Welcome to the ninth issue, a neonatal themed edition of 'The PEN' – the Paediatric Education Newsletter. This is a monthly educational bulletin of cases, clinical questions and learning points from recent teaching.

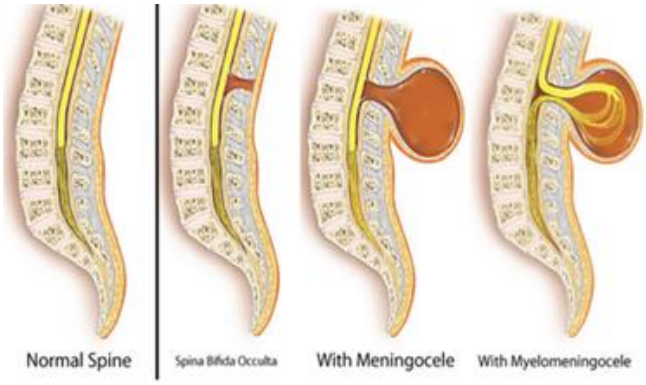
Spinal Dysraphism and sacral dimples

A baby boy was noted to have a small sacral lesion at birth. There were no antenatal concerns on scan. Mum was found to have anti-Ro antibodies 33 weeks (presented with urticaria). Folic acid started at 8 week's gestation.
OE: A 1x1cm non-discharging, circular, ulcerated, midline lesion was noted in the lumbosacral area, covered by a white membrane. There was an underlying bone defect. A benign sacral dimple with visible base was also present to the right of the midline. Neurological examination and ECG were normal. The baby was transferred to SCH for neurosurgical intervention following confirmation of a complex lipomyelomeningocele on MRI scan.

Cutaneous markers of spinal dysraphism:

- Pigmented or hairy patch
- Abnormal skin texture
- Cyst or lipoma
- Skin tag
- Haemangioma
- Swelling

Present in 50-80% Occult spinal dysraphism



Neurological signs (lower limbs):

- Abnormal tone
- Abnormal power
- Abnormal deep tendon reflexes
- Patulous anus
- Abnormal anocutaneous reflex
- Lower limb deformities

Radiology Findings:
Depend on type and severity of condition. See Radiopaedia Section on Spinal Dysraphism.

Any concerns:
Discuss with the Registrar or Consultant before requesting imaging.

When can we reassure parents?

Normal Neurological examination
+
Typical, midline lesion <5mm in size
+
Situated ≤ 2.5cm from anus
+
Base of dimple visualised

Learning points from teaching:

- **Persistent pulmonary hypertension (PPHN):** Delay in decline in pulmonary vascular resistance causes shunting of deoxygenated blood into the systemic circulation. Most common in term infants; seen in sepsis, meconium aspiration and lung hypoplasia. Large difference in pre and post-ductal oxygen saturations.
  Management: 100% O2, keep pH >7.3, maintain good BP, HFOV, Nitric Oxide, ECMO if severe (high oxygenation index)

- **ITP/'Easy bruising':** When presented with a child with 'easy bruising' safeguarding concerns must be considered. Look for red flags in history & examination. Non-accidental injury and bleeding disorders are not mutually exclusive. Children with ITP should be well with no red flag features. There should be no organomegaly or lymphadenopathy and their FBC (except plt), film and clotting should be normal. Discuss with haematology if any concerns about malignancy, pancytopenia, isolated thrombocytopenia > 3 months, severe bleeding, or thrombocytopenia with other unusual features.

Doncaster and Bassetlaw Paediatric Department
The PEN Issue No. 9 January 2021

Doncaster and Bassetlaw Teaching Hospitals NHS Foundation Trust

Congenital CMV

Triggers for cCMV screening:

- Intracranial ventriculomegaly
- Calcification on cranial USS (periventricular)
- Congenital cataracts
- Failed newborn hearing screening
- Petechiae/purpura with thrombocytopenia
- Unexplained thrombocytopenia
- Hepatosplenomegaly
- Prolonged jaundice with transaminitis
- Primary maternal CMV infection in pregnancy
- All babies of HIV positive mothers

Confirmatory tests:

- **CMV PCR saliva swab in first 21 days** (take 2hr after feeding if breastfed, no restriction if formula fed)
- CMV PCR urine in first 21 days (bag or cotton wool)
- 2 independent samples preferred to confirm diagnosis

If PCR positive:

Refer to paediatric infectious disease consultant at Sheffield Children's Hospital

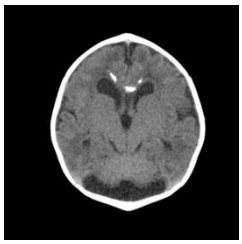
Document evidence of congenital CMV:

- Clinical features – hepatosplenomegaly, jaundice, petechiae/purpura, microcephaly, hypotonia, cataracts, retinitis, hemiparesis, seizures, unilateral/bilateral deafness
- Prematurity & IUGR

Bloods – FBC, clotting, U&Es, LFTs, CMV viral load by PCR

Radiology – Cranial ultrasound & MRI brain (do not delay treatment whilst awaiting MRI)

