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Genetic Healthcare & Public Health Screening in Scotland

03 September 2012

12/55

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This briefing describes genetic healthcare services and public health screening programmes in Scotland. It includes a brief introduction to genetics in relation to healthcare; information on the four regional genetics services and an overview of recent developments in genetic healthcare in Scotland. This briefing also outlines public health screening programmes relating to pregnancy, newborn and adult screening.



The Scottish Parliament
Pàrlamaid na h-Alba

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EXECUTIVE SUMMARY

Genetics services

Genetic conditions, like any other condition, affect one or more parts of the body. They can be inherited from previous generations and passed on to future generations. Genetic healthcare in Scotland is provided by NHS-funded regional genetic services. These regional genetic services are located in Aberdeen, Dundee, Edinburgh and Glasgow and provide a service to patients throughout Scotland. Not only do the regional genetic services provide a face-to-face service through the clinicians and genetic counsellors, they provide a laboratory service for analysing samples taken to test for genetic conditions.

The 2006 'Review of Genetics in Relation to Healthcare in Scotland' (the Calman Review) made a number of recommendations on how the Scottish genetics service could be improved. It recommended an increase in clinical staff, improved public engagement and a consortium approach to the provision of genetic services. As a result of the Calman Review, Gengage was established by the Scottish Government in 2008 to increase public awareness of genetics and to promote dialogue and debate on issues relating to genetic healthcare.

There is a vast amount of genetic healthcare research taking place in Scotland. For example, Generation Scotland is a collaboration between Scottish medical schools, research institutes and the NHS. Generation Scotland aims to identify the genetic basis of common complex disease and use ethically-approved family and population-based studies to understand more about genetic disease. The Chief Scientist Office of the Scottish Government also funds genetic research via the Experimental and Translational Medicine Research Committee and the NHS in Scotland funds a number of clinical research projects.

Public health screening

Public Health Screening is the process used to identify apparently healthy people who may be at an increased risk of a disease or condition. Some screening programmes have a genetic component. There are three strands of screening programmes. These are prenatal screening, newborn screening and adult screening. Screening programmes have advantages and disadvantages and are regulated by the UK National Screening Committee. There are a number of criteria which must be met in order for a screening programme to be implemented. These include factors around the condition, the screening test, the treatment and the screening programme. Screening cannot provide a clear diagnosis of any condition, and instead the results are called 'presumptive'. When screening identifies someone at 'high-risk' of a condition, they will be referred for further diagnostic testing.

Prenatal screening is offered to all pregnant women in Scotland and takes the form of a blood test or an ultrasound scan. Conditions such as sickle cell disease, thalassaemia, Down syndrome, fetal anomalies and infectious diseases are tested routinely as part of pregnancy screening. When screening identifies a pregnancy as 'high-risk', the mother may be offered a diagnostic test such as an amniocentesis or chorionic villi sampling (CVS). Diagnostic tests allow parents to make informed choices about their pregnancy; however, there is a small risk of miscarriage associated with both of these diagnostic procedures.

Parents of all newborn babies in Scotland are offered a blood-spot screening test for their child. This test takes a spot of blood by pricking the heel of the baby and can test for five rare, but serious, conditions including cystic fibrosis. Early diagnosis of any of these five conditions can allow for early treatment of the disease.

Newborn babies are also tested as part of the Newborn Hearing Screening Programme for hearing defects, and the test usually takes place before the baby leaves hospital. Newborns are also routinely given a physical head to toe examination before they leave hospital. Although this is routinely carried out in Scotland, it is not a national screening programme like it is in England.

Adults of certain age groups are offered regular screening for breast cancer (women aged 50-70, every 3 years); bowel cancer (adults aged 50-74, every 2 years) and cervical cancer (women aged 20-60, every 3 years). Uptake of screening for breast cancer and cervical cancer is relatively high at 74.9% and 73.6% respectively, compared to bowel cancer which has a low uptake of 53.7%. Uptake of bowel cancer screening is higher in females than in males and is affected by social factors with the lowest rate of uptake being in the poorest areas.

