Resource cards for midwives to support the National Screening Committee antenatal and newborn screening programmes

Local contact details

Please place sticker here

Test Performance

- Detection Rate (DR) – the proportion of affected individuals who will be identified by the screening test

- False Positive Rate (FPR) – the proportion of unaffected individuals with a higher risk/screen positive result

- False Negative Rate (FNR) - The proportion of affected individuals with a low risk/screen negative result.

Illustration of test performance

6650 women with lower risk result

3 women carrying a baby with Down's syndrome

350 women with a higher risk result

7 women carrying a baby with Down's syndrome

Test performance illustration explained

- The diagram represents a screening test with a 70% detection rate and 5% false positive rate
- 7000 women have been screened
- The pale green figures represent the 6650 women identified as having a lower risk result
- The dark green figures within the pale green/lower risk group represent 3 women identified as having a lower risk result but who are carrying a baby with Down's syndrome
- The pale pink figures represent the 350 women identified as having a higher risk result (i.e 5% of the population)
- The dark pink figures within the pale pink/higher risk group represent 7 women with a pregnancy affected by Down's syndrome (i.e 1:50 identified as higher risk are actually carrying an affected baby).

Reframing Risk

Chance of an affected pregnancy		Chance of an unaffected pregnancy	
1 in 4	25%	3 in 4	75%
1 in 5	20%	4 in 5	80%
1 in 10	10%	9 in 10	90%
1 in 20	5%	19 in 20	95%
1 in 30	3%	29 in 30	97%
1 in 50	2%	49 in 50	98%
1 in 100	1%	99 in 100	99%
1 in 200	0.5%	199 in 200	99.5%

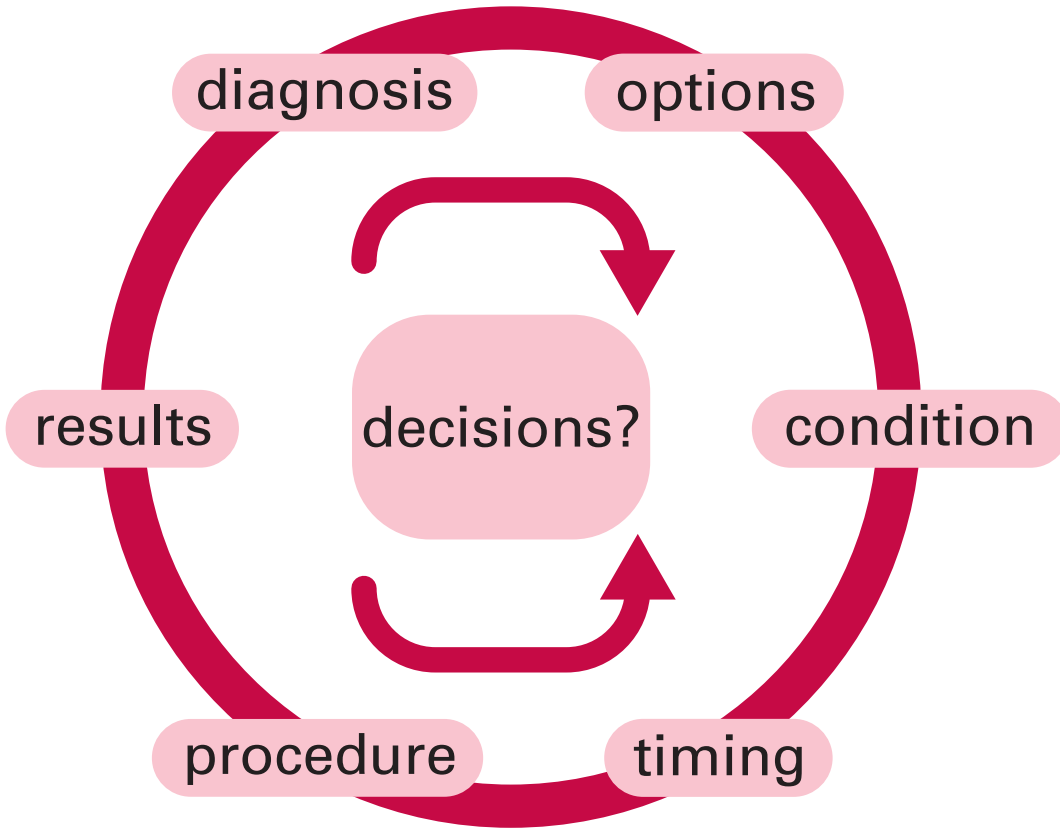
Can be applied to any screening test where the result is reported as a probability

