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Doncaster and Bassetlaw Teaching Hospitals
NHS Foundation Trust

THE PEN

Paediatric Education Newsletter

The Baby Issue

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POSTNATAL WARD REVIEW

CLOTTING DISORDERS

You are asked to review two different babies on the postnatal ward whose mums have clotting disorders.
Baby 1's Mum has Glanzmann's thrombasthenia.
Baby 2's Mum has factor XI deficiency.

The midwives asked if the babies can have Vitamin K and if they need any investigations, treatment or follow up?
(ANSWER ON PAGE 2)

PAED ALERT

IT'S IN THE GENES

Case 1

You are informed by the midwife that a term baby has just been delivered via SVD to a mother with Cleidocranial dysplasia. She says there is a Paed Alerts and the baby requires a review. What will you be looking for on review?
(ANSWER ON PAGE 4)

Case 2

You are performing a NIPE for a baby who has a Paed Alert as the father has Muenke Syndrome. What are your next steps?
(ANSWER ON PAGE 4)

COMMON QUESTIONS ON CLOTTING DISORDERS

How can you find out what you should do for these babies?

- If a mother is a known carrier for a bleeding disorder there should be an **antenatal/delivery plan** agreed between haematology and obstetrics. This should be available in the maternal notes.
- If a baby is expected to be affected by a severe bleeding disorder they should be born in a tertiary centre.
- Mum should be seen in joint haematology/obstetric clinic antenatally.
- ODN neonatal network guideline on haemophilia: https://www.networks.nhs.uk/nhs-networks/yorkshire-humber-neonatal-odn/guidelines-1/guidelines-new/haematological/haemophilia

What do I do if there is bleeding?

Any bleeding e.g. cephalhematoma should be discussed with haematology at SCH as they may need clotting factor replacement.

A cranial USS to look for intraventricular haemorrhage is needed for all babies who have a moderate/severe bleeding disorder (this is not usually indicated for type 2 von Willebrand disease). Any abnormalities should be discussed with the haematologist and an MRI should be considered.

Discharge planning - What happens next?

Babies with bleeding disorders (or where one has not been ruled out) need haematology follow up- the haematology team will decide at what point they need to be seen (e.g. 1 year of age).

Parents and the GP should be informed that they will need subcut rather than IM immunisations.

Mild bleeding disorders

Haemophilia A with factor VIII > 0.05iu/ml
Haemophilia B with factor IX > 0.05iu/ml
Female carriers of haemophilia A or B
Type 1 von Willebrand disease
Factor XI deficiency
Mild deficiencies of other clotting factors II, V, VII, X, XIII

Should you do any blood tests for the baby?

If the baby could be affected by a severe bleeding disorder (see below) they should have cord bloods sent (coagulation screen, factor assays, FBC, glycoproteins – depending on the type of disorder suspected). This is not required in mild bleeding disorders.

Therefore baby 1 should have cord bloods for FBC to check platelets and for glycoproteins (especially if Dad is also a carrier or his status is unknown as it is an autosomal recessive condition). Baby 2 does not need his cord bloods doing.

Can the babies have vitamin K?

Vitamin K should be given orally rather than IV if a moderate/severe bleeding disorder is suspected. Therefore baby 1 should have oral vitamin K however baby 2 may be safe to have IM vitamin K as long as the baby is not severely affected (rare in factor XI deficiency).

Need for prophylactic treatment?

