Duchenne Muscular Dystrophy

The term "Muscular dystrophy" refers to a group of inherited progressive muscle disorders.
Degeneration of muscle fibres occurs, without morphological change.
Affects 1 per 3500 live male births.
The process is non-inflammatory with no central / peripheral nerve abnormalities.
Guillaume Duchenne (a French neurologist) reported on 13 patients with the condition.
The most severe and common form of Muscular Dystrophy is named after him.
Becker Muscular Dystrophy is the 2nd most common and most mild form.

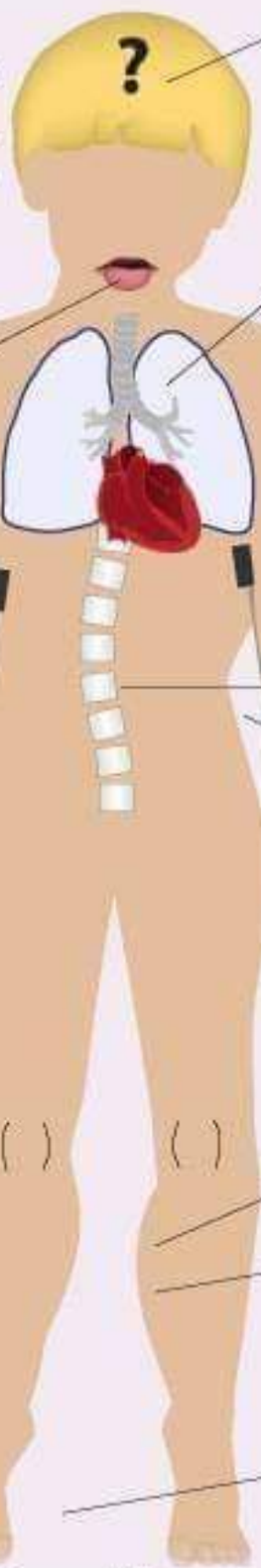
Aetiology - The condition occurs due to defects in the gene coding for dystrophin.
Dystrophin is a very large protein and errors can sometimes occur at multiple sites.
It is found in skeletal (striated), smooth & cardiac muscle fibres and also in the brain.
1 in 3 cases occur due to a spontaneous new mutation.

Inheritance Pattern

Sex linked
(defect located on the X chromosome)
eg Duchenne, Becker, Emery-Dreifuss

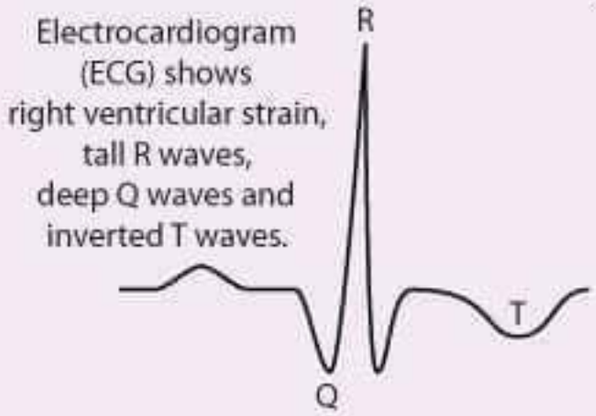
Autosomal dominant
eg Facioscapulohumeral, Distal, Ocular, Oculopharyngeal

Autosomal recessive
eg Limb-girdle form.



Features ...

Muscle weakness occurs from proximal to distal and usually presents when the boy begins to walk.



Apert's Syndrome (1 : 70 000 live births)

A craniosynostosis syndrome caused by a mutation in FGFR2 (**chromosome 10**). Most commonly occurs sporadically.

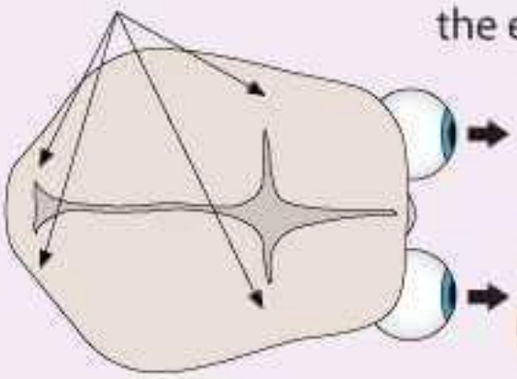
Craniosynostosis refers to premature fusion of one or more the crainial sutures. These sutures are the connections which separate the skull bones and premature fusion leads to an abnormally shaped head.

Other examples of syndromic craniosynostosis include :-

- Crouzon's Syndrome (1 :70 000 live births) Associated with ***normal*** hands & acanthosis nigricans.
- Pfeiffer's Syndrome (1 :120 000 live births) Associated with large thumbs and big toes.
- Muenke's Syndrome (1 :30 000 live births) Not associated with pathognomonic facial or llimb appearances

Bicoronal synostosis
(esp coronal and lambdoid sutures)

EXORBITISM :-
Relatively small orbits gives rise to relative protrusion of the eyes



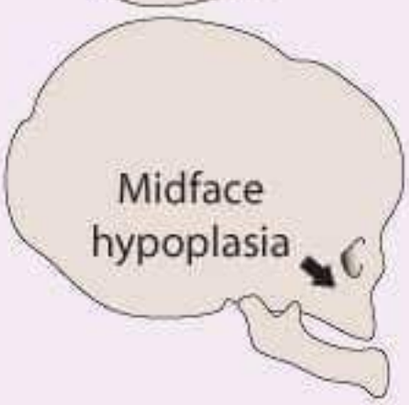
Obstructive sleep apneoa

Mental retardation
- hydrocephalus can occur
- Agenesis of the corpus callosum

Anti-Mongoloid slant

Low set ears

Small beaked nose



Midface hypoplasia

Vertebral fusions C5-6

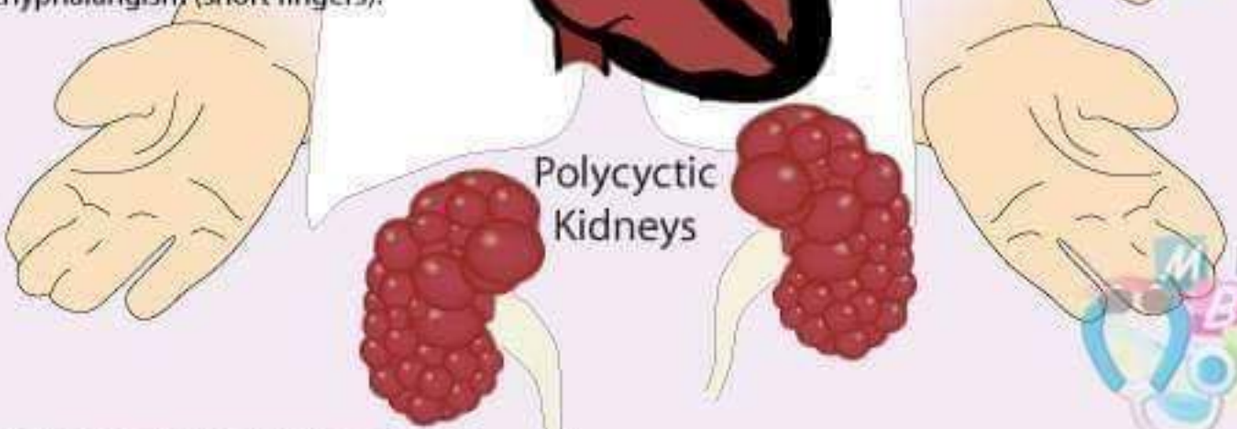
May have Cleft Palate

Type III: "hoof" or "rosebud" hand

Bilateral Complex Syndactyly
A short thumb with radial deviation.
Complex syndactyly (involving bone) of the index finger, long finger & ring finger.
Simple syndactyly (not involving bone) of the fourth webspace.
Symbrachyphalangism (short fingers).

Cardiopulmonary abnormailties

Type II: "spoon" or "mitten" hand.

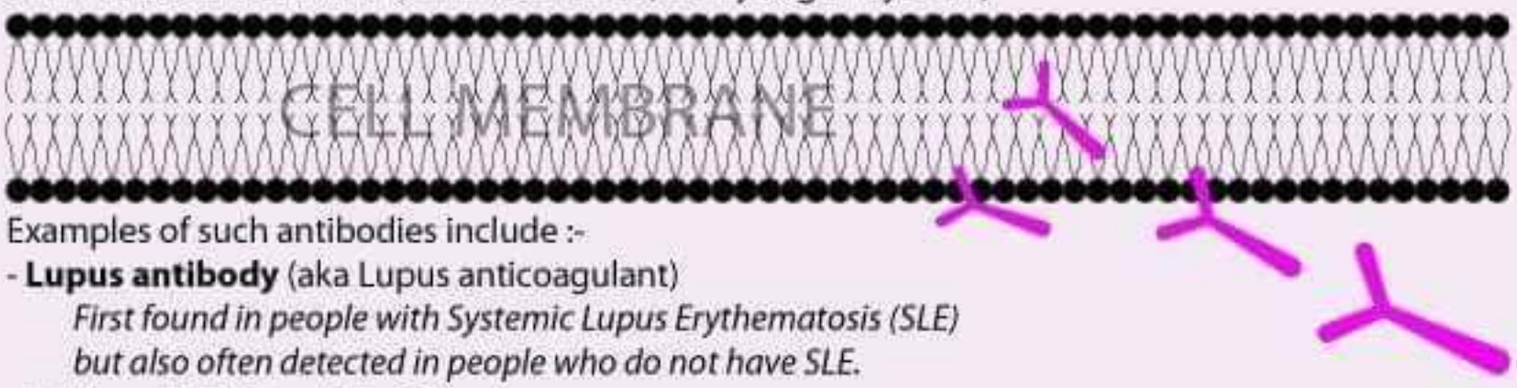


Polycyctic Kidneys

Type I "spade" hand

Antiphospholipid Syndrome (Hughes' Syndrome)

Antiphospholipid syndrome - an autoimmune condition where the affected individual produces antibodies against cell membrane components. It is more common in **women**. It can cause arterial/venous blood clots (in any organ system).



Examples of such antibodies include :-

- **Lupus antibody** (aka Lupus anticoagulant)
  First found in people with Systemic Lupus Erythematosis (SLE) but also often detected in people who do not have SLE.
- **Anti-cardiolipin antibodies**
- **Anti-β2-gycoprotein 1 antibodies**

The most common arterial event is **stroke** (cerebrovascular accident CVA)

Migraine can occur

Pre-eclampsia (assoc with raised blood pressure)

Heart Valve Disease

Oscillopsia (objects in the visual field appear to oscillate)

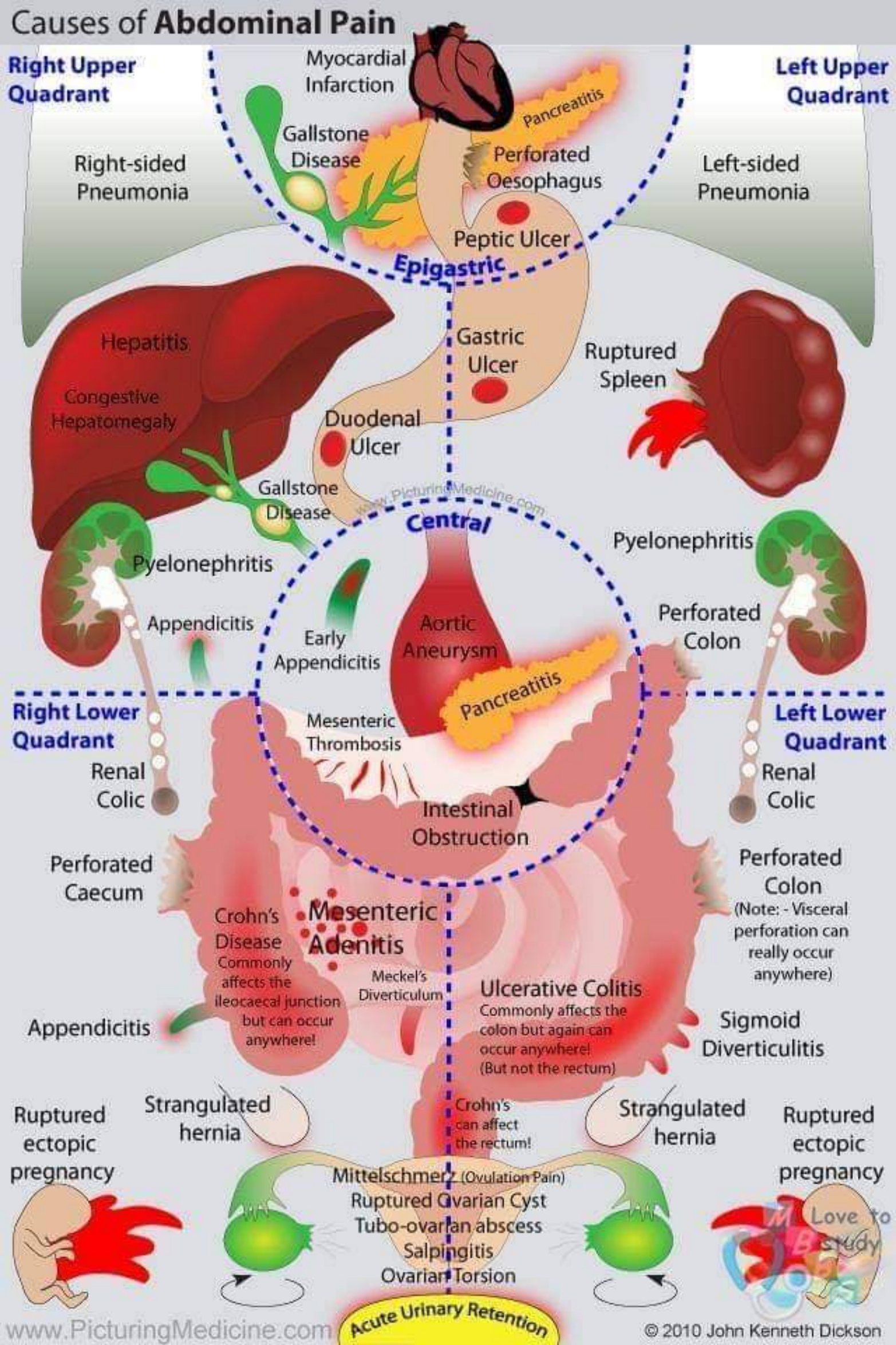
The most common venous event is **deep vein thrombosis** (**DVT**) of the lower leg.

Livedo reticularis (Mottled, "lace-like", purplish, vascular pattern of the lower extremities).