Breast cancer screening in 2011/12 detected over 1,700 cases of breast cancer in Scotland. Regular bowel screening is estimated to reduce the risk of dying from bowel cancer by 16%. Cervical screening is estimated to save around 5,000 lives in the UK each year and regular cervical screening is estimated to prevent 8 out of 10 cervical cancers developing in the first instance. The human papillomavirus (HPV) vaccination was introduced in Scotland in 2008 to help combat cervical cancer. It is offered to school girls aged 12-13 years. The HPV vaccination prevents some forms of cervical cancers from developing in the first instance; however, it does not protect against all cervical cancers and therefore regular screening is still important.

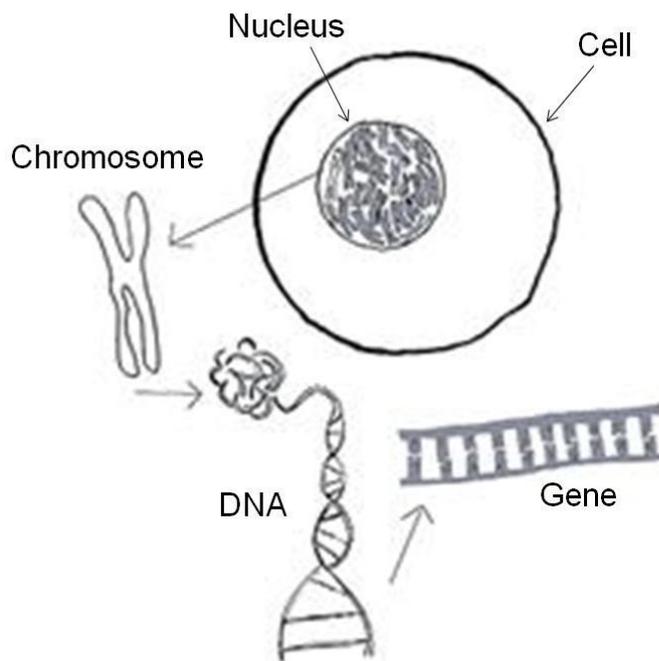
Diabetic retinopathy screening programme is offered annually to everyone aged 12 and over with diabetes. The screening test looks for retinopathy which is a condition that can cause severe damage to the eye and can result in blindness. Early detection can allow for the retinopathy to be treated and can prevent any serious damage from occurring. Diabetic retinopathy is the biggest single cause of blindness and visual impairment in Scotland amongst people with working age. The rising prevalence of diabetes in Scotland means that diabetes is likely to remain a major health and economic problem and this screening programme will help reduce the health and economic problems associated with retinopathy.

In July 2012, the Scottish Government announced its intention to roll out an abdominal aortic aneurysm (AAA) screening programme to all men entering their 65th year. An AAA is an enlarged part of the aorta in the abdomen that has no symptoms unless it bursts, in which case it usually leads to death. AAA screening will only be offered to men aged 65 as 95% of AAA ruptures occur within this group. This new screening programme, to be rolled out across the country by December 2013, is expected to prevent around 170 deaths each year and to shift the balance of care from reactive emergency treatment to preventative management through early diagnosis.

INTRODUCTION

DNA (deoxyribonucleic acid) is the genetic code of life. It determines characteristics from eye colour and the ability/inability to tongue roll to predisposition to genetic (or inheritable) conditions. With the exception of identical twins, everyone has a unique DNA sequence. A gene is a short sequence of DNA. Humans each have around 25,000 genes. Genes are tightly packaged into little bundles known as chromosomes. There are 46 chromosomes in the nucleus (control centre) of each cell (23 inherited from the mother and 23 from the father). Cells are the basic building blocks of life and are like tiny bricks that make up the human body.

Figure 1: DNA is packaged into chromosomes of which there are 23 chromosome pairs in each cell. Image used with kind permission of Genetic Alliance UK.



A genetic condition can arise when there is a change in the DNA code of a particular gene or where there is a change to the structure of chromosomes. Genetic conditions, like any other condition, affect one or more parts of the body. They are, however, unique in that the condition can be inherited from previous generations and passed on to future generations.

