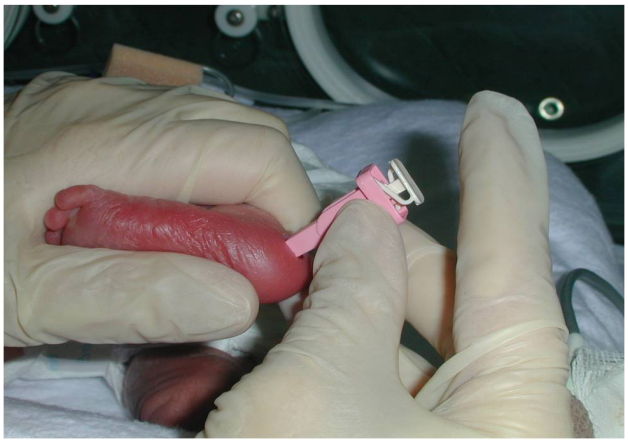
SCREENING THE NEONATE

FACT FILE 2E (b)

In 2008, the Child Health Promotion Programme (CHPP; DoH, 2008), an update of standard one and two of the National Service Framework for Children, Young People and Maternity Services (DFES, 2004), was introduced. This is a programme of information and guidance to support parenting and healthy choices, immunisations, and screening – all necessary services for children and families in order to achieve their optimum health and well being (Davies and Elliman, 2008).

In addition, the UK National Screening Committee set out guidance and recommendations for newborn screening in specific areas which will be the focus of this unit. Refer to the following link to see the timeline of screening as a whole over the pregnancy and newborn period.

http://www.healthscotland.com/uploads/documents/12614-PregnancyAndNewbornTimeline.pdf



Newborn Bloodspot screening
Neonatal screening for phenylketonuria (PKU) using blood from a heel prick onto a card was first undertaken in the 1960's. The programme became nationwide in 1969/70 with congenital hypothyroidism added in 1982, and more recently sickle cell disorders, Medium Chain Acyl Coenzyme A Dehydrogenase Deficiency (MCADD) and Cystic Fibrosis.

Phenylketonuria is an autosomal recessive genetic disorder detected by high levels of phenylalanine in the blood. If it is not detected and treated then metabolites can accumulate and cause brain damage. Treatment is a special diet low in phenylalanine.

Congenital hypothyroidism (CH) is a condition of thyroid hormone deficiency present at birth. The cause is either a problem with thyroid gland development or of a genetic origin. Hypothyroidism can lead to developmental delay if it is not treated with thyroxin supplements.

PKU and congenital hypothyroidism are conditions, which, if untreated, can result in significant developmental delay.

Sickle cell disease including beta thalassaemia are haemoglobinopathies. The conditions affect the normal oxygen carrying capacity of red blood cells. The symptoms can include severe anaemia, intense pain, damage to major organs and infections. Although there is no routine cure for sickle cell, patients can be supported to manage their pain, and regular monitoring can help to avoid life threatening complications such as stroke.

Medium Chain Acyl Coenzyme A Dehydrogenase Deficiency (MCADD) is an autosomal recessive disorder that results from the lack of an enzyme required to metabolise fat into energy. If the child is unable to break down fats fast, the accumulated medium chain fats form toxic metabolites, which can lead to serious life threatening symptoms and even death. The treatment for MCADD is to prevent low blood sugars particularly during illness or fasting.

Cystic Fibrosis (CF) is another autosomal recessive disorder which affects the exocrine glands and the body's ability to move salt and water in and out of cells. This causes the lungs and pancreas to secrete abnormally thick mucus that can blocks the airway and prevents proper function. The accumulation of mucus can also impair the pancreas and intestine.

HIV; Surveillance can be undertaken for maternal Human Immune deficiency virus (HIV) infection based on anonymous testing of spare blood spots allowing monitoring of disease frequency in a defined population. This has been approved by ethics committees and funded by the Department of Health. This is not diagnosis in individual cases.

In other areas / countries, some screening laboratories also include tests for other conditions which are at present not recommended for universal testing in the UK – e.g. galactosaemia and other amino acid disorders

Taking the test
Newborn Bloodspot Screening tests should be carried out on, or after, the 5th day of birth, taking the day of birth as zero (ideally days 5-8). In the community, the blood sample is taken by the midwife or health visitor, placed on the special Bloodspot Screening card and sent to the local neonatal screening laboratory.

Following a positive test:
When a positive screening test is obtained, a firm diagnosis can only be made after further investigation. The infant is referred to a specialist centre for investigation, counselling and clinical management. The bloodspot screening website has parents information leaflets on what the results mean for the child and what further follow up is required.

Once diagnosis is made, treatment should be started as soon as possible. It is recommended that treatment be initiated by 21days or as early as possible before symptoms and subsequent damage occurs. This is particularly important in improving the outcome for children with PKU and hypothyroidism.