Pre-test information - Screening

- Determine woman's knowledge of the conditions being screened for
- Explain that screening is optional
- Check understanding of screening versus diagnosis
- Describe screening test offered locally, how it is done, timing of test
- Explain the likelihood of a 'higher or lower risk' (e.g. Down's), positive, negative or an equivocal result (e.g. infectious diseases)
- Explain and agree how and when results will be given
- Explain confirmatory/repeat testing may occasionally be required
- Discuss meaning and implications of the possible test result
- Discuss the possibility that screening can provide information about other conditions
- Document decision.

Refer to cycle on following card

Pre-test Information Cycle



This depicts a revolving cycle of information giving. Information flows both ways, the starting & finishing point is dependent upon where the individual enters the cycle. A woman can choose at any point to exit the cycle.

Taking the blood sample

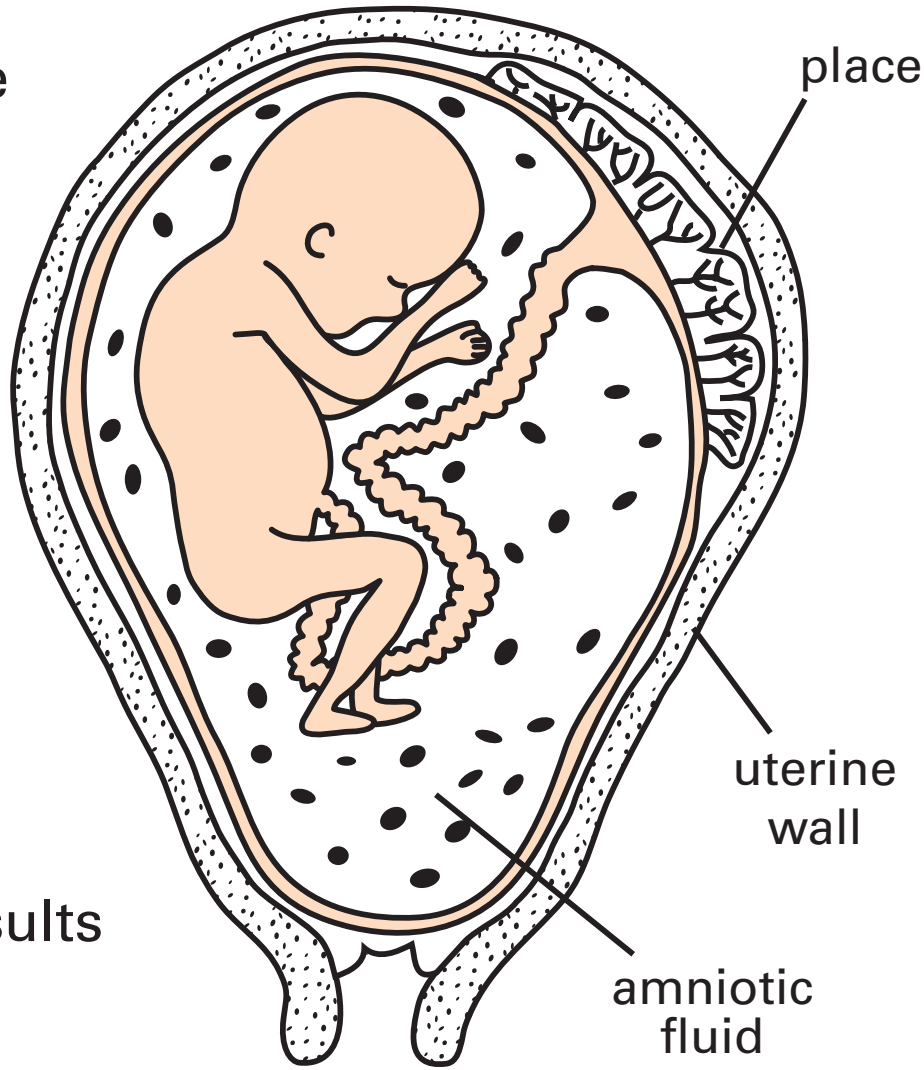
- If taking more than one blood sample at the same time, please take the Down's screening blood first as contamination from the EDTA* in the other vacutainers can affect the result
- Ensure the blood sample is in the correct bottle as per local policy
- Check all personal details are correct with the woman, the request form is fully completed and the sample bottle correctly labelled at time of sample taking
- Delay in sending samples to the laboratory can lead to an inaccurate result.