Pregnancy related complications e.g. **Miscarriage** & **Pre-eclampsia** *(Placental infarctions, early deliveries & stillbirth can also occur more frequently in women with Antiphospholipidsyn)*

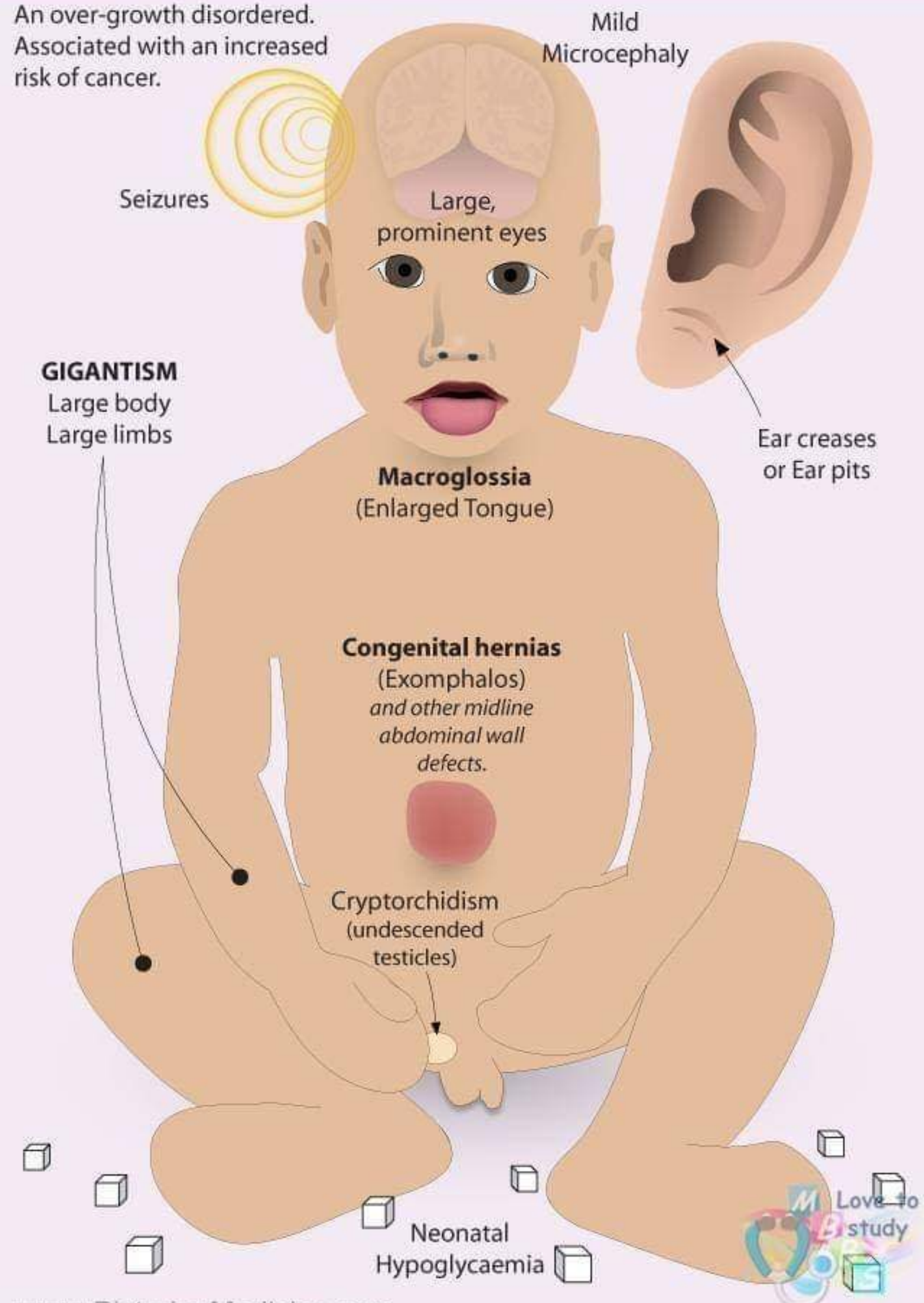
Thrombocytopenia (low platelet count)

Causes of **Abdominal Pain**



Beckwith Wiedemann Syndrome

An over-growth disordered. Associated with an increased risk of cancer.



Systemic Lupus Erythematosis

Most commonly affects women aged 20-40.
Is more common in Afro-Caribbeans.

Anaemia (of chronic dis) (70%)

Anaemia
Normochronic normocytic

Haemolytic anaemia (10%)

Haemolysis

Fever

Hypertension

Epilepsy

Headaches

Arthropathy

CNS (50%)
Headaches
Polyneuropathy
Stroke, psychoses, depression (rare)
Epilepsy (rare)

Sjogren's syn. *(Dry eyes & dry mouth)*

Oral ulceration

Hypothyroidism (rare)

Generalised lymphadenopathy (50%)

Purpura

Raynaud's (10%)

Polyneuropathy

Thrombocytopenia leading to purpura.

Skin (80%)
(photosensitivity)
Malar rash (30%)
Non-specific erythema

Lungs
(Affected in 50%)
Lupus pneumonitis
Pulmonary fibrosis ("honeycomb lung")
Pleurisy +/- effusion.
Patchy consolidation (Areas of collapse).
Shrinking lungs (uncommon but characteristic)

Cardiovascular
(Affected in 40%)
Pericarditis

Splenomegaly (rare)

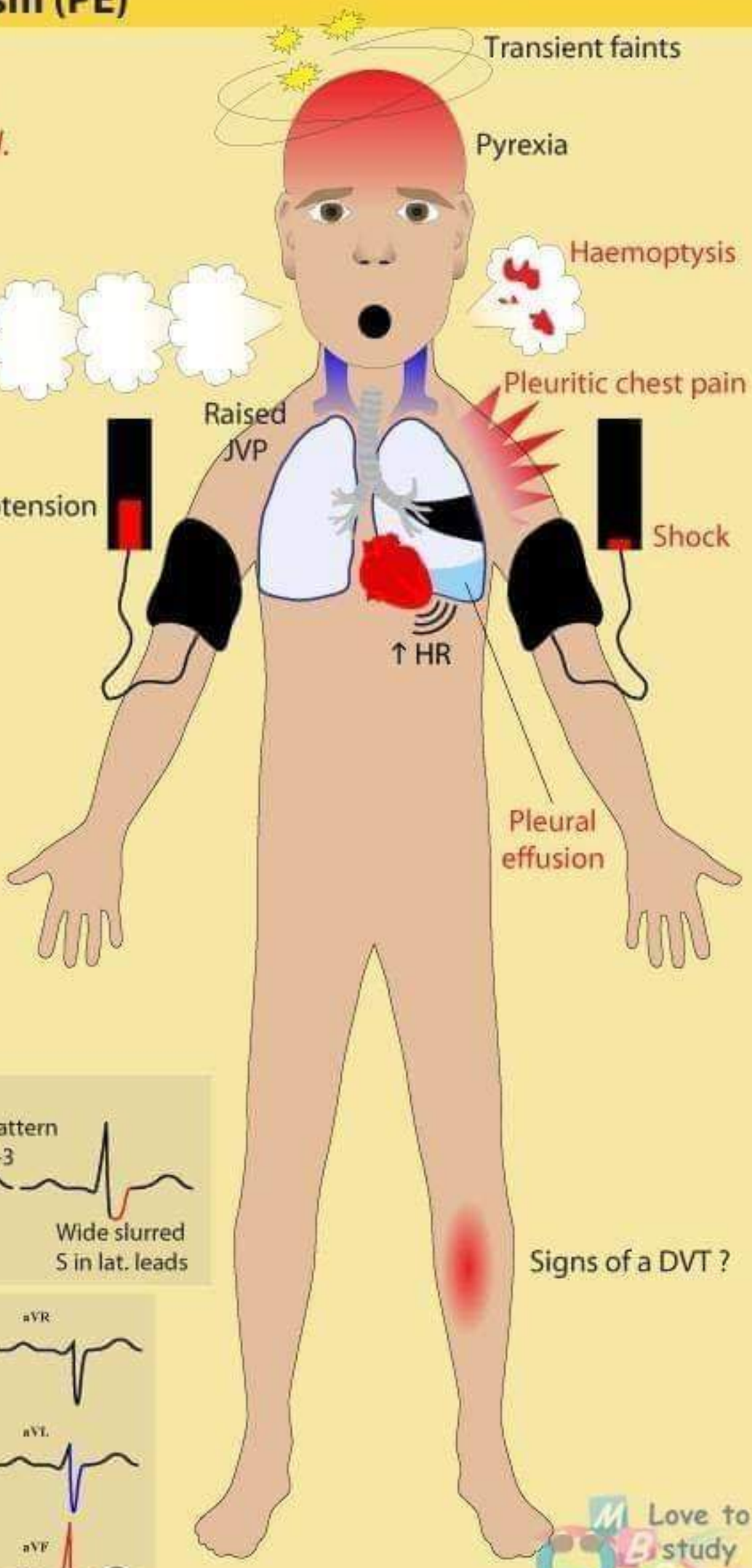
Renal
(Affected in 100%)
Almost any manifestation of renal disease incl....
Hypertension.
Nephrotic syn.

Joints (90%)
Migratory, asymmetrical polyarthropathy.
Finger, Wrists, Elbows, Shoulders, Knees & Ankles.

Haematuria & Proteinuria

Pulmonary Embolism (PE)

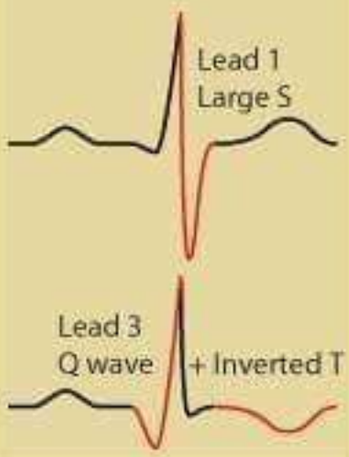
Features of a PE.
Those features typical of a medium-large PE are in red.



ECG Changes

Tachycardia

S1 Q3 T3

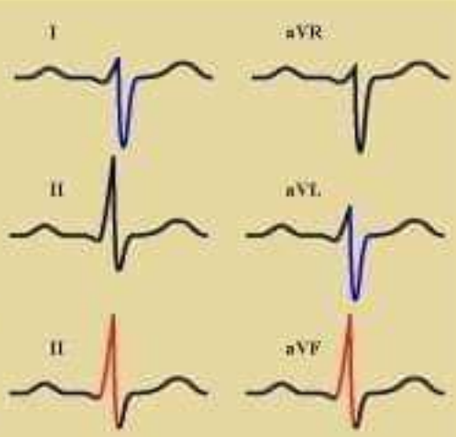


RBBB

Rt Axis Deviation

Leads I & aVL QRS -ve (dom S wave)

Leads III & aVF QRS +ve (dom. R wave)

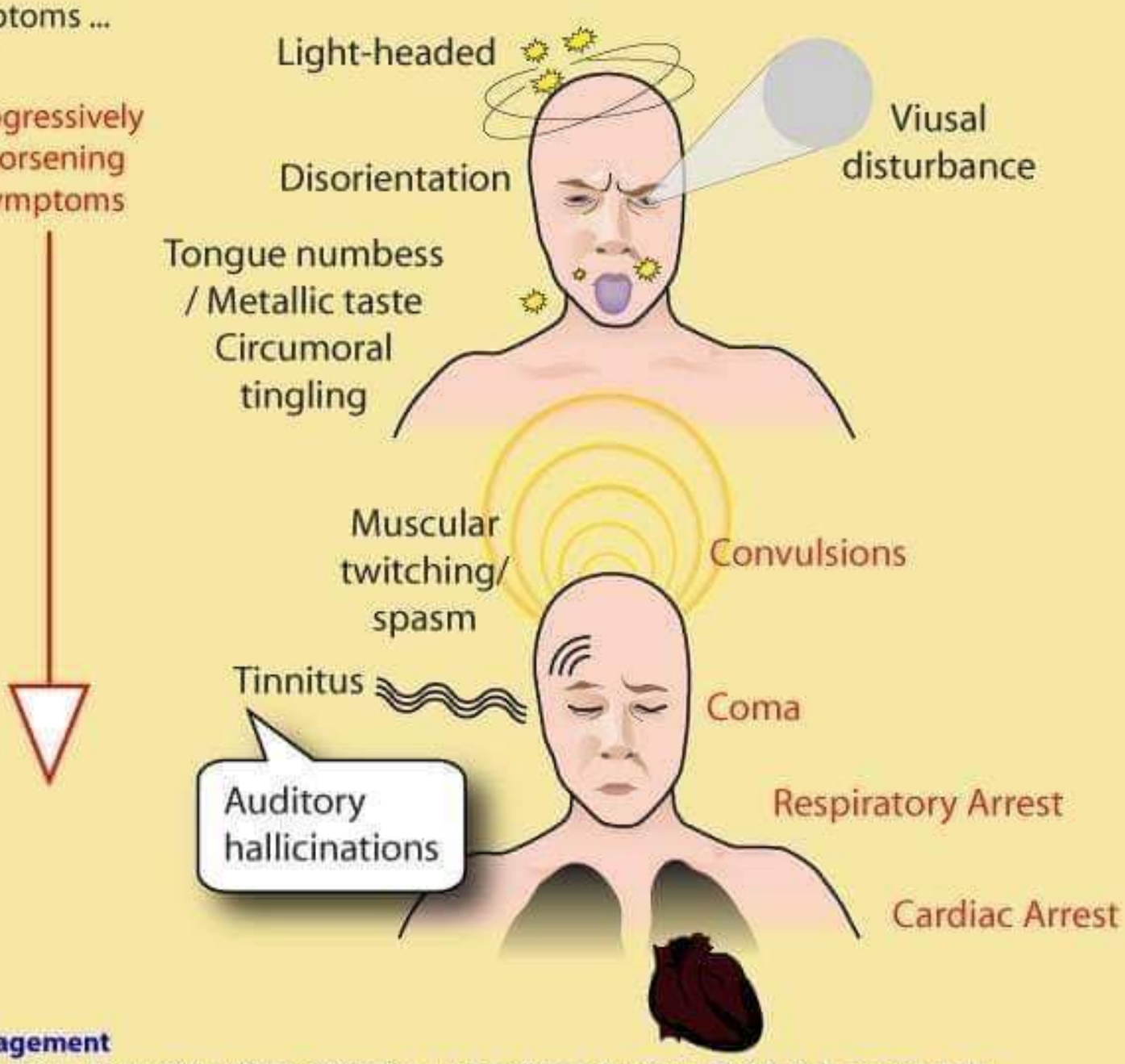


Local Anaesthetic Toxicity

Effects ...

CNS	Excitation / Dizziness / Tinnitus / Circumoral numbness or tingling.
Sezuires	Threshold is lowered by hypoxia/hypercarbia and acidosis. Benzodiazepines can help by increasing the threshold. Thiopental can also be used for treatment.
Cardiovascular	Na channel blockade. Decreased purkinje discharge. Prolonged conduction times. Re-entrant ventricular arrhythmias.