Referrals – ophthalmology, audiology & neurology

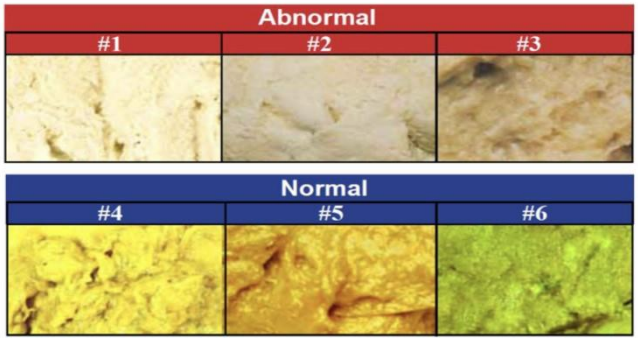


Treatment & follow up:

- Decision made by ID team after discussion with parents in moderate-severe disease
- **Valganciclovir PO/NG 16mg/kg twice daily**
- Ganciclovir IV 6mg/kg twice daily (if significantly unwell/not enterally fed)
- Complete 6 months of antiviral treatment with frequent monitoring
- Follow up – Paediatric ID, audiology, ophthalmology & neurodevelopmental assessment at 1y

Prolonged Jaundice

A 21-day-old female neonate presents with persistent jaundice. Uneventful delivery at term. Her stool appeared clay coloured. On examination she appears jaundiced with 2cm palpable liver.



1. What is your top differential?

A. Breast milk/physiological jaundice
B. Sepsis
C. Biliary Atresia
D. Crigler-Najjar syndrome
E. Galactosemia

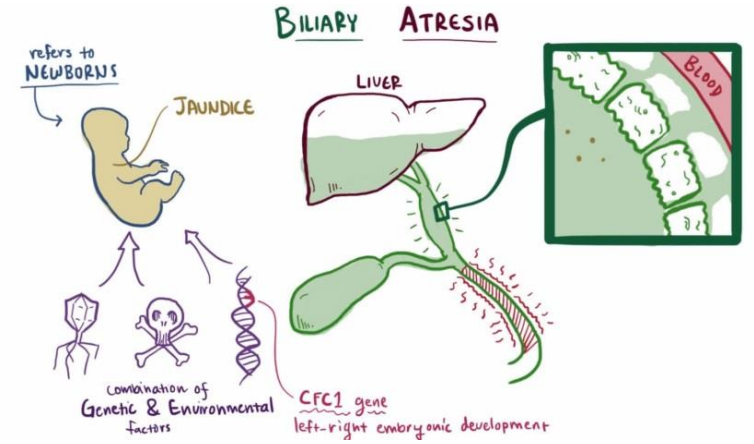
2. What investigation would definitively prove your diagnosis?

A. Bloods tests (including prolonged jaundice screen)
B. Liver USS
C. Liver Biopsy
D. Stool MC&S
E. HIDA scan (hepatobiliary scintigraphy)

3. Once confirmed which of the following would be the most appropriate next step in management?
A. Start glucocorticoids and ursodeoxycholic acid
B. Choledochoduodenostomy
C. Hepatoportoenterostomy
D. Begin evaluation for liver transplantation

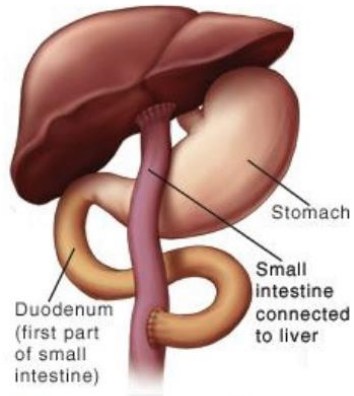
Answers:

1. Biliary Atresia



- Biliary Atresia is a childhood disease in which one or more bile ducts are abnormally narrow, blocked or absent meaning conjugated bilirubin produced by the liver cannot be excreted
- Presents with progressive conjugated jaundice, pale white stool and dark urine
- Bleeding abnormalities can occur due to fat-soluble vitamin malabsorption (Vit K)
- Conjugated bilirubin is unable to cross blood-brain barrier so low risk of kernicterus
- Will progress to liver cirrhosis and portal hypertension if untreated
- Always consider sepsis in jaundiced babies presenting acutely unwell

2. E
- Although blood tests, liver USS +/- biopsy will be done to rule out other causes, the definite diagnosis a HIDA scan. Injection of radioactive isotope with imaging to determine uptake into the liver and measure excretion into the bowel. In biliary atresia there is good hepatic uptake but no excretion into the bowel. Exploratory surgery may be performed if diagnosis remains unclear.



During the Kasai procedure, the intestine is attached to the liver. This allows bile to drain.

3. C
- Also known as a Kasai procedure is the preferred treatment (bile ducts removed + loop of jejunum reattached to liver to allow bile drainage)
- Glucocorticoids and ursodeoxycholic acid are often given postoperatively but limited evidence of benefit
- A liver transplant may be required if the Kasai procedure fails. Early diagnosis of biliary atresia improves the success rate of the Kasai procedure. Prognosis is best if done before 8 weeks of age.

Resources:
Dynamed Spina Bifida
BMJBP Spina bifida and neural tube defects
Fifteen-minute consultation: diagnosis and management of congenital CMV | ADC Education & Practice Edition
Dynamed Biliary Atresia
BMJBP Biliary atresia

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