*Ethylene Diamine Tetra-Acetic Acid (EDTA) is an additive found in some blood bottles e.g. full blood count bottles.

Pre-test information- Diagnostic tests

- Nature & timing of procedure
- Accuracy of results
- Risk of miscarriage
- Limitations of the test
- Possibility of detecting other abnormalities
- Possibility of repeat test
- Care after procedure
- Length of time to results and method of reporting
- Discuss options following results

Please refer to local policy



Neural Tube Defects (NTDs)

- Generic term used to describe anencephaly and spina bifida
- Occurs very early in embryonic life (4-7 weeks)
- Incidence 1-2 per 1000 births
- The effect depends on the size and position of the lesion, 80% of fetuses with spina bifida develop hydrocephalus
- Ultrasound can diagnose up to 99% of anencephaly and up to 90% of open spina bifida
- 400mcg of folic acid is recommended daily from 12 weeks preconception to 12 weeks of pregnancy
- 5mg of folic acid is prescribed for women who have had a previous affected pregnancy or who are on anticonvulsants.

Down's Syndrome

- Genetic condition – presence of three copies of chromosome 21 (trisomy 21)
- Birth prevalence is 1 per 600-800 births
- People with Down's syndrome have learning difficulties but there is a wide spectrum just like there is a wide spectrum of abilities in the general population
- Does not usually run in families, less than 5% are hereditary
- Incidence increases with increasing maternal age
- Individuals with Down's syndrome may also have congenital heart defects (50%), hearing impairment (50%), epilepsy, thyroid problems and Alzheimer's disease
- Life expectancy 50-55 years.

Age related risks for Down's Syndrome

Maternal Age	Risk at term	Maternal Age	Risk at term
20	1:1527	33	1:547
21	1:1507	34	1:446
22	1:1482	35	1:356
23	1:1448	36	1:280
24	1:1406	37	1:218
25	1:1352	38	1:167
26	1:1286	39	1:128
27	1:1206	40	1:97
28	1:1113	41	1:73
29	1:1008	42	1:55
30	1:895	43	1:41
31	1:776	44	1:30
32	1:659	45	1:23