Symptoms ...

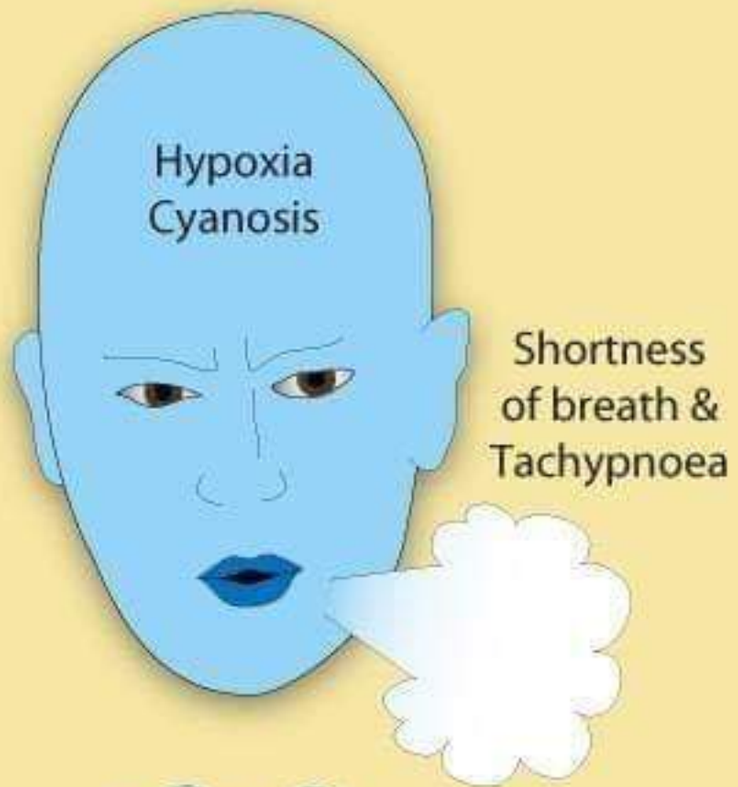


Management

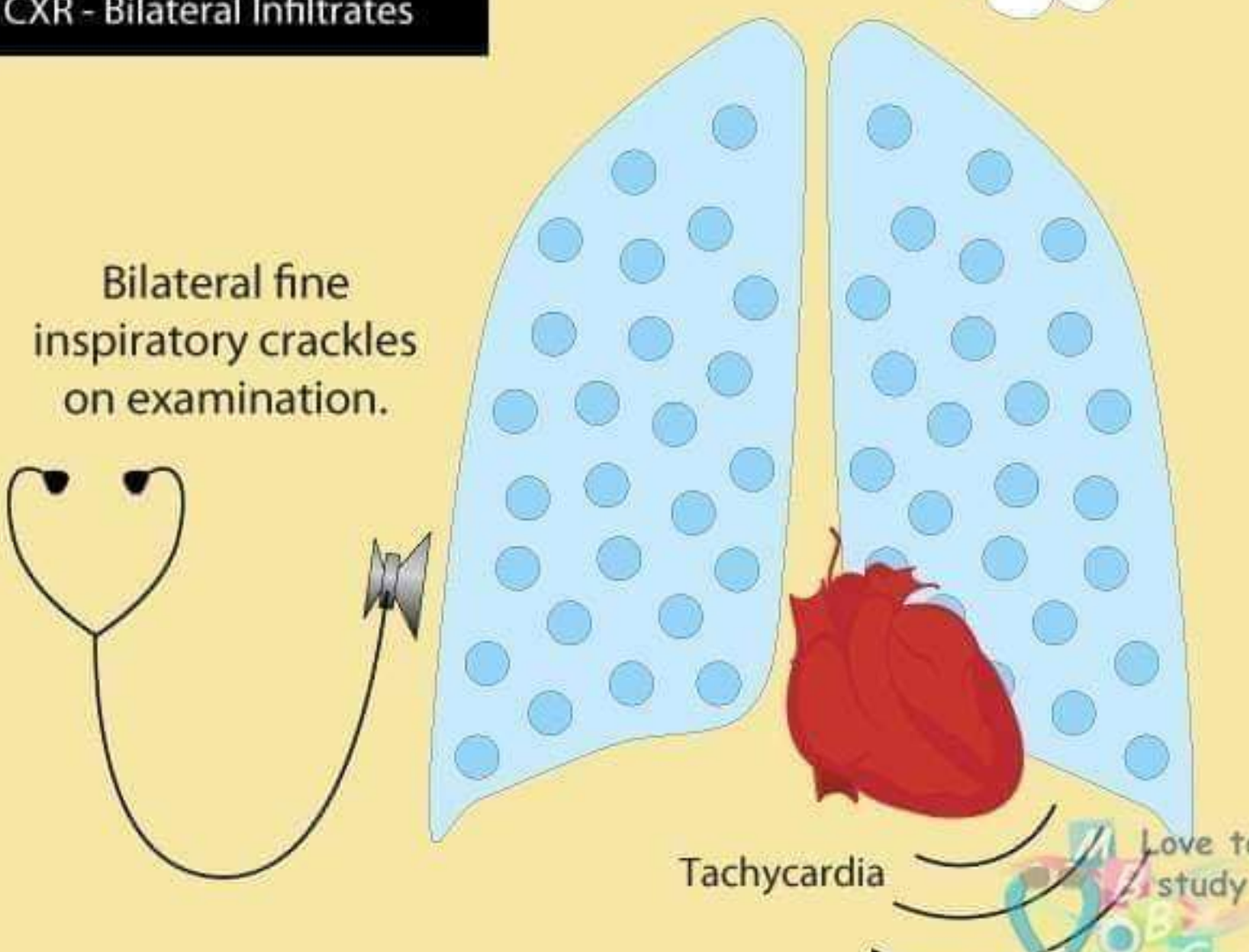
Prevention - Frequent syringe aspirations + Small test doeses + Divided doses can help.
Stop further injections and follow BLS and ALS principles.
Airway & Breathing - Hyperventilate with 100% oxygen.
This conteracts the effects of hypoxia, hypercarbia and acidosis.
Seizure suppression - Benzodiazepines (eg midazolam).
Muscle relaxants may help to stop movements during seizures.
IV 20% lipid emulsion - acts as a binding agent to the local anaesthetic.
Fluid resuscitation. ? Vasopressor, antiarrhythmic and inotropic therapy may be needed.

Acute Respiratory Distress Syndrome (ARDS)

Increased capillary permeability within the lung leading to oedema.
Caused by either direct lung injury or secondary to severe systemic illness.
Often accompanied by multi-organ failure.
Leads to respiratory distress (cyanosis, tachypneoa and tachycardia)



Bilateral fine inspiratory crackles on examination.



Cirrhosis leading to Chronic Liver Failure

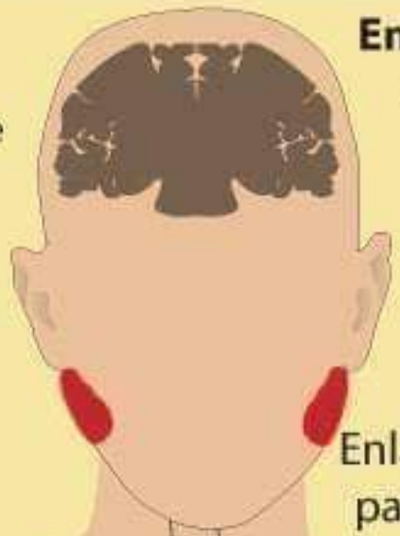
Leuconychia
(white nails with lunulae undemarcated, due to hypoalbuminaemia)

Terry's nails
(white proximally + distal 30% red due to telangiectasias)

Finger Clubbing

Dupuytren's contracture

Palmar erythema
(hyperdynamic circulation)



Encephalopathy
- Confusion
- Coma
- Liver Flap (Asterixis)

Asterixis

Enlarged parotid glands

Spider naevi

Oesophageal varices

Gynaecomastia
Breast development in men.

↑LFTs

Hepatomegaly
(or small liver can occur in late disease)

Portal Hypertension
- Splenomegaly
- Caput medusae
(enlarged superficial periumbilical veins)
- Oesophageal varices
- Ascites

↑Risk of Hepatocellular Carcinoma

Coagulopathy
(↓ production of clotting factors = tendency to bleed)

Ascites

Testicular atrophy

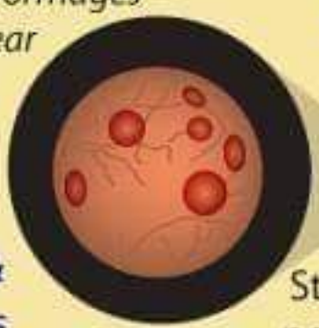
Hypoglycaemia
(low sugar levels)

Hypo-albuminaemia

Infective Endocarditis

An infection of the endocardial surface of the heart.
Intractable congestive heart failure may result.
If left untreated it is generally fatal.
Subacute endocarditis - symptoms are subtle & non-specific (in blue)

Roth spots - *rare*
(retinal haemorrhages with small clear centres)



Flu-like syndrome
Low grade / intermittent fever

Intra-cerebral pathology
Embolic Stroke
Multiple cerebral microabscesses
Intracerebral haemorrhage
Delirum

Conjunctival haemorrhage

Anorexia & weight loss

Stiff neck

Pallor

Pericardial rub
Pleural friction rub

Pleuritic pain

Arrhythmias
Heart murmurs

Abominal symptoms
(RUQ pain, vomiting, Appendicitis-like pain)

Splenomegaly

Janeway lesions
(non-tender macules on the palms & soles)

Petechiae

Subungual (splinter) haemorrhages
(Dark red linear lesions in the nail bed).

Osler nodes
(tender subcutaneous nodules in the pulps of the fingers).

Aquired Immunodeficiency Syndrome (AIDS)

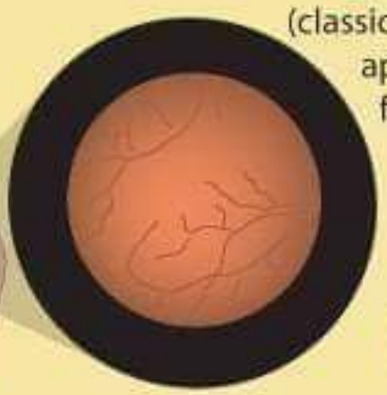
The following are often regarded as ***AIDS defining illnesses***.

Neurological

AIDS-related dementia
HIV-related meningitis
CMV encephalitis
T. gondii - the main CNS pathogen in AIDS (ring-shaped lesions on CT)
Cryptococcus neoformans - causes an insidious meningitis
Tumours
1° cerebral lymphoma.
B-cell non-Hodgkin's Lymphoma

CMV retinitis

(classic "mozarella pizza" appearance on fundoscopy)



Oral Cavity

- Aphthlous ulcers
- Oral Tumours
- Candida
- HSV

Cancers may develop

Lymphoma
Perianal SCC
Brain Tumours
etc

Hepatomegaly

(↑LFTs)
- Drug related
- Viral hepatitis
- AIDS sclerosing cholangitis
- MAI

Pulmonary

Pneumocystis carinii pneumonia (most common life-threatening infection in AIDS)
Pyogenic bacteria
M. Tuberculosis
M. avium intrcellulare
Fungi (Aspergillus, cryptococcus, histoplasma)
Lymphoid interstitial pneumonitis
Non-specific pneumonitis
Lymphoma
CMV

Kaposi Sarcoma

Leishmaniasis

A parasitic disease spread by sand flies. Presents with cutaneous, mucocutaneous or visceral involvement.
Cutaneous disease manifests as ulceration.

Perianal disease

HSV ulceration
Perianal warts
Kaposi sarcoma.
SCC (rare)

Chronic diarrhoea

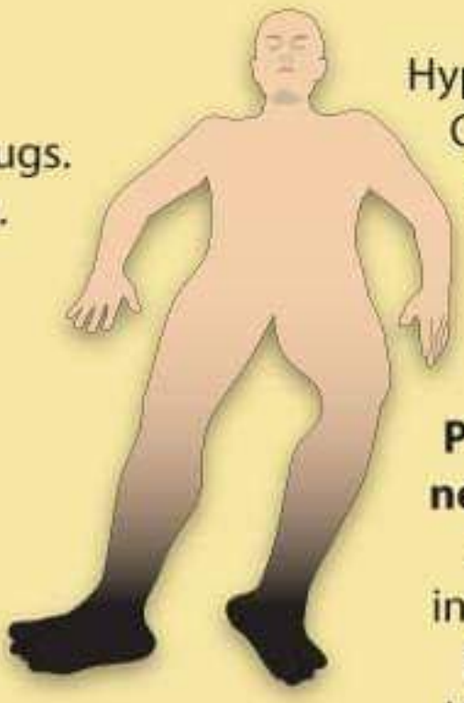
- Bacteria (Salmonella, Shigella, Campytlobacter, Atypical mycobacteria, C. diff)
- Protozoa (Cryptospoidium, Microsporidium)
- Viruses (CMV, adenoviruses)

Acute Porphyria

A number of different types of acute porphyria exist (autosomal dominant).
All are caused by an error in the **haem biosynthesis pathway**.
This leads to the accumulation of **toxic porphyrin precursors**.
Toxins include ***porphobilinogen*** and ***delta-aminolaevulinic acid***.
An acute neurovisceral crisis can result.

Intermittent porphyria
Attacks can be precipitated by various drugs.
Attacks occur more commonly in women.
Autosomal dominant.

Hereditary Coproporphyria
Causes photosensitive skin blisters.
Cutaneous manifestations can occur.



Hypotension
Collapse
Shock

Peripheral neuropathy
Sensory impairment.
Paralysis.
Hypotonia.

