NEWBORN SCREENING

Four diseases screened at newborn screening program

HYPOTHYROIDISM

CAH

PKU

G6PD

UNILATERAL PTOSIS

COMMON CAUSES

- ***Congenital ptosis***. Congenital ptosis is present at birth. It may be unilateral or bilateral, and varies in severity from mild to severe. Congenital ptosis may be familial or associated with an underlying syndrome, but is often an isolated finding in an otherwise healthy child.Severe congenital ptosis requires early repair due to the risk of amblyopia
- ***Third nerve palsy***. Patients with complete third nerve palsies usually have marked ptosis on the affected side, severe strabismus with the eye out and down, and a larger pupil on the affected side (although the pupil in some patients with congenital third nerve palsy may be smaller Severe ptosis from a third nerve palsy may cause amblyopia in young patients.The presence of an acquired third nerve palsy requires prompt evaluation
- ***Eyelid or orbital mass***. A large number of eyelid and orbital lesions may cause secondary ptosis. In most eyelid lesions, this is a mechanical effect due to the increased weight of the eyelids, and the etiology is obvious on examination. Early orbital lesions may cause ptosis without marked proptosis, and this possibility should be kept in mind in patients with acquired ptosis.
- ***Horner syndrome***. The ptosis in patients with Horner syndrome is usually mild to moderate. Patients have unequal pupils (smaller on the affected side), and may demonstrate decreased sweating of the brow on the affected side Horner syndrome itself does not cause vision problems. Its importance lies in possible associations with systemic diseases,such as neuroblastoma.

UNILATERAL EDEMA LOWER LIMB

Localized Edema

- Localized edema is a more common presenting complaint in pediatrics than generalized edema). Usually, these areas of localized swelling are caused by minor trauma, infection, or secondary to an allergic reaction. Historical factors and physical examination findings will often lead to a particular diagnosis without the need for further testing.
- Tenderness to palpation points to trauma or infection, while fever, erythema, and overlying warmth more commonly occur with an infectious cause. On the face and distal extremities, insect bites may produce swelling and warmth, which can be difficult to distinguish from cellulitis.
- A therapeutic response to an oral antihistamine or to an intramuscular dose of epinephrine can help to differentiate an allergic reaction from other causes of localized swelling

Causes of Edema

Localized

Allergic reaction

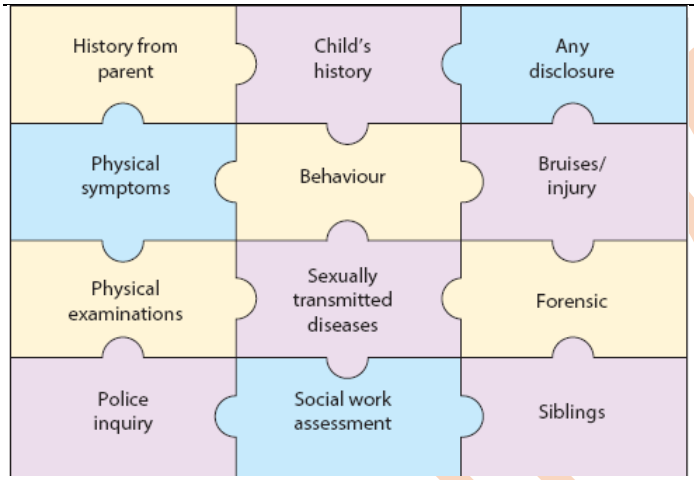
Cellulitis
Trauma
Dependent edema from immobility of extremity
Congenital lymphatic abnormality

Life-Threatening Causes of Edema

Localized

Allergic reaction
venous thrombosis
Pit viper envenomations
Thrombophlebitis

GENITAL INJURY IN A 3 YR OLD



Recognition

The child or young person may:
• tell someone about the abuse
• be identified in pornographic material
• be pregnant (by legal definition this is due to sexual abuse for a girl under the age of 13 years)
• have a sexually transmitted infection with no clear explanation (but some sexually transmitted infections can be passed from the mother to the baby during pregnancy or birth).

Physical symptoms

• Vaginal bleeding, itching, discharge
• Rectal bleeding.

Behavioural symptoms

• Soiling, secondary enuresis
• Self-harm, aggressive or sexualised behaviours, regression, poor school performance.