Severe deficiencies require clotting factor prophylaxis if there are risk factors:

§ Prematurity <36 weeks

§ Instrumental delivery

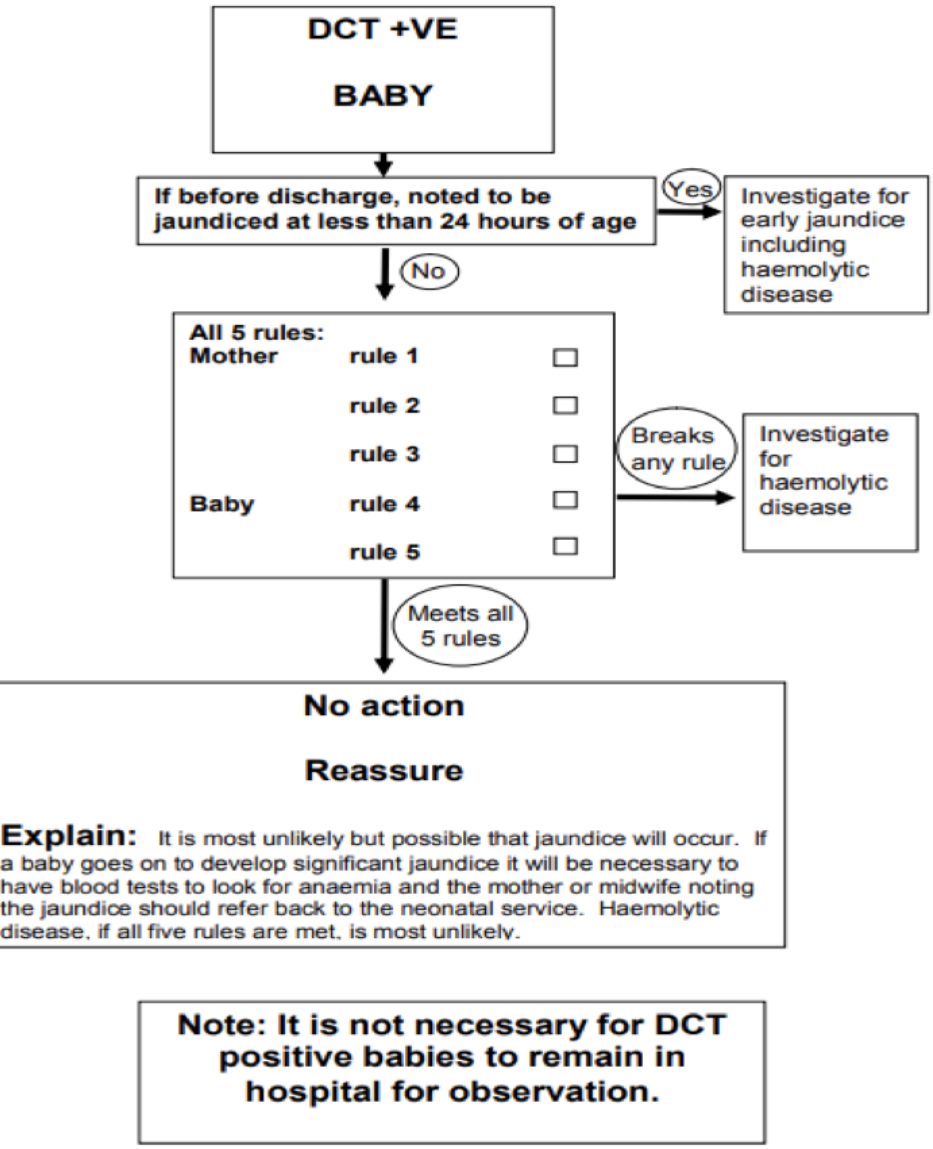
§ Prolonged 2nd stage of labour (e.g. pushing for >2 hrs in primigravida or >1 hour in parous women).

§ Superficial cranial bruising

Moderate/severe bleeding disorders

Haemophilia A with factor VIII < 0.05iu/ml
Haemophilia B with factor IX <0.05iu/ml
Type 2 and 3 von Willebrand disease
Rare severe congenital clotting factor deficiency of II, V, VII, X, XIII
Severe platelet function defects e.g. Glanzmann's thrombasthenia and Bernard Soulier Syndrome.

QUICK RECAP ON DCT



Note: It is not necessary for DCT positive babies to remain in hospital for observation.