↑WCC

Hyponatraemia
Hypokalaemia

K+

Na+

Psychosis
Odd behaviour
Seizures

Fever

Sight affected (hallucinations)

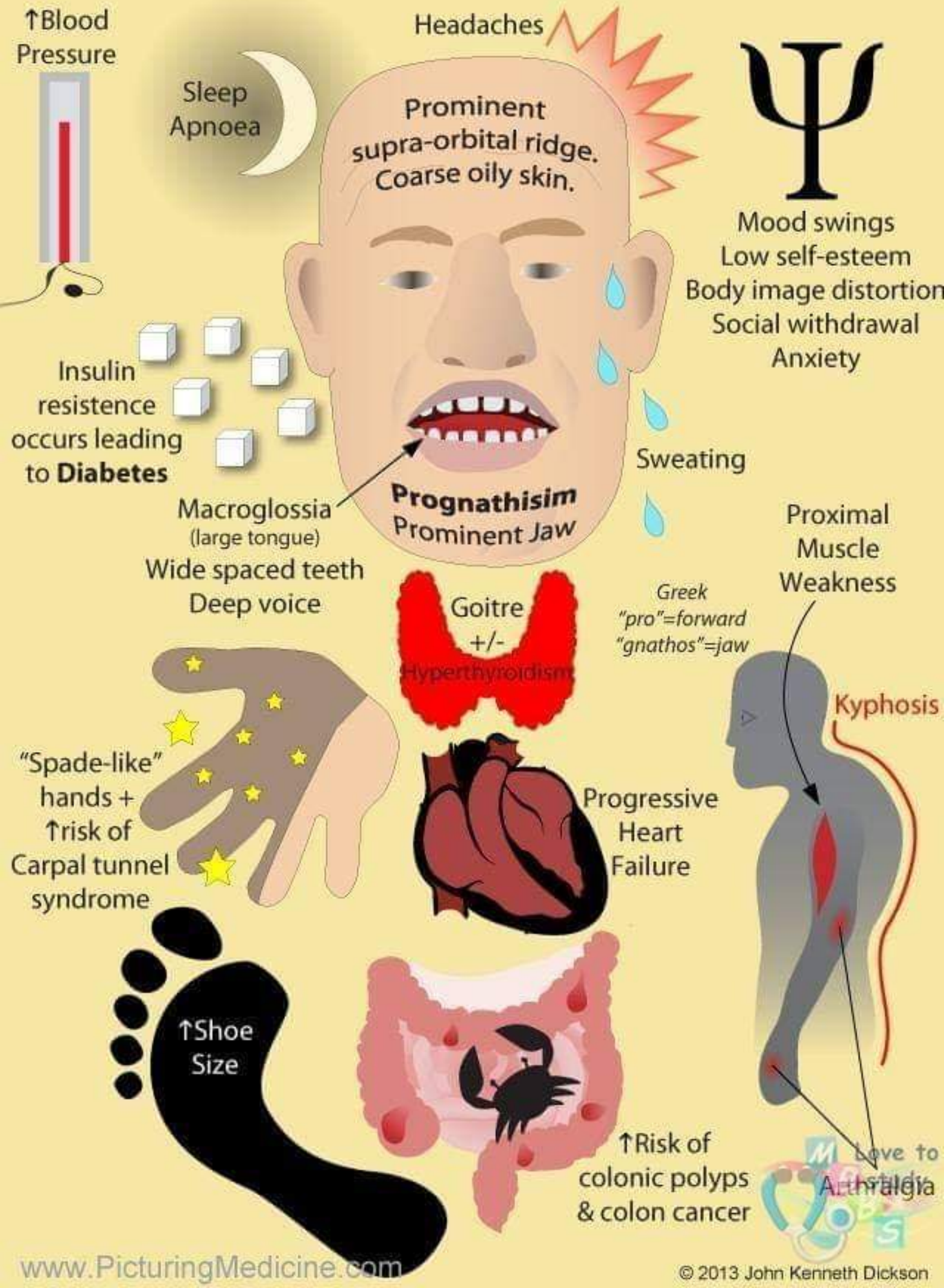
Can present with **Acute Abdominal Pain**
Beware - anaesthesia can be disasterous!

Proteinuria
Porphyrins can be detected in the urine

Colic & Vomiting

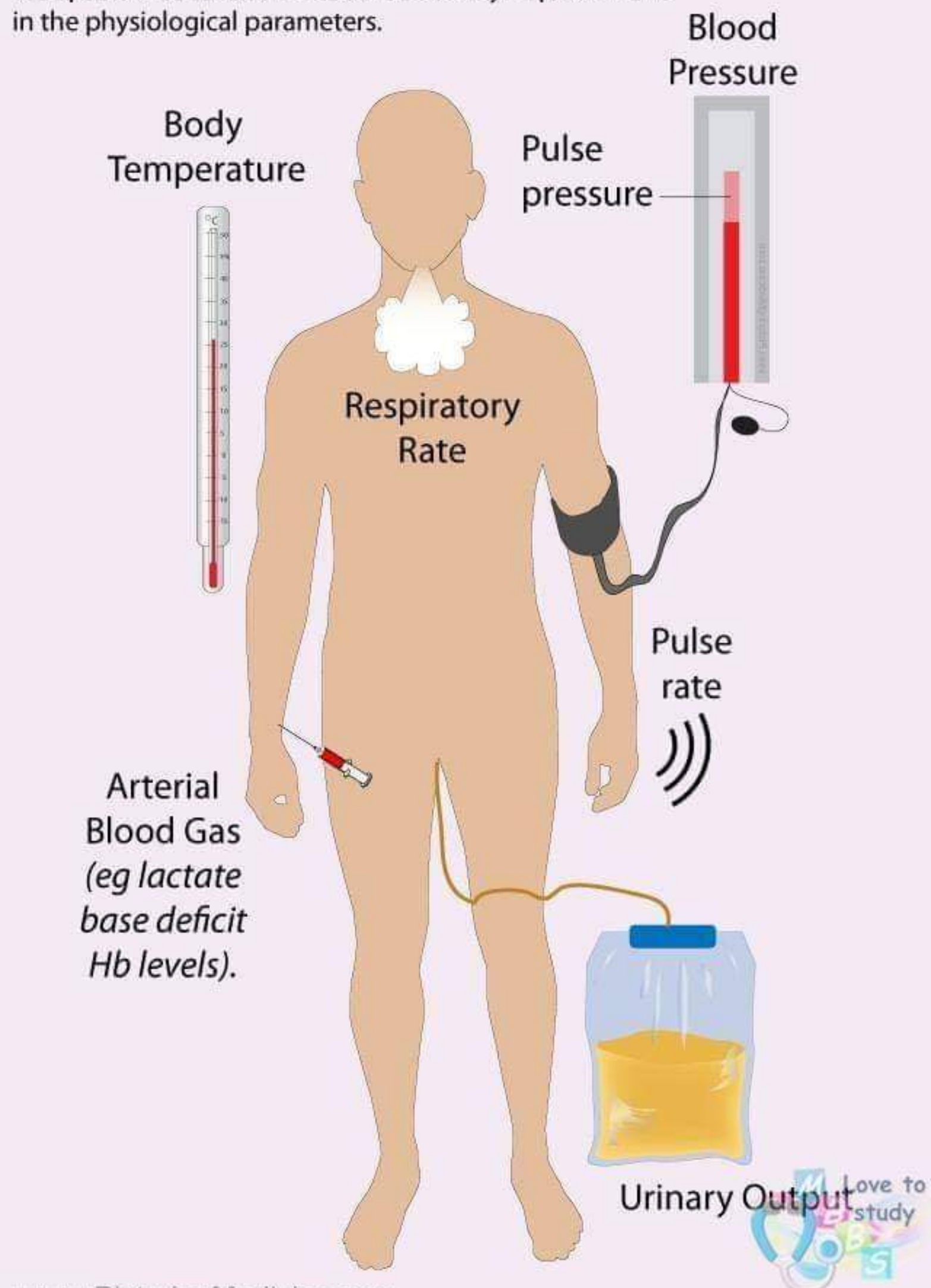
Acromegaly

Caused by over-secretion of **growth hormone (GH)** from the **pituitary gland**.
The condition is rare and tends to present between 30 and 50 years of age.



Assessing the adequacy of resuscitation

Adequate resuscitation is best assessed by improvement in the physiological parameters.



Down's Syndrome

Trisomy of chromosome 21.

Alzheimer's disease can occur.

Trisomy of chromosome 21.

Dysmorphic Round Face

Learning difficulties & Developmental Delay (Mean IQ of 50%)

Epicanthic folds

Brushfield spots on iris

Small Ears

Flat occiput

Protruding tongue (not macroglossia though!)

Abundant Neck Skin

Atlantoaxial instability

Hypothyroidism

Single palmar crease

Incurved little fingers (Clinodactyly)

Cardiac defects in 50%
(Ventricular septal defect, Patent ductus arteriosus, Atrial septal defect)

Respiratory infections

Increased risk of Leukaemia

Gap between 1st & 2nd toes (Sandal toe gap)

Duodenal atresia (double bubble sign on AXR)

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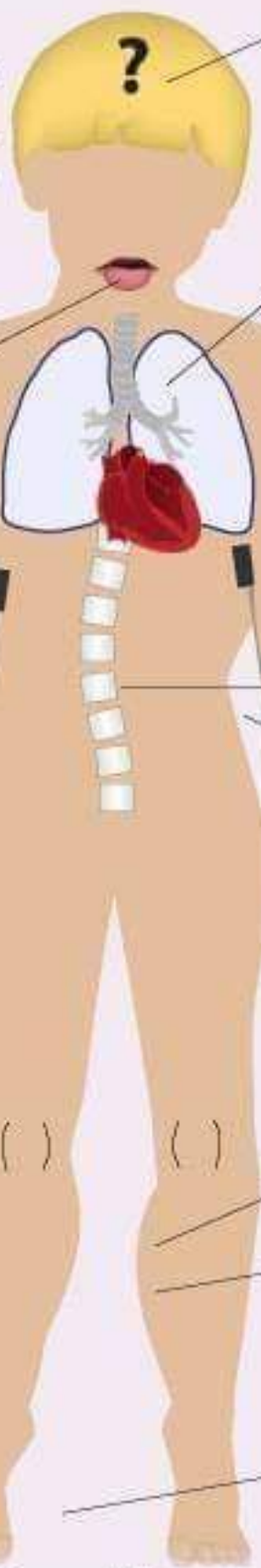

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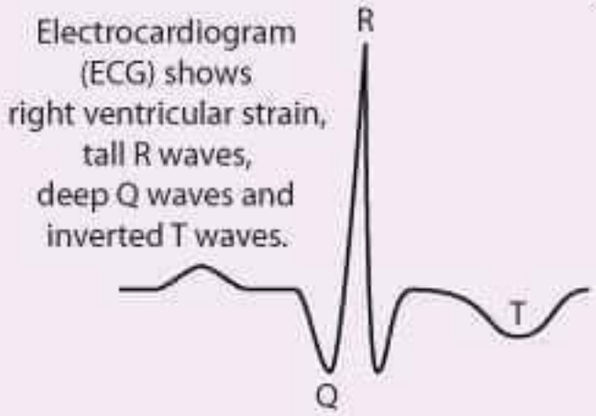

Secondary Survey = Head-to-toe evaluation of the patient

Performed **after the primary survey**, once **resuscitation is underway** and **vital signs are normal**.
Revisit the history - Blunt or penetrating trauma? Ejection from a vehicle?

Review CXR
Pneumothorax?
Wide mediastinum?

Pentrating injury to the eyes?

Pupillary size?

?Lens dislocation

? Visual acuity

Scalp lacerations

Pen Torch

Check eye movements! Entrapment of ocular muscles?

CSF leak?

Contact lenses?

MAXILLO-FACIAL
Skull, facial & cribriform fractures?

Mid-face Stability?
Missing teeth?
Malocclusion?

Subcutaneous Emphysema?
Laryngeal Fractures?

Lacerations deep to platysma
Consider angiography or surgery.

C-SPINE

Carotid Artery Injury?
Blunt trauma? Bruits?

CHEST (front & back)
Palpate & Ascultate.
Pneumothorax?
Flail segments?

Tension Pneumo-thorax?

? Brachial Plexus Injury

Examine the Back & Spine.

Complete neurological exam

Bruising + limb paralysis

Rib Fractures?

Cardiac Tamponade

LIMBS
Tenderness?
Deformity?
Brusing?
Crepitus?
Fractures?
Compartment Syn?

Abdominal Tenderness/Distension?
Injuries to retroperitoneal organs can be difficult to identify (even on CT)

Distal limb iaschaemia?
Pulse oximetry?
Capillary refill?

Seat belt bruising?
(?Ruptured viscus)

Pelvic Fractures?

PERINEUM
Brusing?
Urethral blood?

Meatal blood?

BURNS?
Circumferential
? escharotomy
Brassy cough?
Productive of soot?
Soot/blisters/oedema of tongue/pharynx

PER RECTUM
Blood?
Lacerations?
Normal sphincter tone?
High-riding prostate?

VAGINA
Foreign bodies?
Lacerations?
Pregnancy test?

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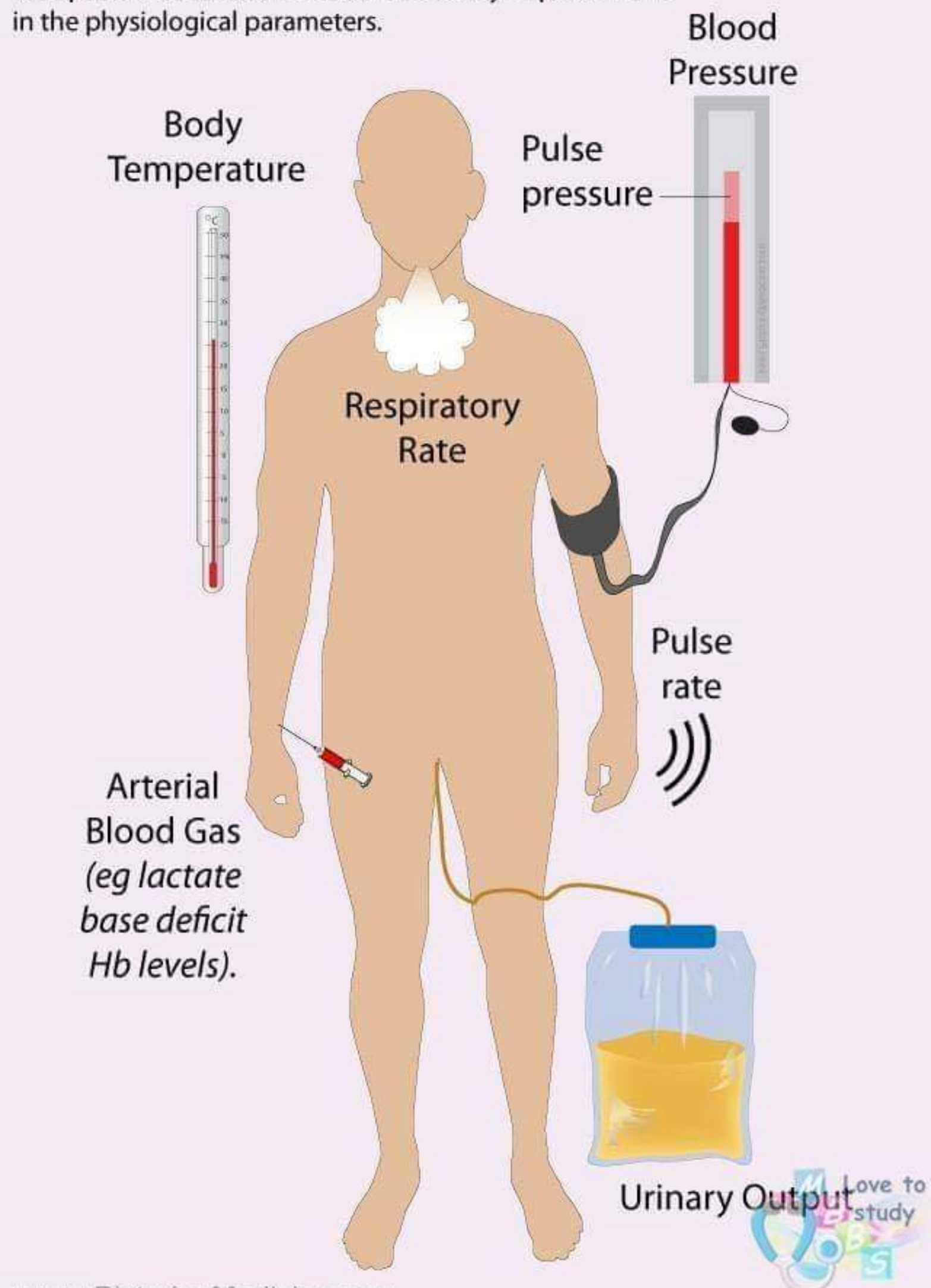


Primary Survery Adjuncts

Ventilatory Rate & ABG
Can be used to monitor adequancy of respiration. ET tubes can be dislodged when the patient is moved. The pCO2 reflects the adequacy of ventilation.