Signs

- There are few clearly diagnostic signs of sexual abuse on examination. This is because sexual abuse of children often comprises touching or kissing or other activities that do not involve significant physical force. Furthermore, the genital area heals very quickly in young children,so signs may be absent even a few days after significant trauma. Forensic material also decays rapidly

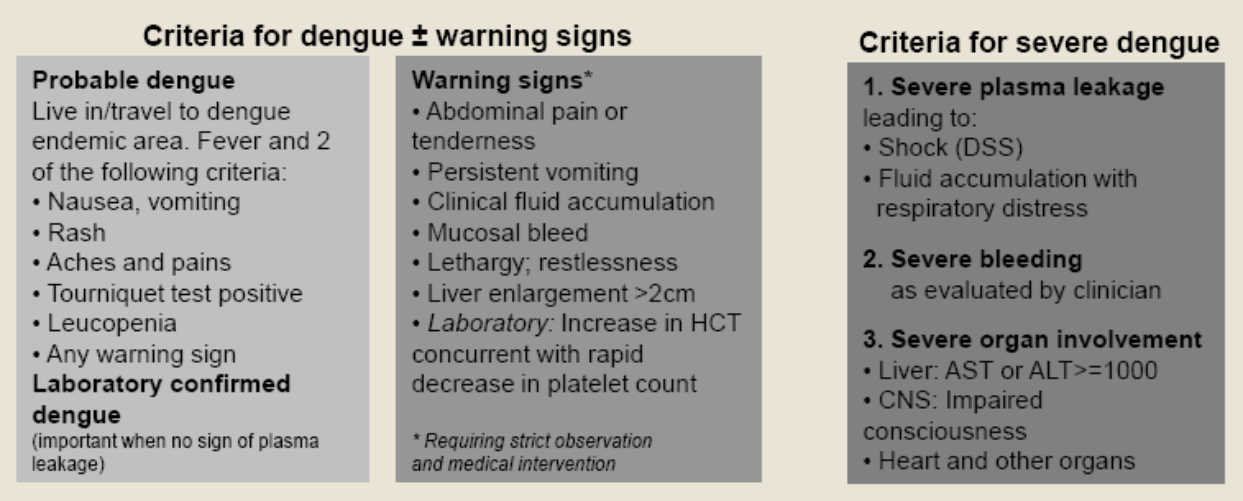
- Examination of children suspected of having been sexually abused requires a doctor with specific expertise and training, facilities for photographic documentation, sexually transmitted infection screening and management and, where indicated, forensic testing (Forensic testing of swabs from the child or his/her clothing/bedding may reveal DNA from the sperm of the perpetrator

Management

- Abused children may present to doctors in the hospital or to medical or nursing staff in the community. They may also be brought for a medical opinion by social services or the police. In all cases, the procedures of the local safeguarding children board should be followed.
- The medical consultation should be the same as for any medical condition, with a detailed history and full examination. It is usually most productive when this is conducted in a sensitive and concerned way without being accusatory or condemning.
- Any injuries or medical findings should be carefully noted, measured, recorded and drawn on a body map and photographed (with consent). The height, weight and head circumference(where appropriate) should be recorded and plotted on a centile chart. The interaction between the child and parents should be noted.
- All notes must be meticulous, dated, timed and signed on each page. Treatment of specific injuries should be instigated and blood tests and X-rays undertaken