POLICY CONTEXT

Provision of Healthcare Services in Scotland, including the provision of Genetic Healthcare Services, is devolved to the Scottish Parliament. However, ethical and legal matters relating to genetics are reserved to Westminster. Reserved matters include regulation of:

- Genetic research
- Genetic testing
- Assisted conception (e.g. IVF, sperm/egg donation and surrogacy)
- Xenotransplantation (transplantation of cells or organs from one species to another)
- Embryology and
- Abortion

The Emerging Science and Bioethics Advisory Committee (ESBAC) is responsible for providing advice to the four UK Health Ministers on emerging scientific developments and their ethical, legal and economic implications. Its remit involves the field of human genetics. The newly-established ESBAC has taken over the responsibilities of the former Human Genetics Commission (HGC) which was the former UK Government's advisor on new developments in human genetics. The HGC was disbanded in June 2012 following a UK Government review of Arm's Length Bodies. The HGC's role was to advise the government on social, ethical and legal issues arising from genetics.

GENETICS SERVICES

WHAT ARE GENETICS SERVICES

Genetics services encompass clinical genetics, molecular genetics, cytogenetics and biochemical genetics.

Clinical Genetics

Clinical Genetics is the branch of genetics healthcare that directly deals with patients. It is generally an outpatient service and involves:

- Taking a detailed pedigree (family tree) analysis of genetic conditions and providing clinical assessment by family history;
- Deciding on further investigations and taking samples for genetic testing (molecular or cytogenetic testing);
- Genetic counselling: bringing all the information together and communicating it in an understandable way to allow families to make informed choices and decisions. Providing information about genetic risk for patients and family members;
- Discussions about options available to patients, including therapies, diagnostic testing, management plans and support groups;
- Diagnosis of rare genetic conditions;
- Co-ordination of screening for specific rare syndromes;
- Multidisciplinary working to direct investigation and management of specific diseases with significant genetic component.

Molecular Genetic and Cytogenetic laboratories

Molecular Genetic laboratories (or 'DNA labs') provide a genetic testing facility for genetic conditions that arise from specific changes (mutations) in the DNA code. For example, some inheritable breast cancers can result from changes in one of two specific genes: BRCA1 and BRCA2.

Cytogenetic laboratories (or 'chromosome labs') provide a genetic testing facility for genetic conditions that arise from chromosomal abnormalities. For example, Down syndrome is caused by 3 copies of chromosome 21 (an individual normally would have only two copies of each chromosome – one maternal and one paternal).

Biochemical Genetics

Biochemical Genetics labs provide a genetic testing facility for genetic conditions for conditions that produce an unusual amount of protein in a blood sample. This includes testing prenatal screening samples for neural tube defects and Down syndrome. The Scottish Newborn Screening Laboratory is based at the Biochemical Genetics Department at the West of Scotland Regional Genetics Service (See Figure 2) and tests newborn babies for rare but serious genetic conditions.

GENETIC TESTING

A genetic test is a test that looks for specific changes to a particular gene that is known to cause a particular genetic condition. There are four types of genetic testing (ScotGEN, 2012):

Diagnostic Genetic Test – this is undertaken because a diagnosis is suspected in a patient. Most of these tests can be requested directly by a doctor, although more complex cases might be referred to one of Scotland's regional genetic services. This includes diagnostic testing for suspected cancer cases.

Predictive Genetic Test – this is undertaken for a patient who is well but has a family history of a particular genetic condition. For example, someone with a family history of Huntington's disease might be offered a predictive test to determine if they are going to develop the condition. Predictive genetic testing will only be authorised by a Clinical Geneticist or Genetics Counsellor.

Carrier Test – this is undertaken for a patient who is well and will never develop the condition themselves, but may transmit the condition to their children. Testing for cystic fibrosis is an example. Carrier testing allows women (and partners) to make informed choices about existing or future pregnancies.

Screening Test – this is offered to a particular group of people for a particular condition. An example is the screening offered to pregnant women for Down syndrome. Screening tests are discussed in more detail below (see page 10).

The UK Genetic Testing Network (UKGTN) advises the NHS in all four UK countries about genetic testing for inheritable genetic conditions. It aims to ensure the provision of high quality genetic testing services. UKGTN is a network of genetic testing laboratories, clinicians and commissioners of NHS genetic services and involves input from patient groups (UKGTN, 2012a). The UKGTN assess whether or not a genetic test is likely to be of benefit to patients. If it is, then a recommendation is passed to the Genetics Commissioning Advisory Group (GenCAG) who will then request that the relevant commissioners provide funding for the test. In Scotland, the National Services Division is responsible for commissioning genetic testing (UKGTN, 2012b).

