazepine	Partial	Phenytoin	Partial/generalized
Phenytoin	Partial	Topiramate	Partial/generalized
		Valproate	Partial/generalized

Rheumatology



➤ **Case (1) :**

• A **4 year old child** presents with history of **pain in Rt knee** ,and both ankles, for 3 months, and her mother has been giving her ibuprofen to control, she is otherwise well.

➤ **What is the most likely diagnosis ?**

- Growing pains
- Oligoarticular JIA
- Dermatomyositis
- Systemic onset JIA
- Polyarticular JIA

* JIA = Juvenile idiopathic Arthritis

*** Examination :**

- Normal temp.&mild irritability
- Arthritis of the Rt wrist joint
- Swollen left knee joint

*** Investigations :**

- CBC: normal
- ESR : 17
- RF : -Ve

What should you do ?

- ↓ Joint Pain
- ↓ Joint Swelling
- No Diffuse/Systemic manifestations

1. Is it articular ?

2. Is it acute or chronic ?

3. Is inflammation present ?

4. How many/which joints are involved ?

5. Are there RED FLAGS ?

N.B. RED FLAG CONDITIONS :

- Septic non-gonococcal arthritis
- Indolent infections (as TB)
- Trauma
- Haemarthrosis
- Intra-articular metastatic cancer
- Fracture

➤ **Is it inflammatory or non inflammatory ?**

SEARCH FOR SIGNS AND SYMPTOMS OF INFLAMMATION

➤ **Is it mono & oligo or poly articular?**

- Oligoarthritis (involving 2-4 joints)
- Polyarthritis (involving 5 joints or more)
- Monoarthritis

➤ **Differential diagnosis you might think of in this patient ?**

- There is oligo arthritis
- In a 4 ys old child
- No systemic manifestations
- No hist of trauma ¬ septic &no hist. of bleeding disorder
- Investigations are within normal
- Response to NSAID

➤ **Most likely diagnosis is ?** • **Oligoarticular JRA**

Comment :

• as IT FULLFILLS THE CRITERIA OF DIAGNOSIS OF **JEUVENILE IDIOPATHIC ARTHRITIS** .. WHICH IS :

- Age of onset below 16 years
- Arthritis involving ,one or more joints(less than 5)
- Duration more than 6 ws
- Exclude other causes of arthritis.
- Why **oligo-articular** :
 - Age 4ys
 - Involving ≤4 joints
 - Involving large joints
 - No systemic manifestations

➤ **Case (2) :**

- 10 years old girl presents with a **high fever, rash, and hip and knee joint pain**
- slit lamp examination of her eyes is **normal**
- blood tests are **-ve for auto antibodies**

*** Examination :**

- Fever 38.5
- Rash
- Arthritis
- No uveitis

*** Investigations :**

- CBC: HB: 9.2 & WBCS : 19.000 & PLT: 430.000
- ESR: 110
- ANA : -Ve
- RF : -Ve

➤ **What is the most likely diagnosis ?**

- Enthesitis related arthritis
- Extended oligoarticular JIA
- Henoch schonliein purpura
- Systemic onset JIA
- Polyarticular JIA

➤ **Differential diagnosis you might think of in this patient ?**

- **Rheumatic:** SLE , systemic sclerosis, FMF
- **Nonrheumatic:** HIV, viral infections, causes of purpura, vasculitis
- **“Overlap Syndrome”** UCTD, MCTD

- This case is one of the common presentations of **Systemic onset JIA**

Comment :

- as IT FULLFILLS THE CRITERIA OF DIAGNOSIS OF **JEUVENILE IDIOPATHIC ARTHRITIS** .. WHICH IS :

- Age of onset below 16 years
- Arthritis involving ,one or more joints(less than 5)
- Duration more than 6 ws - Exclude other causes of arthritis.
- **Why Systemic onset JIA :**
- prominent systemic symptoms: fever, rash,
- arthritis
- No Uveitis
- Anaemia ,leucocytosis and thrombocytosis
- Increased ESR
- Negative ANA

➤ **Case (3) :**

- **12 years** old Egyptian **girl**, presents with :
 - **symmetrically painful wrists**
 - Intermittent episodes of **fatigue, fever** associated with **Arthralgia** and **Myalgia**
 - Over the past six month, she has **progressive weight loss** and **alopecia**
 - Recently on returning from the beach, she had **facial** and **body rash**

*** Examination :**

- Temperature: 38.5
- Blood pressure: 100/70
- Body weight: at the **3rd centile for age**
- Examination revealed **mild alopecia**
- **Erythematous facial rash**
- **Body rash**: over the trunk & shoulders & palms of both hands .
- **Bilateral wrist arthritis**

*** Investigations :**

- CBC:
normocytic hypochromic anaemia, lymphopenia & thrombocytopenia
- **Decreased C3,C4**
- **Raised immunoglobulins**
- **Positive ANA, Anti-DNA**
- **Positive anti-cardiolipin, anti RO-LA, anti-smooth ms. cytoplasmic abs**

➤ **What is the most likely diagnosis ?**

- Rheumatoid arthritis.
- Henoch schnolien purpura.
- Systemic lupus erythematosus.
- Cytomegaloviral infection .

➤ **Differential diagnosis you might think of in this patient ?**

- **Rheumatic**: SLE , systemic sclerosis, FMF
- **Nonrheumatic**: HIV, viral infections, causes of purpura, vasculitis
- **“Overlap Syndrome”** UCTD, MCTD

- Based on the clinical presentation, differential diagnosis might be difficult between :

Systemic onset JIA & SLE & CMV & EPV & HEMATOLOGICAL
MALIGNANCIES

>> Based on these investigations :

- This case is one of the common presentations of **SLE**

Comment :

- It **fulfills >4** of the ACR for diagnosis of lupus .

Can you enumerate them from the case scenario ?

- 1- Arthritis
- 2- Malar rash
- 3- lymphopenia, thrombocytopenia
- 3- Positive ANA
- 4- Positive anti-DNA
- 5- Positive anti-cardiolipin

- اللهم علمنا ما ينفعنا و انفعنا بما علمتنا وَ زِدْنَا علماً