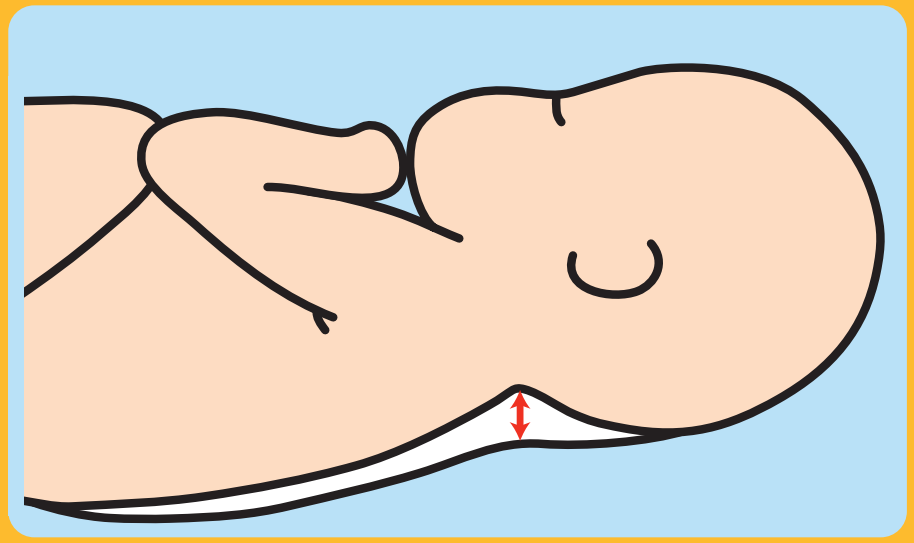
Cuckle et al, 1987

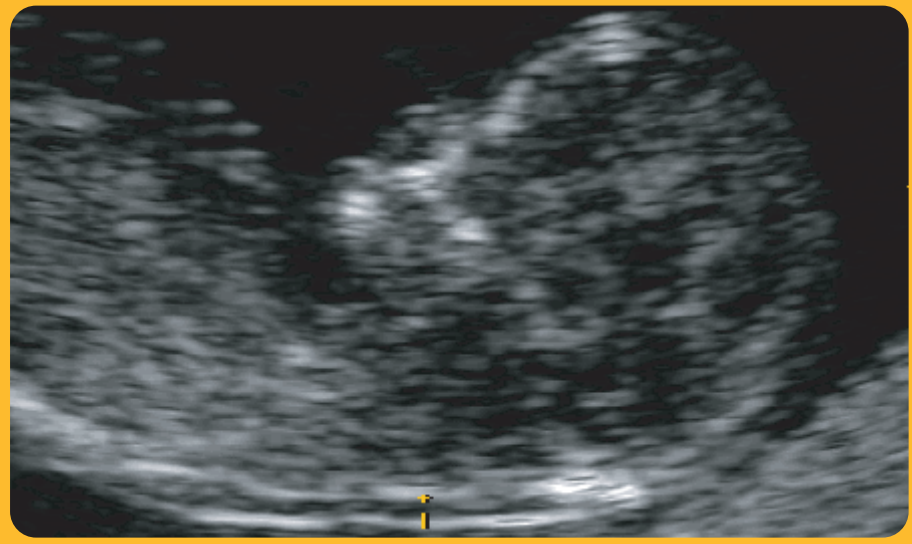
Down's syndrome screening - tests available

Test	Markers	False Positive Rate for 85% Detection Rate
Triple	AFP, hCG, uE_3	9.3%
Quadruple	AFP, hCG, uE_3, Inhibin A	6.2%
Nuchal Translucency	NT only	20%
Combined	NT, PAPP-A, hCG	6.1%
Integrated	1st trimester - combined test 2nd trimester - quadruple test	1.2%
Serum integrated	1st trimester - PAPP-A, hCG 2nd trimester - quadruple test	2.7%

HTA SURUSS report, 2003
Please refer to local policy where appropriate

Nuchal Translucency





Adapted from BroTaf Health Authority, 2000

Rubella

- Rubella (German Measles) is an illness caused by the rubella virus and presents with a rash, incubation period 14 - 21 days
- HPA & RCOG recommend all rashes in pregnancy be investigated
- Screening in pregnancy is to identify women who require vaccination postpartum in order to prevent congenital rubella in subsequent pregnancies
- Congenital rubella can cause multiple problems including deafness, heart and eye defects
- In the first 8-10 weeks of pregnancy infection results in severe fetal damage in up to 90% of cases. After this period the risk of damage is lower and likely to involve hearing impairment. Rubella defects are rare after 16 weeks gestation
- An antibody level of less than 10iu/ml is IgG negative (non immune)
- The MMR vaccine must be offered to IgG negative women postpartum. The woman should be advised not to conceive within one month of vaccination (can be given at the same time as Anti D but must be given in the opposite arm).