The Scottish Genetics Laboratory Consortium (which is funded by National Services Division) determines which genetic tests molecular genetics and cytogenetics laboratories perform. The consortium ensures that the Scotland's four regional genetics services work together to avoid duplication and to provide genetic testing for a wide range of conditions efficiently (National Services Division, 2012b). Tests for rare genetic conditions not available in Scotland are sourced from England or abroad (National Services Division, 2012b). The Scottish Genetics Laboratory Consortium has the role to:

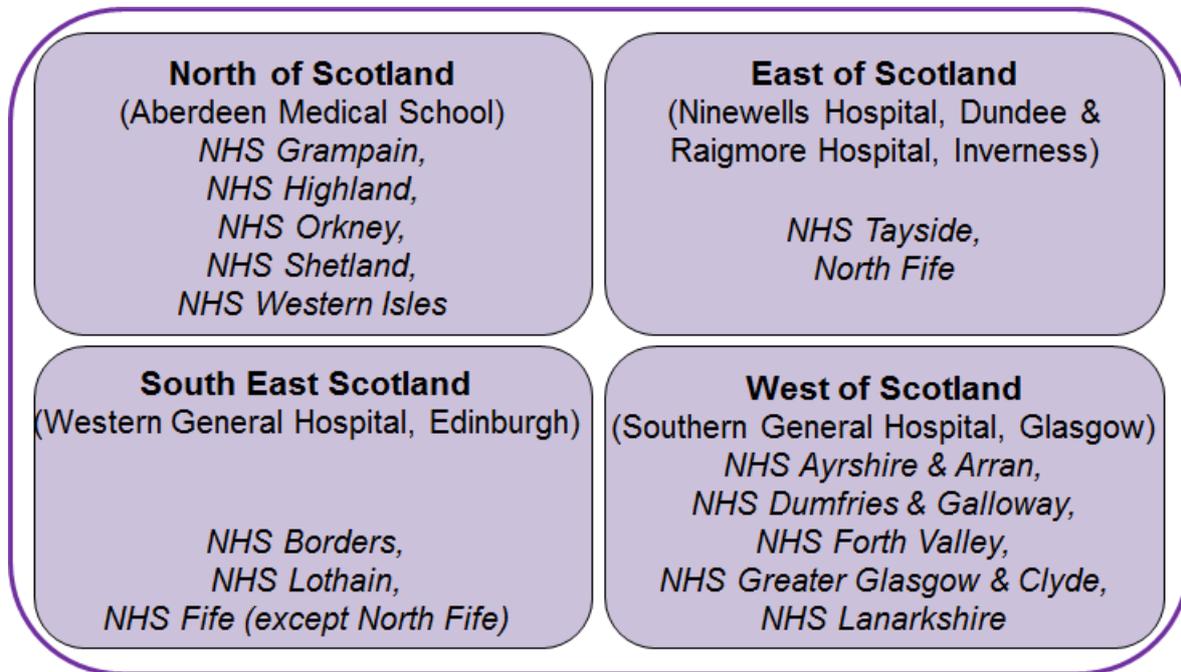
- Openly discuss and agree workload distribution between the four regional genetics services so as to make the most effective use of available resources;

- Consider the classification and declassification of genetic tests by the UK Genetic Testing Network (UKGTN) so as to ensure that all tests undertaken are evidence based (National Services Division, 2012b).

GENETICS SERVICES IN SCOTLAND

Healthcare for patients affected by or at risk of a genetic condition is delivered by regional genetics services. In Scotland, there are four regional genetics services as shown in Figure 2.

Figure 2: Scotland's four Regional Genetics Services and their health board catchment areas



National Services Division

National Services Division is a division within NHS National Services Scotland (NSS). National Services Division receives top-sliced, ring-fenced funding from the Scottish Government to commission and performance-manage nationally designated specialist services, national managed clinical networks and screening programmes on behalf of NHS Scotland. The National Services Division is responsible for commissioning and performance-managing specialist services (including molecular genetic and cytogenetic laboratories) on behalf of NHS Scotland. National Services Division aims to:

- Ensure equity of access for all Scottish residents to specialist and screening services;
- Ensure the best possible clinical outcomes within the funding available;
- Provide a secure funded environment for the establishment and development of new national services;
- Provide a risk-sharing arrangement for NHS Boards where incidence is sporadic and treatment involves specialist skills or expensive equipment; and
- Avoid the unnecessary proliferation of duplicate services, thus promoting clinical quality and cost effectiveness (National Services Division, 2012a).