ECG monitoring

Blood Pressure
A poor measure and late indicator of tissue perfusion.

X-rays (chest & pelvis)

Gastric Catheter
Decompression reduces but does not eliminate the risk of aspiration. The tube should be inserted orally if there is risk of a cribriform fracture.

Pulse Oximetry Measures oxygen saturation of Hb colorimetrically. It does not measure the paO2
Do not place distal to the BP cuff as misleading results are produced.

Urinary Catheter (plus urine specimen)
Urine output reflects renal perfusion.
It is a sensitive indicator of volume status. The rectum and genitalia should be examined prior to placement.
A catheter should not be placed trans-urethrally if there is a urtheral injury. Retrograde urethrogram may be indicated.

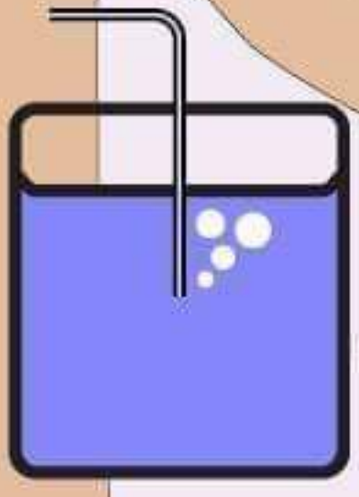
Breathing - Management

Give **supplemental oxygen** to all patients. If not intubated oxygen should be administered via a mask-reservoir device.

If **tension pneumothorax** is supected ... **Chest decompression** is required.

Open ("sucking") pneumothorax requires occulsive dressing with tape on 3 sides

Use **pulse oximetry** to monitor oxygen saturation of Hb.



A chest drain may be needed

Intubation and mechanical ventilation, can help manage "breathing-related" issues too. eg severe hypoxemia because of contusions etc
(i.e. It is not just helpful for airway management)

Analgesia may also be very important to enable adequate breathing and ventilation (eg for patients with rib fractures).

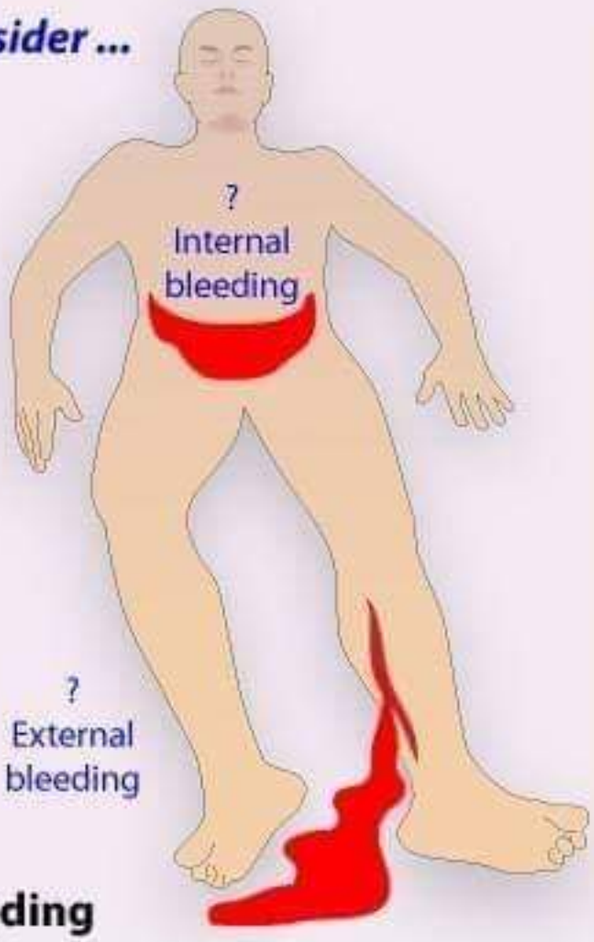
Circulation - Assessment

Blood volume, cardiac output and bleeding are the major circulatory issues.

Blood Volume and Cardiac Output

Haemorrhage is the major cause of preventable death after injury.
Once tension pneumothorax has been eliminated as a cause of shock, low BP following injury is considered hypo***volaemic*** until proven otherwise.

Consider ...



Level of consciousness - can help to indicate perfusion of the brain.

Pulse - A full slow steady pulse is usually a sign of normovolaemia. Caution re patients who are taking beta-blockers.

Skin colour - a patient with pink skin (esp face & extremities) rarely has critical hypovolaemia after injury. Ashen, gray face and pale extremities is worrying.

Bleeding

The source of bleeding should be identified as either external or internal.
External haemorrhage is identified and controlled during the primary survery.
Direct pressure is usually effective in treating bleeding.
Tourniquets are rarely required but are effective for massive exanguinating haemorrhage from an extremity (there is a risk however of ischaemia).
Haemostats can damage nerves and veins.

The source of bleeding can usually be determined by physical examination and imaging (eg CXR, pelvic x-ray, FAST)

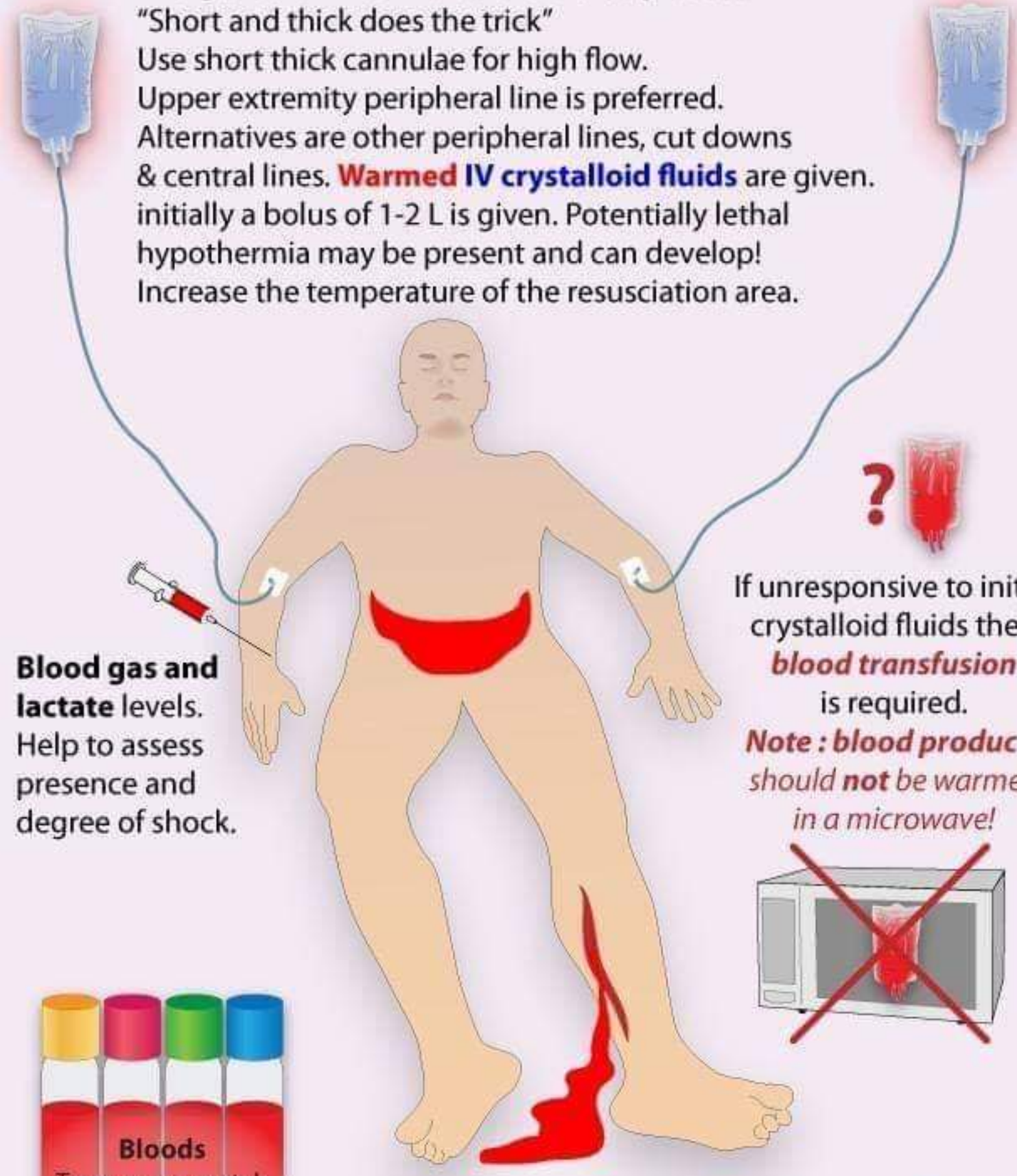
Major areas of internal haemorrhage (imaging) *potential management* :-

- Chest (CXR) *chest decompression, chest drain*
- Abdomen (FAST) *surgical intervention*
- Retroperitoneum (serial examination/double- or triple-contrast CT) *surgical intervention*
- Pelvis (x-ray) *pelvic binder*
- Long bones (x-ray) *splint application.*

Circulation - Management

Control bleeding and replace lost fluid. *Aggressive volume resuscitation is* ***not*** *a substitute for definitive haemorrhage control which may require ...* **surgery**, **angio-embolisation** *and* **pelvic stabilisation**.

2 large-bore IV cannulae should be placed.
"Short and thick does the trick"
Use short thick cannulae for high flow.
Upper extremity peripheral line is preferred.
Alternatives are other peripheral lines, cut downs & central lines. **Warmed IV crystalloid fluids** are given. initially a bolus of 1-2 L is given. Potentially lethal hypothermia may be present and can develop!
Increase the temperature of the resusciation area.



Blood gas and lactate levels. Help to assess presence and degree of shock.

If unresponsive to initial crystalloid fluids then ***blood transfusion*** is required.
Note : blood products *should* ***not*** *be warmed in a microwave!*

At the time of insertion, **take blood** for ...
- type and cross-match and baseline haematology.
- pregnancy test (for females of child-bearing age).
- toxicology / alcohol levels.

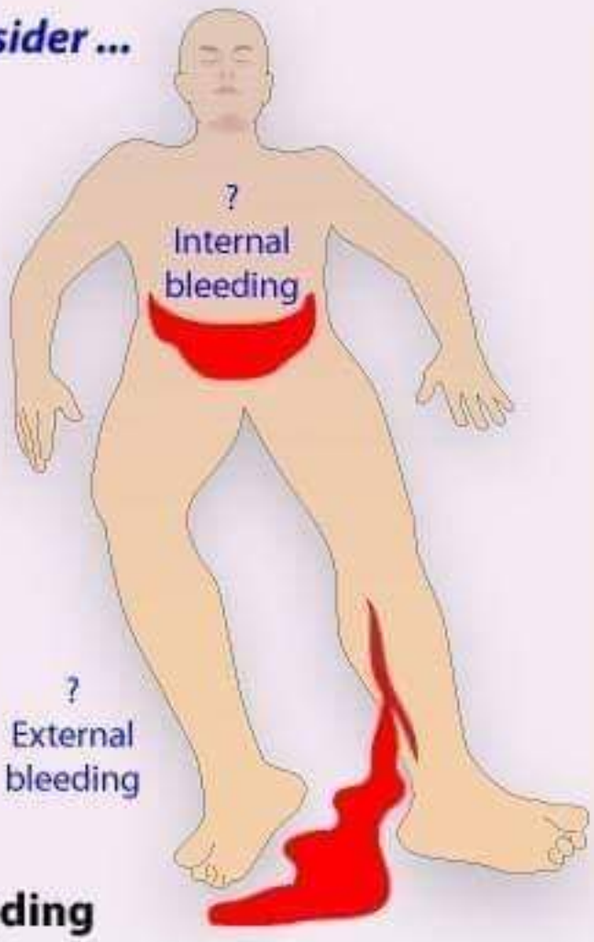
Circulation - Assessment

Blood volume, cardiac output and bleeding are the major circulatory issues.

Blood Volume and Cardiac Output

Haemorrhage is the major cause of preventable death after injury.
Once tension pneumothorax has been eliminated as a cause of shock, low BP following injury is considered hypo***volaemic*** until proven otherwise.

Consider ...



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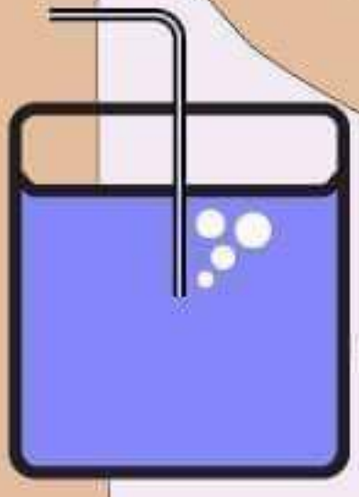
Breathing - Management

Give **supplemental oxygen** to all patients. If not intubated oxygen should be administered via a mask-reservoir device.

If **tension pneumothorax** is supected ... **Chest decompression** is required.

Open ("sucking") pneumothorax requires occulsive dressing with tape on 3 sides

Use **pulse oximetry** to monitor oxygen saturation of Hb.



A chest drain may be needed

Intubation and mechanical ventilation, can help manage "breathing-related" issues too. eg severe hypoxemia because of contusions etc
(i.e. It is not just helpful for airway management)

Analgesia may also be very important to enable adequate breathing and ventilation (eg for patients with rib fractures).

Breathing - Assessment

If the airway is clear ***then*** the next consideration is **adequate gas exchange (i.e. ventilation).**

Remember to *expose the chest* ***FULLY***

Inspect ? Dyspnoea / Tachypnoea

Ascultate

Percuss

Investigations ...