HPA- Health Protection Agency-RCOG-Royal College of Obstetricians & Gynaecologists

Syphilis

- Syphilis is a sexually transmitted infection, caused by the bacterium Treponema pallidum
- Screening in pregnancy is to identify women with an infection and offer treatment which will reduce the risks of the baby developing congenital syphilis
- If the pregnant woman has an untreated syphilis infection, the fetal loss rate is approximately 50%
- Babies that survive suffer considerable morbidity including: naso-facial hypoplasia, blindness, deafness, bone abnormalities etc.
- Congenital syphilis is transmitted via the placenta
- The screening tests have an accuracy of over 99%
- Positive or equivocal results require urgent referral to GUM, antibiotic treatment and discussion regarding the risks to the baby.

GUM – Genito-Urinary Medicine

HIV

- HIV is a retrovirus that attacks and destroys CD4 cells, resulting in immune suppression that may lead to AIDS
- Screening in pregnancy for HIV is to identify women with the infection, offer early treatment and appropriate care to reduce mother to baby transmission
- HIV is transmitted through: sexual contact, contact with contaminated blood products e.g. needle sharing, vertical transmission during pregnancy, delivery or breastfeeding
- The vertical transmission rate can be reduced from 25% to less than 2% with optimal management which includes anti retroviral therapy and appropriate obstetric and midwifery management
- A false negative result* may occur if a woman is tested in the window period between infection and sero-conversion
- There is a risk of acquiring HIV during pregnancy if a woman or her partner participates in high risk behaviour
- Confirmed positive results require specialist counselling, urgent referral to GUM.

GUM – Genito-Urinary Medicine

* see test performance

Hepatitis B (HBV)

- HBV is an infectious disease of the liver caused by the HBV virus resulting in both acute and chronic infection
- Screening in pregnancy for HBV is to identify women who are infected or carriers, as their babies will be at significant risk of contracting HBV
- A newborn programme of vaccination if completed is 95% effective. This involves an initial dose within 48 hours of birth, 2nd dose at one month, 3rd dose at two months, 4th dose at 12 months (with a blood test to check immunity)
- Chronic infectivity may result in cirrhosis and carcinoma, about 20% of chronic HBV carriers die from liver failure
- HBV is transmitted through sexual contact, contaminated blood e.g. needle sharing or by vertical transmission
- The screening test identifies Hepatitis B surface antigen (HBsAg) and has an accuracy of 99.9%
- The presence of Hepatitis B e antigen (HBeAg) indicates high infectivity
- Confirmed positive results require referral to gastroenterology and/or hepatology services.

Sickle Cell Disorders

- Sickle Cell Disorders are inherited blood conditions that affect haemoglobin. Inheritance of an affected gene from both parents results in a disorder and inheritance of only one affected gene result in a healthy carrier

- Screening in pregnancy should be performed early in the 1st trimester. If a woman is a carrier or has a disorder partner testing should be offered to assess risk

- If a couple who are both healthy carriers decide to have children, there is a **1 in 4** chance with each pregnancy that the child will have a Sickle Cell Disorder.

Sickle Cell Disorders

- There are approximately 240,000 healthy carriers and >12,500 people with Sickle Cell Disorders in the UK

- Sickle Cell Disorders (which include HbSS, HbSC, HbSD Punjab, HbSβ ThaI [β+, β0, δβ, Lepore], HbSO Arab, Hb-S/HPFH) are a variable set of conditions but can cause chronic anaemia, jaundice, painful crisis, organ damage, infections and strokes

- All pregnant women with Sickle Cell Disorders should receive specialist obstetric and haematological care.