DENGUE SHOCK SYNDROME

Current terminology



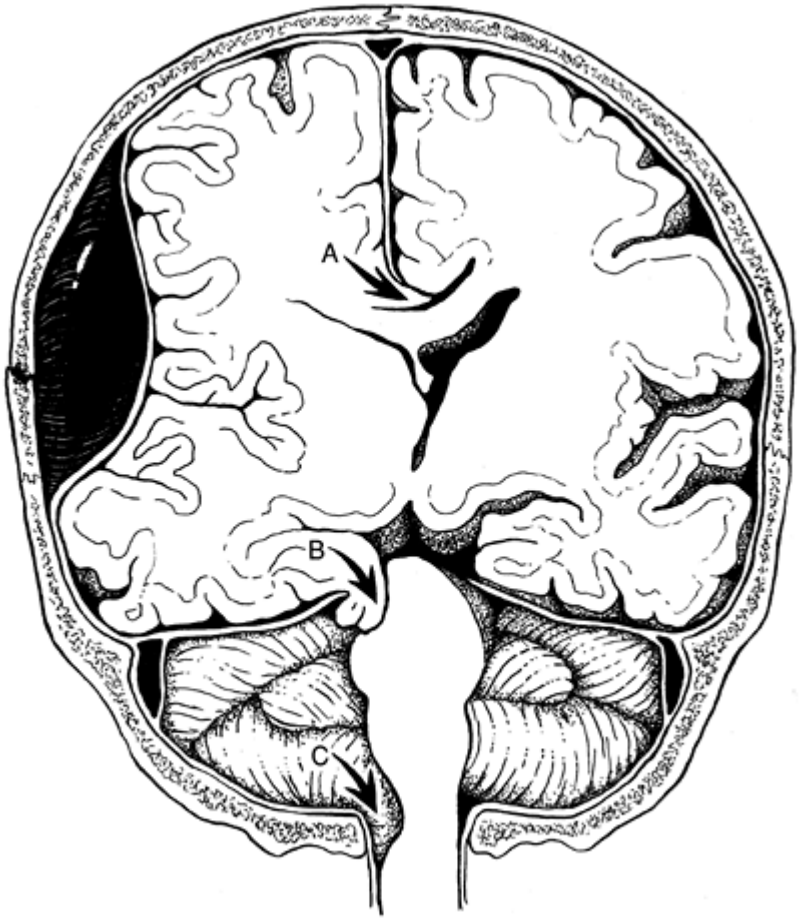
CLINICAL FEATURES OF INTRACRANIAL HYPERTENSION

Increased intracranial pressure

- An increase in the volume of any intracranial compartment (blood, CSF, or parenchyma) can cause an elevation in intracranial pressure (ICP) and is a true neurosurgical emergency.Head trauma is the most common cause of increased ICP in children.
- Although the onset of increased ICP is usually acute, onset may be delayed in patients with subdural or epiduralhematomas. Other causes are brain tumor, meningitis, hemorrhage from a vascular malformation,intracerebral abscess, pseudotumor cerebri, and shunt obstruction in a patientwith hydrocephalus. Intracranial hypertension is also the major life-threatening complication of Reye's syndrome.

Clinical presentation

- Lethargy is an important finding in a patient with increased ICP. A patient with a GCS <9 or a falling GCS after head trauma requires an immediate evaluation for intracranial hypertension.A patient with either a history of head injury or increased ICP on a nontraumatic basis may complain of early-morning headache and vomiting, or headaches of recent onset that have become more frequent and severe. A verbal child may complain of blurry vision, frank diplopia, or intermittent loss of vision. Altered mental status, a change in personality (constant crying or irritability in an infant), neck pain, and a head tilt are also suggestive of increased ICP. Papilledema is a highly specific but insensitive finding within the first 24-hours of acutely increased ICP.
- In an infant, increased ICP causes irritability, vomiting, widening of the sutures, increasing head circumference, a full or bulging fontanelle, and possibly “sunsetting” of the eyes.
- Clinical signs of imminent cerebral herniation include a deteriorating level of consciousness, unequal pupils (the dilated pupil is usually on the same side as the herniation), asymmetric EOMs, and decorticate (flexor-early) or decerebrate (extensor-late) posturing.
- Abnormal respirations, bradycardia, and hypertension (Cushing’s triad) occur with severely increased ICP.



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- Herniation syndromes: (A) subfalcine and cingulate, (B) uncal, (C) foramen magnum (see Table 51.4). Subfalcine herniation occurs when one cerebral hemisphere is displaced under the falx cerebri across the midline (A). Uncal herniation refers to displacement of supratentorial structures inferiorly under the tentorium cerebelli, causing distortion and compression of the

blood supply to infratentorial structures (B). Downward herniation of the cerebellum causes compression of the brainstem (C).

Diagnosis

- Any patient who has suffered significant head trauma is at risk for increased ICP. Always ask about persistent vomiting or visual or behavioral changes. Check the pupillary responses, EOMs, and level of consciousness. Perform a careful ophthalmoscopic exam and a sensory and motor exam, comparing one side of the body to the other. Coma can be due to metabolic causes, but typically the pupils are equal and reactive and there are no focal findings.
- Congenital anisocoria, the use of mydriatic drops, or traumatic mydriasis or iritis can cause pupillary inequality. These are always diagnoses of exclusion in a patient with lethargy or any other signs suggesting increased ICP.
- Morning headaches or headaches that are increasing in frequency and intensity are worrisome. However, headaches in an alert patient with no other abnormal neurologic findings are most likely to be psychogenic, tension, or migraine headaches