Passive immunisation with anti-D globulin during any pregnancy will remove any Rhesus D positive cells from the fetus without the mother showing an active immunological response. The anti-D administered crosses the placenta and if the fetus is Rhesus positive, small amounts will attach to the fetal red blood cells. This does not lead to fetal haemolytic disease, but it will give a positive Coombs' test in the newborn infant. This can make interpretation of a positive Coombs' test difficult.

Most infants with a positive Coombs' test have no risk of haemolysis! It is simply the result of passive maternal immunisation. **Few** may have haemolytic disease so it is important to make an assessment, to determine if the baby's positive DCT is the result of maternal passive immunisation or maternal alloimmunisation to Rhesus D antigen or other red blood cell antigens.

Baby with a positive DAT is most unlikely to have haemolytic disease if :

5 GOLDEN RULES

1. Mother has had anti-D antibody passive immunisation before birth.

2. Mother does not have a rising titre of anti-D antibody during pregnancy.

3. Mother has no antibody titre against other red cell antigens (eg, c, E, Kell etc)

4. Baby is not jaundiced or has jaundice controlled by single phototherapy at more than 24 hours of age. [Jaundice requiring phototherapy within the first 24 hours is assumed to be pathological and may be due to haemolysis.]

5. Baby is not clinically anaemic or has normal haemoglobin on testing.

If all 5 rules are met then no investigation is required, reassurance can be given and we can explain that it is most unlikely but possible that jaundice will occur.

If a baby goes on to develop severe jaundice, it will be necessary to have blood tests- FBC with blood film, reticulocyte and split bilirubin. If needed, discuss with haematologist

APPROACH TO NEONATAL ALLOIMMUNE THROMBOCYTOPENIA

Platelet version of haemolytic disease of the newborn. Unlike Rhesus disease, NAIT can occur in the first pregnancy, due to the early expression of platelet antigens. The thrombocytopenia can be severe and result in intracranial haemorrhage (ICH) and fetal death.

There is platelet incompatibility between the parents, leading to trans-placental transfer of platelet antibodies to the paternal HPA antigens and thrombocytopenia. Common antibody: HPA-1a, HPA-5B and HPA-15b.

Diagnosis

NAIT should be suspected in a term infant with a platelet count <50x10 9/l.

Investigations

Blood for platelet genotyping and antibody testing for both parents and baby.

Cranial USS - Risk of ICH

Treatment

While waiting for the results, the infant should be treated as if they have this condition due to the high risk of ICH.

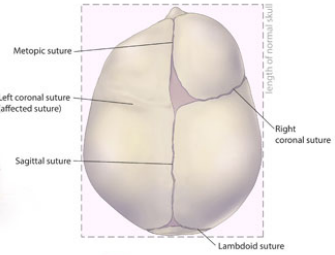
Aim

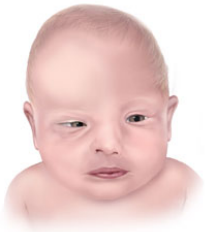
Platelet count > 50 if there is any bleeding
Platelet count > 25 if otherwise well with no IVH.

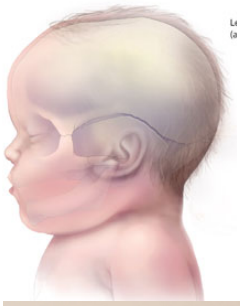
High dose steroids and immunoglobulins given to the mother seem to reduce the need for the very high-risk in-utero transfusions of platelets.

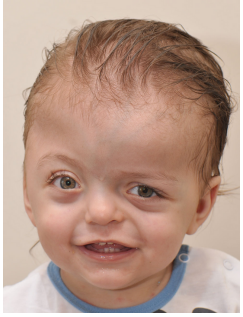
Platelet transfusion with HPA 1a and 5b negative platelets.
If an emergency random donor platelets may be used.