Thalassaemia Disorders

- Alpha and Beta Thalassaemia Major are inherited blood conditions that affect haemoglobin. Alpha Thalassaemia Major is incompatible with life. Beta Thalassaemia Major results in severe anaemia. Inheritance of an affected gene from both parents results in a disorder and inheritance of only one affected gene results in a healthy carrier
- There are other less serious Thalassaemia Disorders which can be detected by the screening programme
- Screening in pregnancy should be performed in the 1st trimester. If a woman is a carrier or has a Thalassaemia Disorder, then partner testing should be offered to assess risk
- If a couple who are both healthy carriers decide to have children, there is a **1 in 4** chance with each pregnancy that the child will have a Thalassaemia Disorder.

Thalassaemia Disorders

- There are approximately 214,000 carriers and >700 people with Thalassaemia Disorders in the UK
- Beta Thalassaemia Major results in life threatening anaemia and requires blood transfusions every 4 to 6 weeks and iron chelation to prevent further illness
- All pregnant women with a Thalassaemia Disorder should receive specialist obstetric and haematological care.

For further information visit:
http://www.kcl-phs.org.uk/haemscreening/

Sickle Cell & Thalassaemia Screening Flow Chart

Ask EVERY woman about her and her baby's father's family origins to identify risk

High Prevalence Trusts
- Offer screening for haemoglobin variants
- Offer screening for Thalassaemia

Low Prevalence Trusts
- Offer screening for Thalassaemia
- Determine family origin of woman & baby's father
- Offer screening for haemoglobin variants if at high risk or woman asks for screening

If positive

Offer partner screening

Partner is a carrier

Partner is absent

Offer pre-natal diagnosis

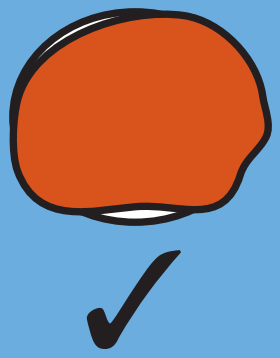
Newborn blood spot screening

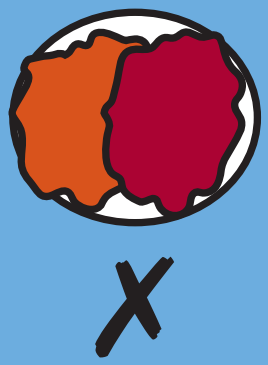
- Newborn screening tests are recommended for all babies
- Offer parents screening (use national parent information leaflet) and record parental choice. Conditions screened may vary locally, please check
- Take blood sample at 5-8 days of age, ideally 5 days (irrespective of prematurity, milk feeds or transfusion)
- If possible take sample prior to transfusion
- Use only an approved automated device
- Ensure baby's NHS number and correct name is on the card.

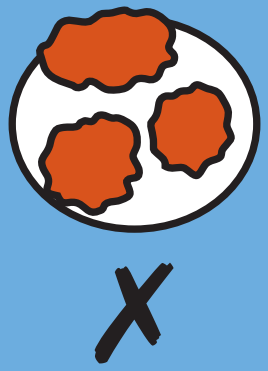
For further professional and parent information see:
www.newbornscreening-bloodspot.org.uk

Newborn blood spot screening
Taking a sample

- Ensure skin is clean
- Activate recommended device
- Wait for blood to flow naturally
- Allow blood to fill each circle and soak completely through
- Do not add layers of small blood spots
- Complete the card as requested and post to laboratory within 24 hours of sampling.