Pulse oximetry

Measures ***oxygen saturation*** of ***Hb*** colorimetrically. Does not measure the partial pressure of oxygen or carbon dioxide (which can more accurately reflect adequate ventilation).

Arterial Blood Gas

? USS

May be helpful for assessing pneumothorax

Must be identified during the **Primary Survey ...**

Flail chest

Tension Pneumothorax

Open Pneumothorax

Massive Haemothorax

More likely to be identified during the **Secondary Survey ...**

Simple Pneumothorax

Tracheobronchial tree injury

Rib Fractures

Pulmonary contusion

Traumatic aortic disruption

Traumatic diaphragmatic injury

Blunt cardiac injury

Simple Haemothorax

Blunt oesophageal rupture

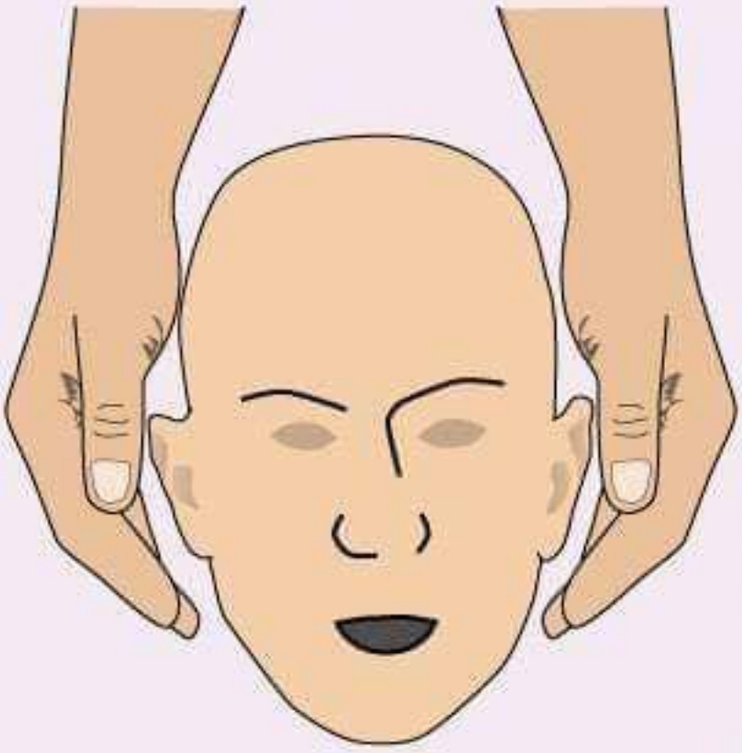
Airway : CERVICAL SPINE CONTROL

With any history of trauma **suspect a injury to the cervical spine**...

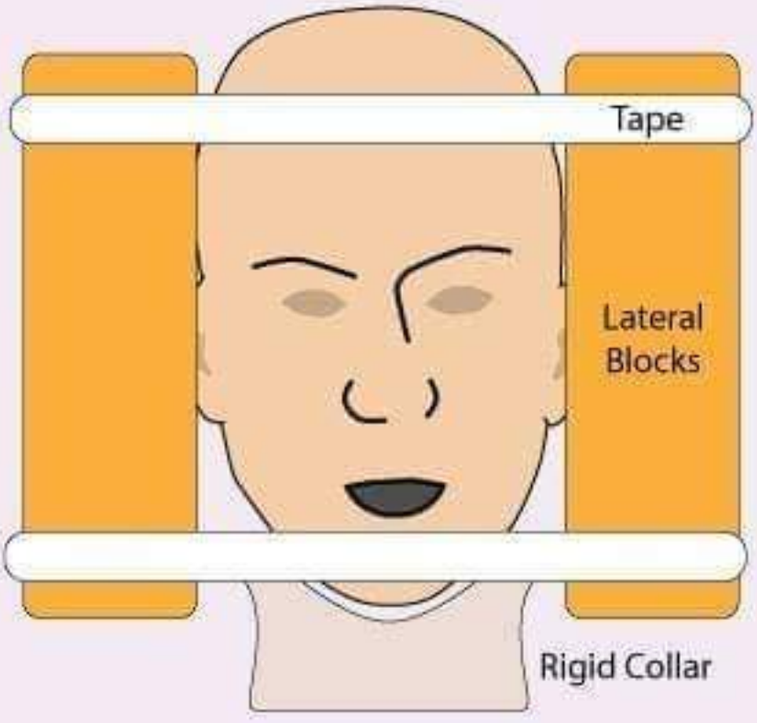
Protect the cervical spine at all times!

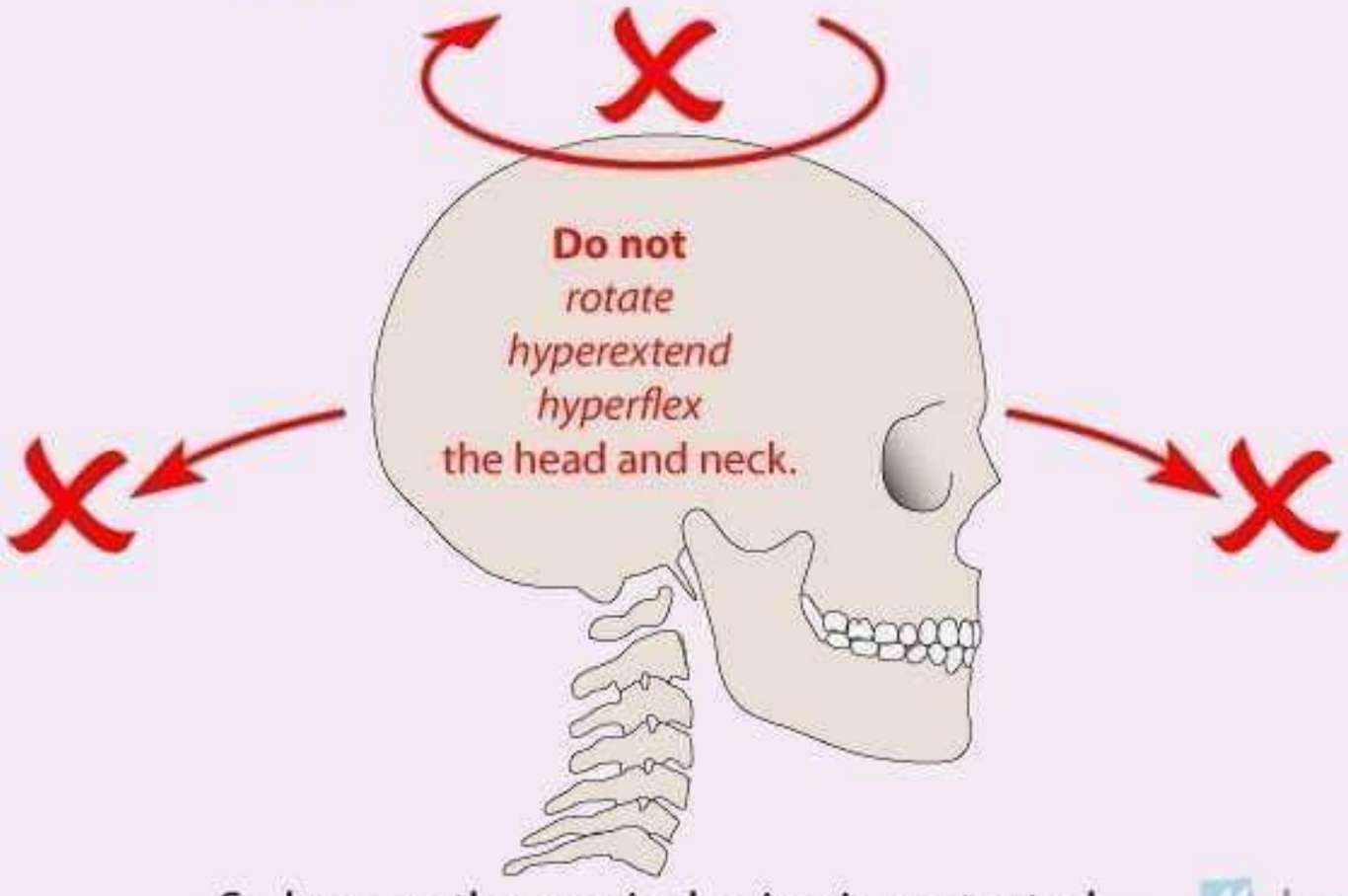
Either by manual stabilization or by using appropriate stabilization devices.

Manual Stabilisation



Stabilisation Devices





So long as the cervical spine is protected, further assessment and appropriate imaging can be obtained ***after*** potentially life-threatening injuries have been addressed.

Airway - Assesssment to ascertain patency

Protect the cervical Spine at all times!

Can the patient **communicate verbally?** *If so the airway is unlikely to be in immediate jeopardy.*

Hoarseness? ? laryngeal obstruction

Agitation
May indicate hypoxia

Cyanosis
Indicates hypoxaemia (inspect **nail-beds** & **circumoral skin**)

Obtundation
May suggest hypercarbia

GCS ≤ 8
usually requires a **definitive airway**

Noisy breathing?
Stridor? *(an airway emergency!*
Obstruction?
Use SUCTION
But take care not to push visible foreign bodies further in!

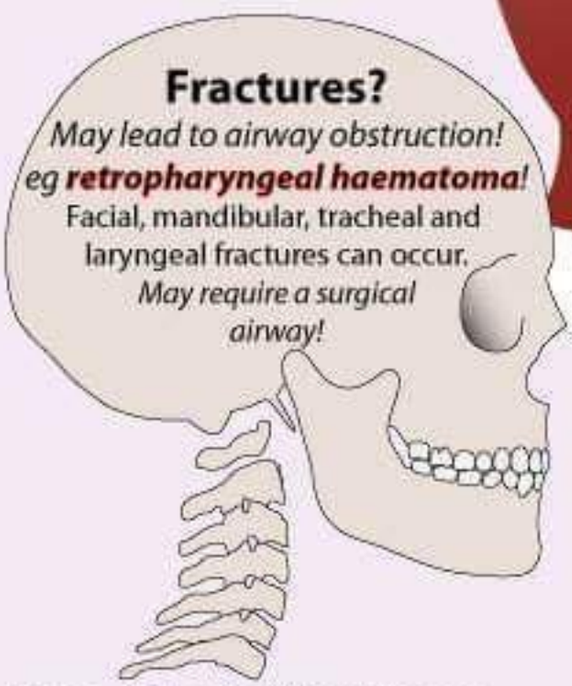
Snoring?
Gurgling?
Stridor?

Remember to **Reassess** frequently

Use of **accessory muscles** of ventilation e.g. sternocleidomastoid and scalene muscles

? Laryngeal Fracture
(sometimes **palpable**)
Can cause hoarseness & subcutaneous emphysema.

Fractures?
May lead to airway obstruction!
*eg **retropharyngeal haematoma**!*
Facial, mandibular, tracheal and laryngeal fractures can occur.
May require a surgical airway!



Check trachea
Is it in the midline?

Paediatic Airway
Has unique anatomical features!

Beware aspiration of gastric contents

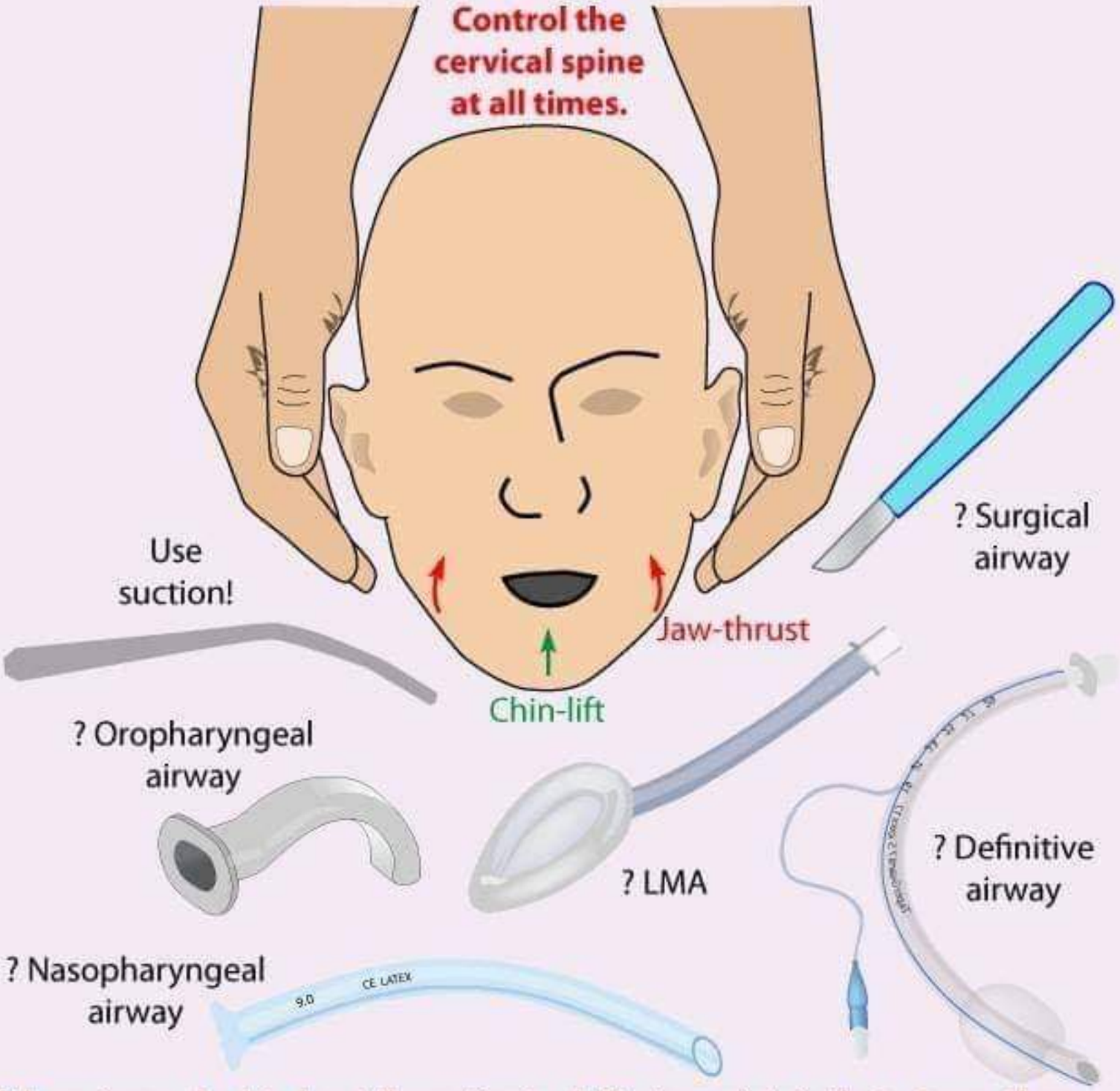
Note - Some facial fractures can cause an airway problem such that the patient refuses to lie down.