Image source: http://newbornbloodspot.screening.nhs.uk/
Used with kind permission

Hearing screening in the Newborn: Hearing screening is carried out in all newborns as part of the UK Hearing Screening Programme. Done usually before weeks 4-6, the aim is to identify any hearing loss or difficulty that could go onto affect language acquisition in the developing infant / child. In turn, intervention is given as soon as possible to prevent a delay in this vital area of development.

Physical Examination; Examination of the Newborn: The UK National Screening Committee (and in 2004 the Child Health Promotion programme) set the context for the examination of the newborn as do the NICE guidelines for Post-natal care (See Reading list). The Newborn and Infant Physical examination (NIPE) document focuses on pathways, standards and competencies for screening. After a baby is born an initial physical examination should be carried out. Parents are then offered a more detailed physical examination carried out, ideally, within the first 24 hours of birth, and certainly within 72 hours, to detect conditions that may need early treatment. It is repeated at the end of the postnatal period.

The procedure is outlined as follows ….
Review the health history of the family, woman and baby and address any parental concerns. The physical assessment should include the following:

- appearance, including colour, breathing, behaviour
- activity and posture
- head (including fontanelles), face, nose,
- mouth including palate, ears, neck and general
- symmetry of head and facial features. Note head circumference
- eyes; check opacities and 'red reflex'
- neck and clavicles, limbs, hands, feet and digits;
- assess proportions and symmetry
- heart; check position, rate, rhythm and sounds, murmurs and femoral pulse volume
- lungs; check effort, rate and sounds
- abdomen; check shape and palpate to identify any organomegaly
- umbilical cord
- genitalia and anus; check completeness and patency and undescended testes in males
- spine; palpate bony structures and check integrity of skin
- skin; note colour and texture as well as birthmarks or rashes
- central nervous system; check tone, behaviour, movements and posture, and elicit reflexes only if concerned
- hips; check symmetry of limbs and skin folds; perform Barlow and Ortolani's manoeuvres
- cry; note sound
- weight; note.

Appropriate recommendations are made by the NHS National Screening Committee (www.nsc.nhs.uk/ch_screen/child_ind.htm) for all areas of screening including both antenatal and postnatal tests.

KEY READING

WEBSITES / DIRECT LINKS....

Screening Timeline
http://www.healthscotland.com/uploads/documents/12614-PregnancyAndNewbornTimeline.pdf

NEWBORN BLOODSPOT SCREENING
http://newbornbloodspot.screening.nhs.uk/

http://www.screening.nhs.uk/annbpublications

NEWBORN PHYSICAL EXAMINATION SCREENING
http://www.screening.nhs.uk/newborninfantphysical-england
http://newbornphysical.screening.nhs.uk/
http://www.infantgrapevine.co.uk/journal_article.html?RecordNumber=5752&number=22
http://www.infantgrapevine.co.uk/pdf/inf_022_ens.pdf
http://newbornphysical.screening.nhs.uk/publications

NICE Postnatal Guideline (includes examination of the newborn)
http://www.nice.org.uk/CG037

NHS Hearing Screening
http://hearing.screening.nhs.uk/
http://www.ndcs.org.uk/family_support/newborn_hearing_screening/index.html

CDH-
http://www.screening.nhs.uk/hipdislocation

HIV SCREENING
http://www.infantgrapevine.co.uk/journal_article.html?RecordNumber=5623&number=13
http://www.infantgrapevine.co.uk/pdf/inf_013_msk.pdf

SICKLE CELL
http://www.screening.nhs.uk/sct-england

Screening for Fetal Anomalies – Map of Medicine
http://healthguides.mapofmedicine.com/choices/map/fetal_anomaly_screening1.html

Downs Syndrome screening – Map of Medicine
http://healthguides.mapofmedicine.com/choices/map/down_s_syndrome_screening1.html

NHS SCREENING CONTINUOUS PROFESSIONAL DEVELOPMENT WEBSITE
http://cpd.screening.nhs.uk/programme-specific

ONLINE / E-LEARNING MODULES IN SCREENJNG (includes antenatal and newborn screening, Group b Strep, Health knowledge, Midwifery competencies, PEGASUS (Sickle cell and Thalassaemia) and Newborn Physical Examination) .....REQUIRES free registration to access modules) http://cpd.screening.nhs.uk/elearning

RESOURCE CARDS FOR SCREENING – ANTENATAL AND NEONATAL

http://cpd.screening.nhs.uk/resource-cards

CHIMAT LINK TO SCREENING TIMELINE

http://www.chimat.org.uk/resource/item.aspx?RID=88294

http://www.patient.co.uk/doctor/Newborn-Screening.htm

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