Phenylketonuria (PKU)

- PKU is an autosomal recessive genetic condition that affects 1 in 10,000 babies in the UK
- Babies with this condition are unable to metabolise phenylalanine (an amino acid found in proteins)
- Untreated babies develop serious permanent mental disability
- Ongoing treatment, with a strictly controlled diet, prevents disability
- Screen positive babies are seen by a specialist and a low protein diet should be started by 21 days of age. Treatment is very effective.

Congenital Hypothyroidism (CHT)

- CHT affects 1 in 4,000 babies in UK
- Babies with this condition produce insufficient thyroxine
- Untreated babies develop permanent physical and mental disability. Treatment can prevent disability
- Screen positive babies are seen by a specialist and treatment with thyroxine should be started by 21 days of age
- Treatment is effective in preventing severe mental disability.

Cystic Fibrosis (CF) - Newborn screening

- CF is an autosomal recessive genetic condition that affects 1 in 2,500 babies in UK
- Sticky mucus secretions cause digestive problems, recurrent chest infections leading to lung damage, poor growth and development. Survival is to mean age 31
- Screening enables early detection of pre-symptomatic babies
- Early treatment, including dietary supplements, medication and physiotherapy may improve health
- Some babies will require a second blood sample at approx 21 days for further testing
- Affected babies are seen by a specialist and started on treatment by 30 days of age
- Screening can identify some CF carriers.

Sickle Cell Disorders – Newborn screening

- Sickle Cell Disorders affect 1 in 2,500 babies in the UK
- Affected babies are treated with penicillin and prevenar vaccine before 3 months of age
- Untreated babies are at high risk of death or complications from treatable infections or severe acute anaemia in the first few years of life
- Parents of affected children need to be fully informed and involved in the management of their child's condition.

Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD)

- Pilot implementation of screening in 6 laboratories across UK presently
- MCADD is an autosomal recessive genetic condition that affects 1:10,000-20,000 babies in the UK
- MCADD affects breakdown of fat and blocks energy production. This can result in drowsiness, lethargy, vomiting, seizures and in some cases coma and death
- 20-25% mortality, 30% survivors with central nervous system sequelae at first clinical presentation
- Symptoms can occur quickly in infants who are not feeding well or are unwell with an intercurrent infection, e.g. diarrhoea & vomiting
- Mean age at presentation is 14 months
- Treatment is by prevention of metabolic crisis: Avoid fasting and close monitoring to determine safe periods between meals
- Emergency regime if unwell involves administration of a Glucose polymer (maxijul) and IV dextrose.

Useful Addresses

- Antenatal Results and Choices
  www.arc-uk.org
- Congenital hypothyroidism: www.ich.ucl.ac.uk/factsheets
- Contact a Family
  www.cafamily.org.uk
- Cystic Fibrosis: www.cftrust.org.uk
- Down's Syndrome Association
  www.dsa-uk.com
- Down's Syndrome Screening Programme
  http://www.screening.nhs.uk/downs/home.htm
- Health Protection Agency: www.hpa.org.uk
- MCADD: www.climb.org.uk
- Miscarriage Association
  www.miscarriageassociation.org.uk
- NELH (National Electronic Library for Health)
  http://www.library.nhs.uk/screening

Useful Addresses

- NHS Sickle Cell & Thalassaemia Screening Programme:
  www.kcl-phs.org.uk/haemscreening/
- PEGASUS (professional education for genetic assessment & screening)
  www.pegasus.nhs.uk
- Personal experiences of health and illness
  www.dipex.org/antenatalscreening
- Phenylketonuria: www.nspku.org.uk
- Positively Women (Support for women living with HIV)
  www.positivelywomen.org.uk
- Sickle cell disease: www.sicklecellsociety.org
- UK Newborn Screening Programme Centre:
  www.newbornscreening-bloodspot.org.uk
- UK Thalassaemia Society
  office@ukts.org, www.ukts.org
- Visit http://www.screening.nhs.uk/cpd/home.htm for training resources and the Screening Choices Programme