Airway - Management

Maintain the airway as a priority.

If unconscious (with no gag reflex) an oropharyngeal airway can be helpful.

GCS ≤ 8 usually requires a **definitive airway**



If there is any doubt about the patients ability to maintain their airway then a definitive airway (ie intubation) should be established.

The cervical spine should be controlled at all times however.

Colorimetry / capnography - can confirm correct endotracheal tube placement. (i.e. not in the oesophagus!)

Intubation & Ventilation in the unconscious patient can unmask or aggrevate a pneumothorax. The patients chest must therefore be re-evaluated.

Obtain CXR soon after intubation.

The causes of **Shortness of Breath**

Anaphylaxis

Central causes
(leading to
hypoventilation)

Anxiety
(hyperventilation)

H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+ H+

Metabolic
Acidosis

STRIDOR

Upper Airway Obstruction
often with STRIDOR

Asthma

Aspirin OD

Laryngeal
Fractures

Phrenic Nerve
lesion (C3,4,5)

Pulmonary
Fibrosis
"honeycomb
lung"

Pneumo
-thorax

Chronic
Obstructive
Lung Disease

Lung Cancer

Pulmonary
Embolus

Bronchiectasis

Kyphoscoliosis

Pulmonary
Collapse

Basal Atelectasis
Pneumonia

Heart
Failure

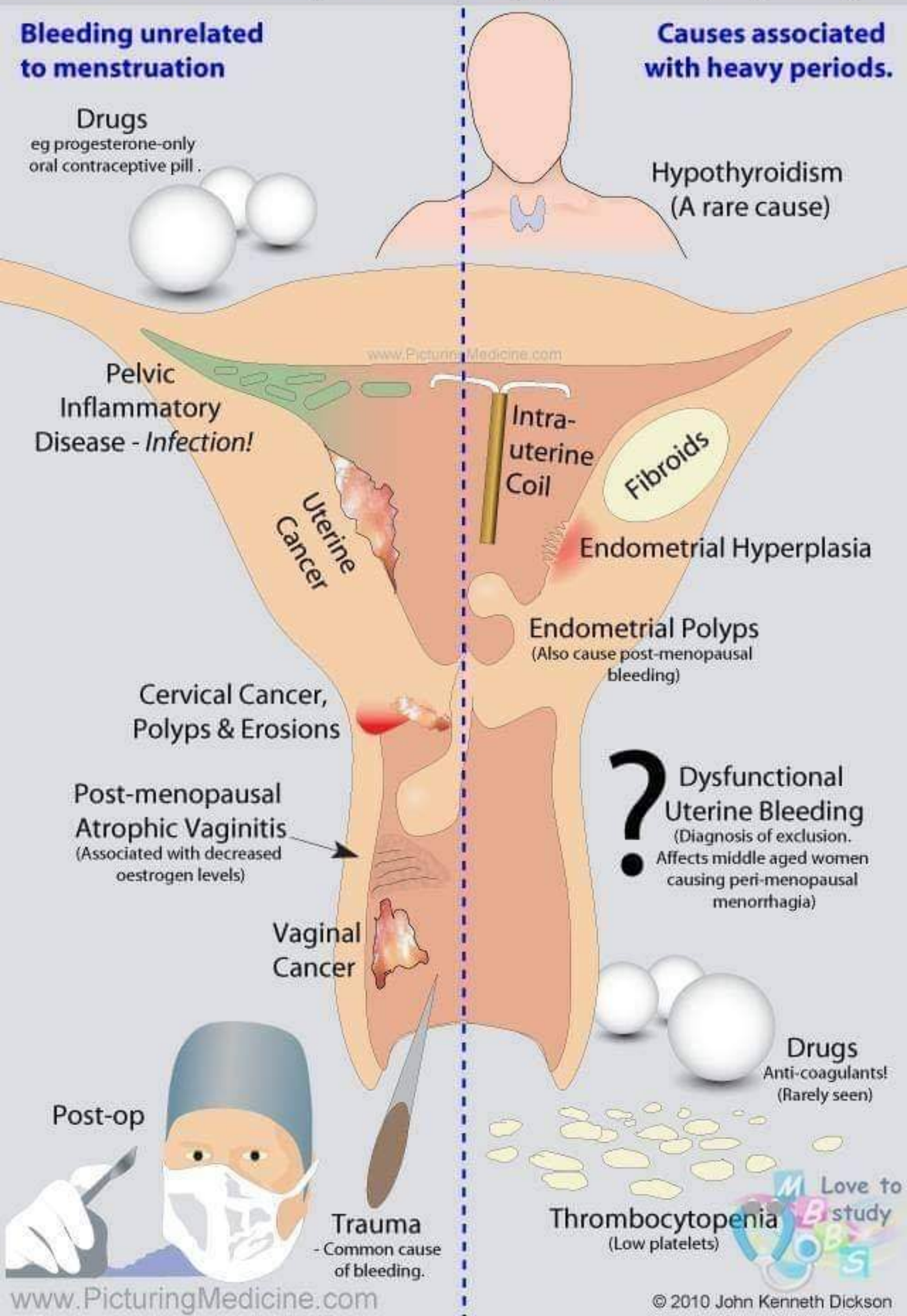
Myocardial
Infarction

Pleural
Effusion

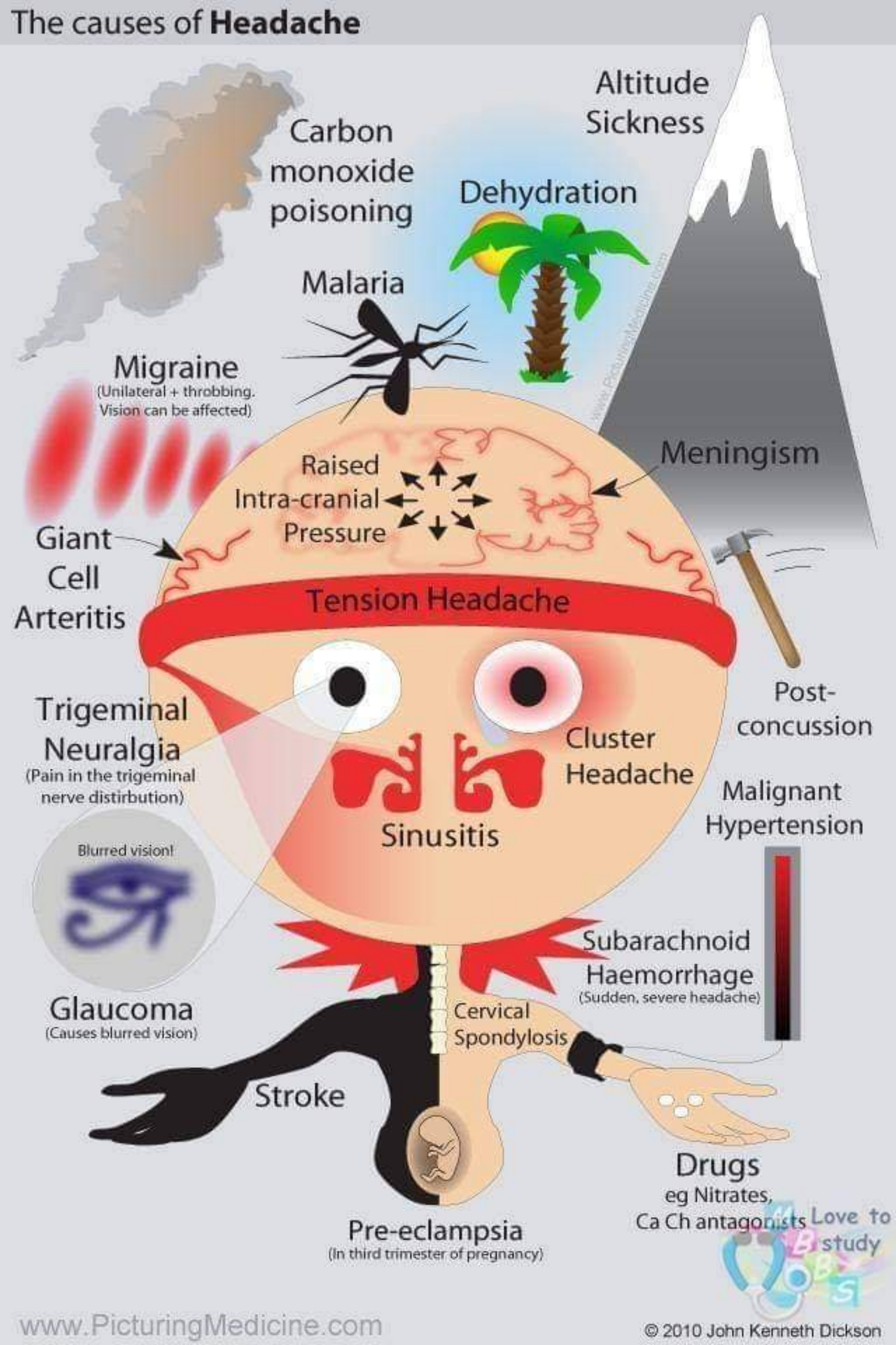
Arrhythmias

Diaphragmatic Weakness
eg Myasthenia Gravis, Gullain-Barre Syndrome
MS, Polio, MND, Muscular Dystrophy

The causes of **Per Vaginal Bleeding** (outside of pregnancy)

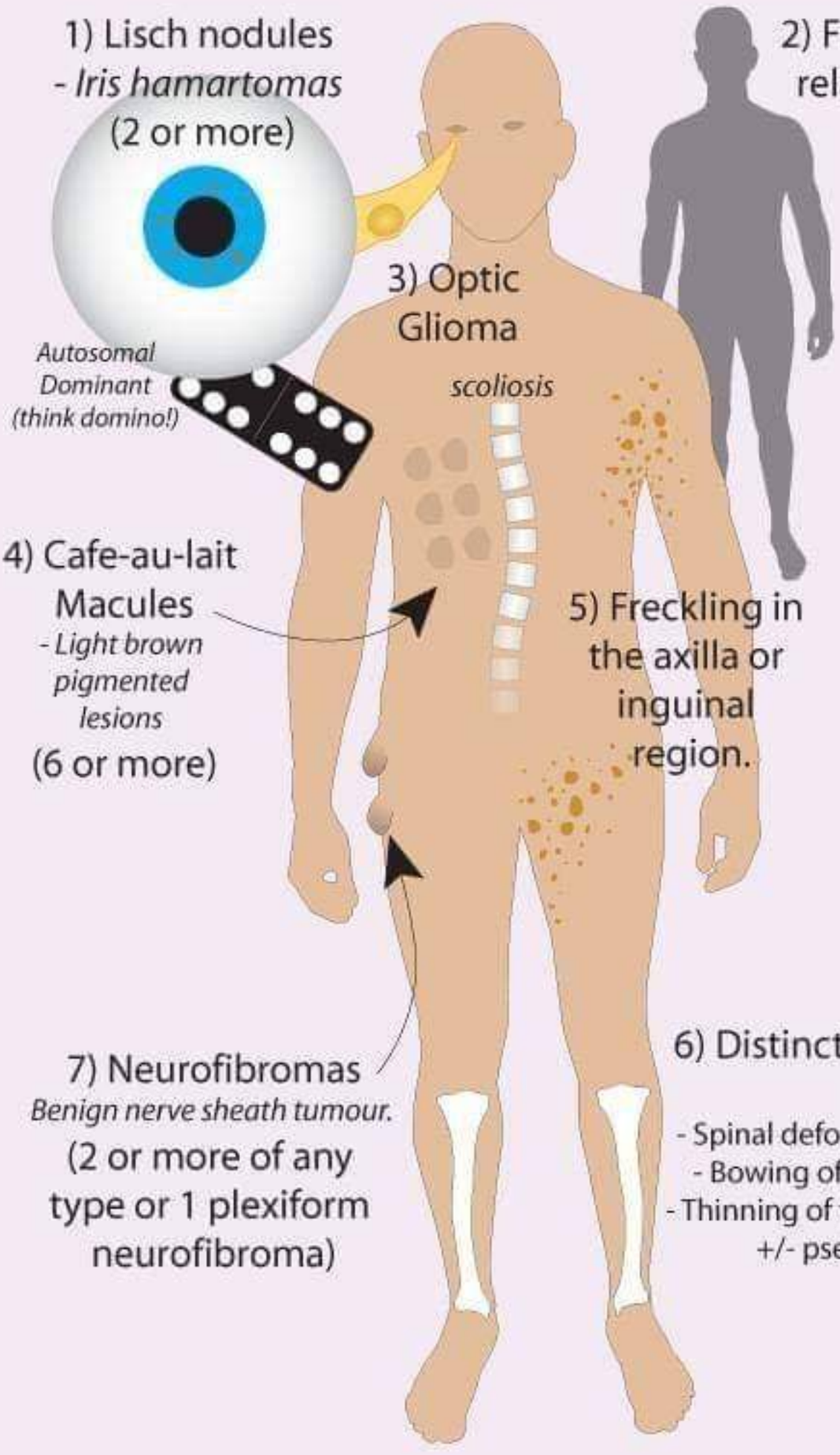


The causes of **Headache**



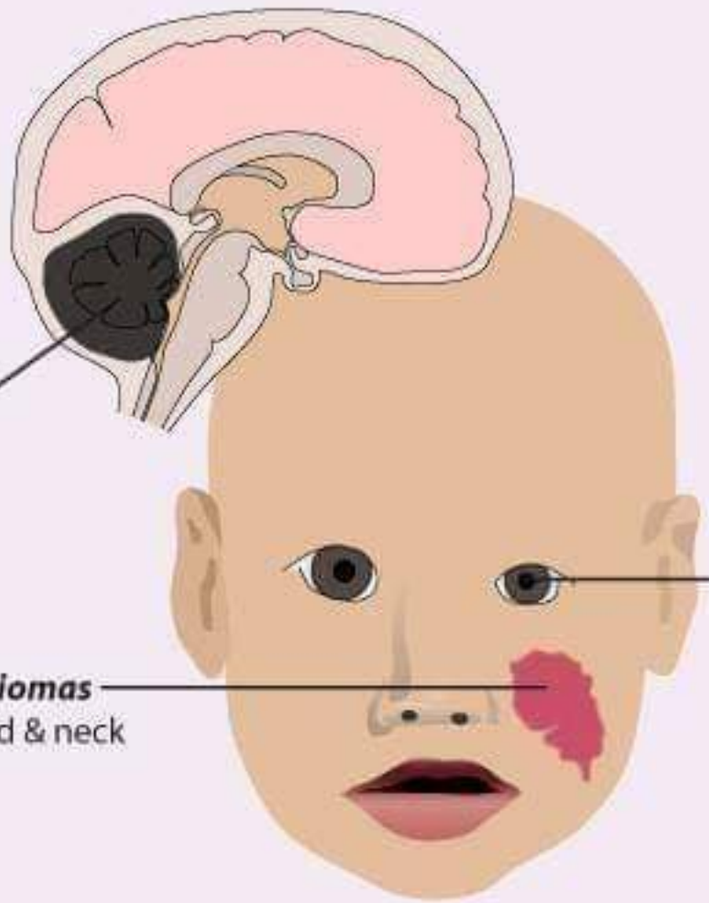
Von Recklinghausen's Syndrome - Neurofibromatosis Type 1 (NF1)

Hereditary multiple neurofibromas.
Autosomal dominant with high rate of penetrance.
The NF1 gene on chromosome 17 is affected.
Diagnostic Criteria - Two or more of the following...