Workforce

Regional genetics services are staffed by a number of specialists in addition to the administrative, technical and support staff. The specialists are:

Clinical Geneticists – these are medically qualified doctors who specialise in clinical genetics. Clinical Geneticists usually deal with more complex cases of inheritable genetic conditions.

Genetic Counsellors – these are healthcare professionals usually with a science or nursing background. Genetic Counsellors can specialise in one or more types of Genetic Conditions. Genetic counsellors can register with the Genetic Counselling Registration Board (GCRB), although registration is currently voluntary. In Scotland, there are 26 Genetic Counsellors, 13 of whom are GCRB-registered (GCRB, 2012). The GCRB was established by the profession to regulate the standards of practice of Genetic Counsellors in the UK. Its purpose is to establish, maintain and improve standards of practice to assure public safety. Workforce levels for Genetic Counsellors at the four Regional Genetics Services are shown in Table 1.

Table 1: Genetic Counsellor Workforce in Scotland

Regional Genetics Service	Number of Genetic Counsellors
North of Scotland	6
East of Scotland	4
West of Scotland	10
South East Scotland	6

Genetic Counsellors with a scientific background require a GCRB-approved MSc in Genetic Counselling (in the UK these courses are only available at Manchester University and Cardiff University). Genetic Counsellors with a nursing background are not required to have the MSc on appointment; however, there is the expectation that they will gain it at some point. In England & Wales, the Department of Health has set up Genetic Counsellor training posts; however, no such scheme has been implemented in Scotland.

Clinical Scientists – these healthcare professionals provide the laboratory service for genetic testing. Clinical Scientists in the regional genetics services usually specialise in either molecular genetics or cytogenetics. However, traineeships have recently been established in molecular pathology, which integrates both genetic specialities and incorporates analysis of cancerous tumours.

Total staffing for the regional genetics laboratories in Scotland is approximately 155 whole time equivalents (WTE). The workforce is divided up as shown in Table 2 (National Services Division, 2012c), below.

The head of a regional genetics service is usually a Consultant Clinical Scientist or Consultant Clinical Geneticist. There are a number of associations representing genetics healthcare professionals (Appendix 1).

[NHS Education for Scotland](#) is the special health board responsible for delivering and developing education and training for those who work in NHS Scotland. NHS Education for Scotland covers national postgraduate pre-registration training for clinical scientists, support for other groups, leadership development across healthcare science, support staff initiatives and trainers and assessors (NHS Education for Scotland, 2012).

Table 2: Genetic Laboratories Workforce in Scotland

Role	WTE
Consultant Clinical Scientist	29.0
Principal Clinical Scientist	10.1
Clinical Scientist	46.8
Biomedical Scientist	3.6
Biomedical Support	1.0
Healthcare Scientist	0.5
Healthcare Scientist Associate	2.0
Healthcare Assistant	2.0
Quality Manager	2.3
Laboratory Manager	2.0
Senior Genetic Technologist	9.5
Genetic Technologist	27.3
Medical Technical Officer	8.0
Administrative	11.9
Information Technology	0.2
TOTAL	155

Budgets

As shown in Table 3, total public funding for molecular and cytogenetic laboratory services in Scotland across the four regional genetics services in 2011/12 was £10,725,388. This includes staffing, equipment, consumables and maintenance (National Services Division, 2012c). In addition, £464,574 was spent on tests that were sent to other laboratories outside Scotland (National Services Division, 2012c):

Table 3: Molecular Genetic & Cytogenetic Laboratory Services Public Spending 2011/12

Spending 2011/12	£ million
Total in Scotland	10.725
Tests sent to labs outside Scotland	0.465
Total Genetics Spending	11.190

Research Funding

There are a number of research centres in Scotland specialising in genetics research. For example, the Medical Research Council (MRC) [Human Genetics Unit](#), based at the Southern General Hospital in Edinburgh, studies the genetic basis of disease. In August 2012, the MRC announced £60m of funding for Edinburgh University's Institute of Genetics and Molecular Medicine ([IGGM](#)) (BBC, 2012). This funding aims to help IGGM scientists gain insights into conditions such as cancer, arthritis and schizophrenia and could help the scientists develop new tests and therapies for patients (BBC, 2012).