GENETICS

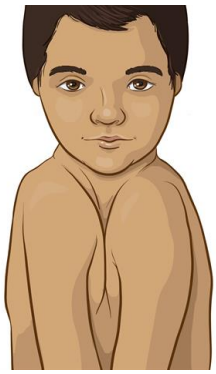


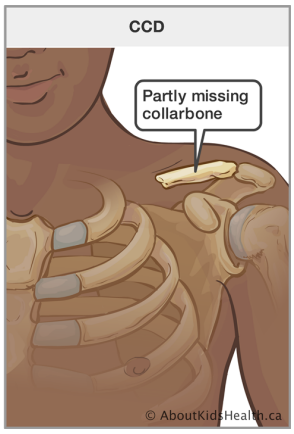


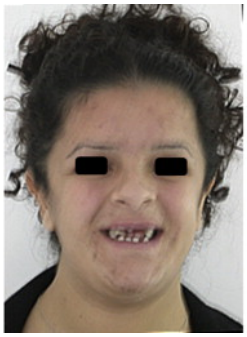




MUENKE SYNDROME	CLEIDOCRANIAL DYSPLASIA
AUTOSOMAL DOMINANT INHERITANCE May be sporadic	
Fibroblast Growth Factor Receptor 3 (FGFR3) gene	*RUNX2* gene
DIAGNOSIS	
Do not always have Clinical Features Mainstay is genetic testing	Clinical Features Imaging Genetic Testing
CLINICAL FEATURES	
If present Macrocephaly with no fused sutures Early fusion of coronal sutures leading to misshapen skull Short fingers and toes Webbing of toes Short stature normal growth velocity	Delayed closure or large of fontanelles Early fusion of coronal sutures Underdevelopment/short clavicles Bone abnormalities in the hands Abnormal teeth - delayed eruption/extra teeth Osteopenia Osteoporosis Scoliosis Delayed Growth & short stature Prominent forehead Prominent chin Maxillary hypoplasia Coxa vara Genu valgum High-arch palate/ cleft palate
INVESTIGATIONS	
Blood for genetic testing Hearing test - both can have hearing loss X-rays CrUSS if misshapen/early fusion of sutures	
DISCHARGE PLANNING	
General Paediatric follow up **Genetics referral and follow up** **(Most started during the antenatal period and require re-referral post delivery)**	
LONG TERM OUTCOMES Normal Development Good prognosis ** Cleidocranial dysplasia have an increased risk for recurrent ear and sinus infections, and upper respiratory complications. **	







ADDITIONAL READING & RESOURCES

CLINICAL REPORT.

Identifying the Misshapen Head: Craniosynostosis and Related Disorders

Mark S. Dias, MD, FAAP, FAANS,a Thomas Samson, MD, FAAP,b Elias B. Rizk, MD, FAAP, FAANS,a Lance S. Governale, MD, FAAP, FAANS,c Joan T. Richtsmeier, PhD,d

A detailed clinical report that reviews the characteristic head shape changes, as well as secondary craniofacial characteristics, that occur in the setting of the various primary craniosynostosis and deformations. It encompasses the physiology and genetics of suture growth, pathophysiology of craniosynostosis, description of the types as well as the syndromes associated. Identifying the various types of head shape abnormalities is important for aesthetics, to identify candidates for future monitoring, and in some, to prevent increases in intracranial pressure (ICP) and allow proper brain development.

EVIDENCE BASED SUMMARIES

In-Depth information from Online Mendelian Inheritance in Man (OMIM) on

- Cleidocranial dysplasia.
- Muenke Syndrome

FOR MORE LIBRARY RESOURCES CLICK HERE

USEFUL RESOURCES FOR PARENTS

Headlines

A charity that provides information and support for people with craniosynostosis and their families.
http://www.headlines.org.uk/

The Haemophilia Society
Glanzmann's Thrombasthenia & Factor XI Deficiency

Offers patient information and support via Facebook groups for individual conditions.
https://haemophilia.org.uk/

CLICK HERE FOR MORE INFORMATION ON SUPPORT GROUPS FOR PATIENTS AND CARERS