PHACE / PHACES Syndrome

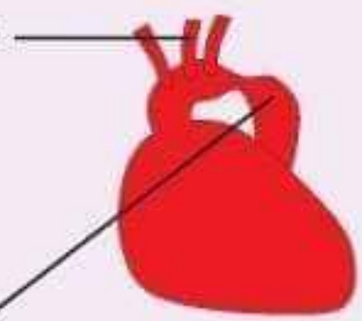
P = Posterior fossa abnormalities (affecting the cerebellum)
H = Haemangiomas of the head & neck
A = Arterial abnormalities (carotid/cerebral)
C = Cardiac Defects (eg coarctation of the aorta)
E = Eye abnormalities (underdeveloped optic nerve or small eye - microphthalmia)
Usually on the same side as the haemangioma.
S = Sternal defects (notch or absent sternum)



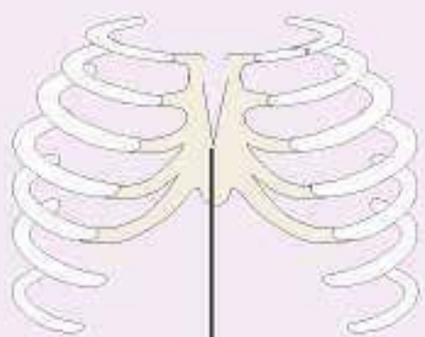
Posterior fossa abnormalities (affecting the cerebellum)

Haemangiomas of the head & neck

Eye abnormalities (underdeveloped optic nerve or small eye - microphthalmia) Usually on the same side as the haemangioma.



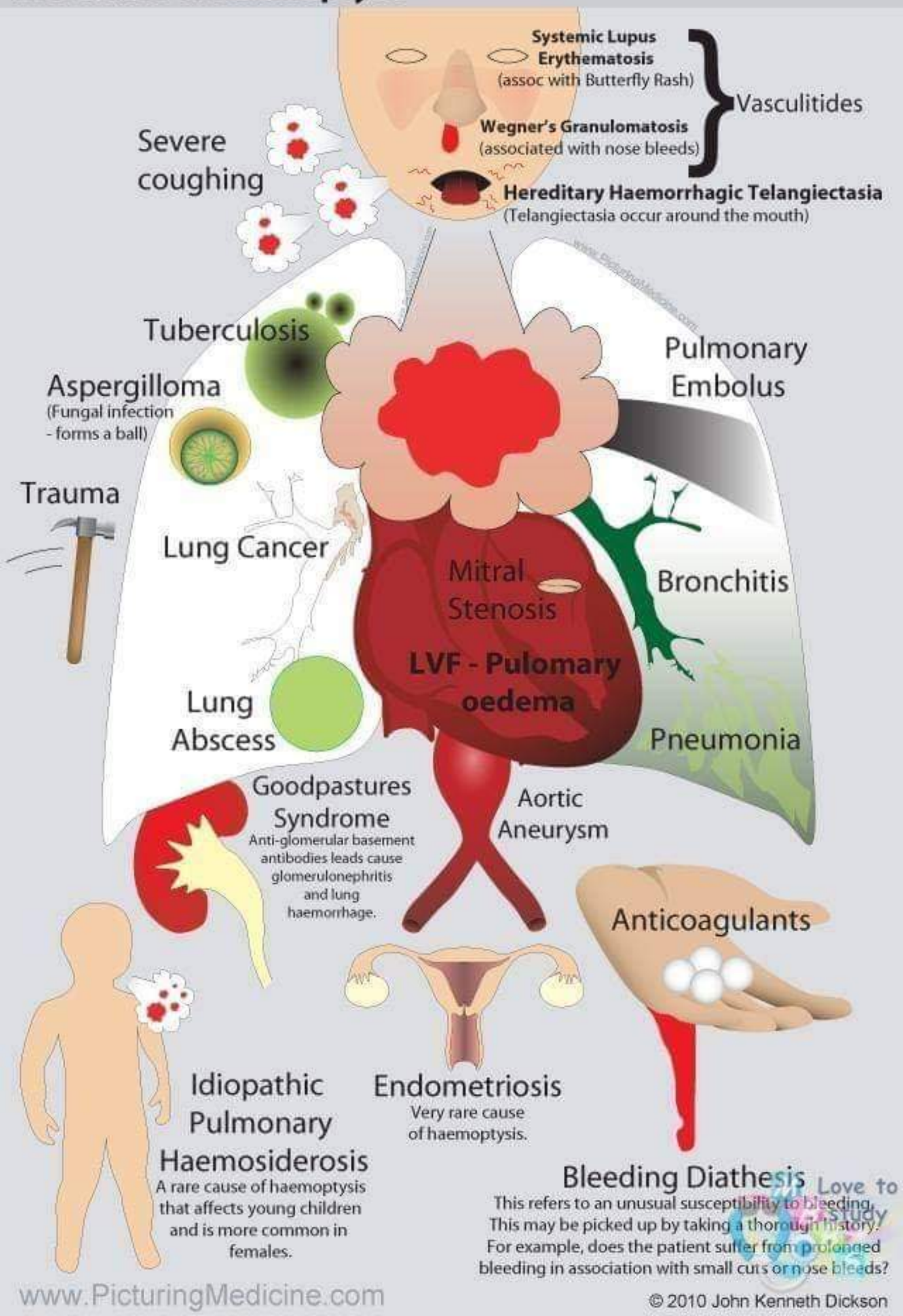
Arterial abnormalities (carotid/cerebral)



Cardiac Defects (eg coarctation of the aorta)

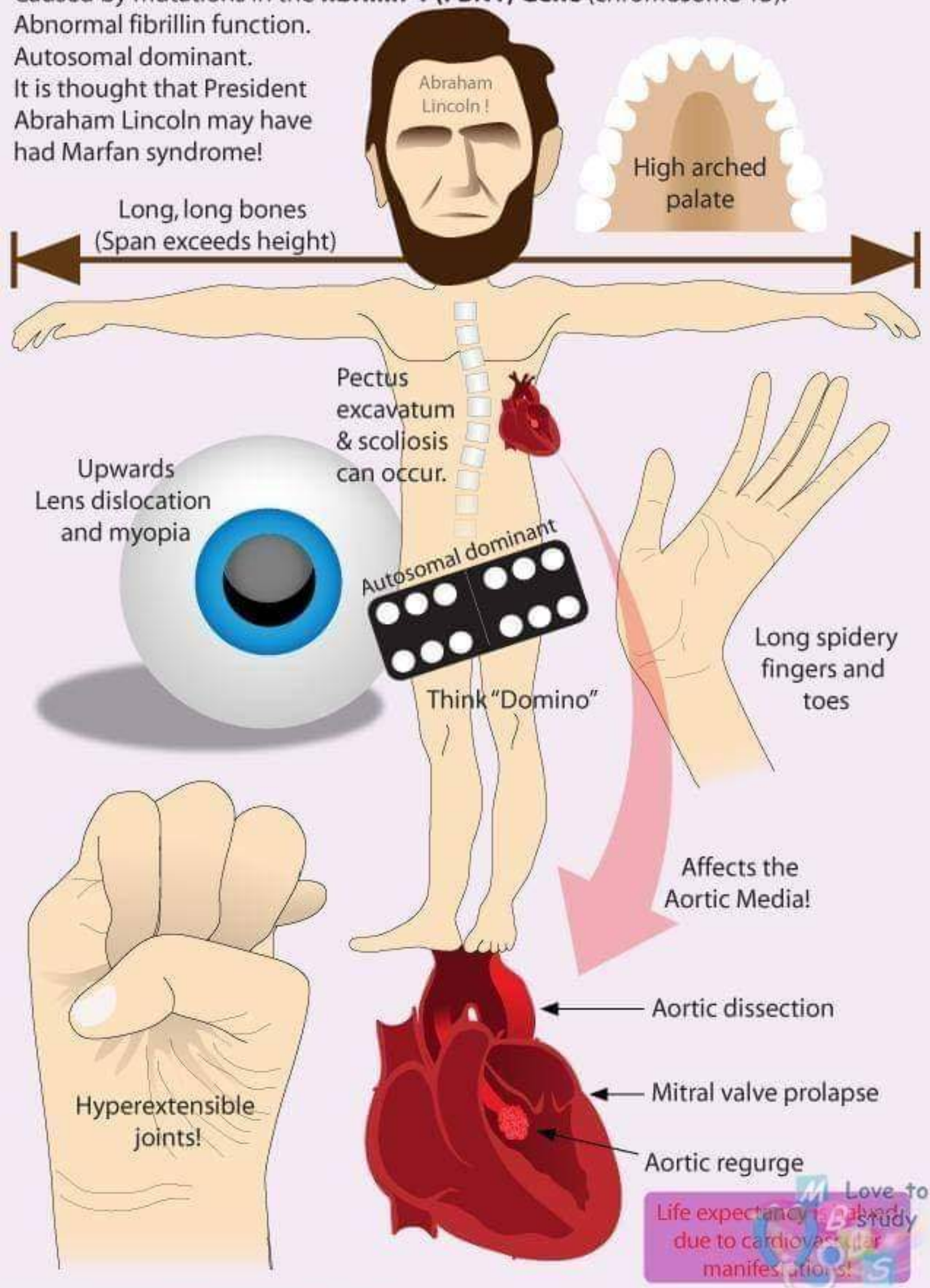
Sternal defects (notch or absent sternum)

The causes of **Haemoptysis**



Marfan Syndrome *(Connective Tissue Disorder)*

Caused by mutations in the **fibrillin-1 (FBN1) Gene** (chromosome 15).
Abnormal fibrillin function.
Autosomal dominant.
It is thought that President Abraham Lincoln may have had Marfan syndrome!

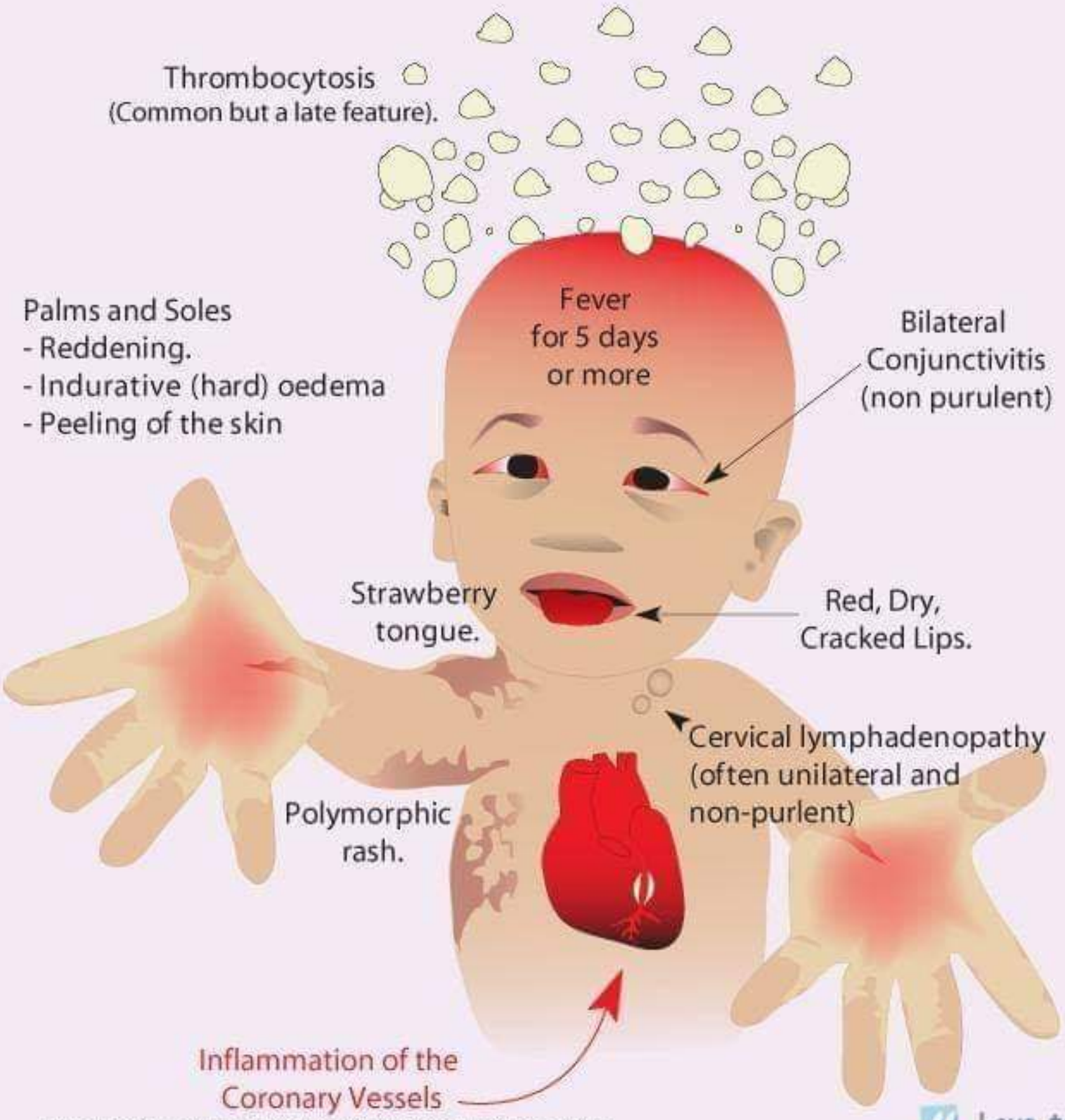


Life expectancy is reduced due to cardiovascular manifestations!

Love to study

Kawasaki Disease (Mucocutaneous Lymph Node Syndrome)

An rare systemic acute febrile vasculitic syndrome. Aetiology unknown.
Affects children between 6mths and 4yrs (peak at 1yr).
More common in asians.
Early diagnosis can prevent lethal cardiac complications.
Early administration of intravenous immunoglobulin reduces the risk of developing cardiac involvement.



This leads to aneurysms with subsequent scarring. Narrowing, ischaemia and even infarction can occur leading to sudden death!