The Scottish Government Chief Scientist Office (CSO) has the responsibility for encouraging and supporting research into health and health care needs in Scotland. This includes research into genetics. Genetics research is a very broad field and the figures for research funding are

therefore interpretive. The CSO funds a number of genetics healthcare research projects. These include funding through the Experimental and Translational Medicine Research Committee (ETMRC). The ETMRC has an annual budget of £5.2m. The Scottish Government estimates that around 25% of the research projects funded through ETMRC have a genetics component (Scottish Government, 2012a). In addition, the CSO funds Generation Scotland (discussed below). Funding over the past 5 years for this project is shown in Table 4 (Scottish Government, 2012a).

Table 4: Scottish Government Chief Scientist Office funding of Generation Scotland

Year	£ million
2007	1.0
2008	1.0
2009	1.0
2010	0.7
2011	0.4

The CSO also funds two Scottish Experimental Cancer Medicine Centres (ECMCs), one at Glasgow and a second ECMC which is a collaboration between Edinburgh and Dundee. ECMCs are set up to conduct early phase clinical trials, all of which have a genetic component to their research. Total funding to the Scottish ECMC's by the CSO from 2007-11 was £325K (Scottish Government, 2012a). The CSO also invests the majority of its annual budget in the infrastructure that allows NHS Scotland to participate in high impact clinical research. This could be labelled as genetics research. The CSO estimates of funding for clinical research is shown in Table 5 (Scottish Government, 2012a).

Table 5: Scottish Government Chief Scientist Office estimated funding for Clinical Research

Year	£ million
2007	2.6
2008	2.6
2009	2.6
2010	2.3
2011	2.0

Calman Review

In 2006, Professor Sir Kenneth Calman led the '[Review of Genetics in Relation to Healthcare in Scotland](#)' (Calman Review) (Scottish Government, 2006). The Calman Review made a number of recommendations about genetic healthcare in Scotland with regards to staffing, public engagement and clinical and laboratory services.

The Calman Review resulted in significant funding being made available to upgrade clinical and laboratory facilities and recruit more medical, clinical, scientific, administrative and nursing staff within the regional genetics services. It led to further funding in genetic healthcare services and an increase in the number of Consultant Geneticists and Genetic Counsellors in Scotland. It also paved the way for a Neuromuscular Clinical Geneticist role with pan-Scotland responsibilities. The review also made funding available for specialist genetic care co-ordinators for complex single gene disorders.

The Calman Review also resulted in the extension of the consortium approach from molecular genetic to include cytogenetic services. As a result, the Scottish Genetics Laboratory Consortium was established to allow for sharing of resources and workforce across Scotland's four regional genetics services. The aim of this has been to achieve workflow balancing, resulting in lower reporting times.

Public engagement in genetics is another area that was discussed in detail in the Calman Review. As a result of the review, Gengage (discussed below) was established as a formal funded network to link all of those with a background or interest in genetics with the general public.

GENETICS ORGANISATIONS

There are a number of professional associations for genetics healthcare specialists and clinical scientists. These associations usually have a UK-wide remit. For more information about these associations, see Appendix 1. In addition to these, there are a number of genetics organisations in Scotland.

Scottish Genetics Education Network ([ScotGEN](#))

ScotGEN is funded by the Scottish Government, is a collaboration between Scotland's four regional genetics services and Scotland's universities. It is a network for people involved in the teaching of genetics for healthcare in Scotland and provides teaching materials.

ScotGEN is also home to the [Scottish Clinical Genetics Forum](#). This is a network between the four regional genetics services, National Services Division, Gengage, Scottish Genetic Alliance UK, the Scottish Government and other interested parties, including those from overlapping healthcare specialities and patient groups. The Scottish Clinical Genetics Forum aims to help develop an innovative, seamless service and integrate patient and public views into service planning and care guidelines (ScotGEN, 2012). It allows for the sharing of knowledge and expertise whilst setting a series of standards to measure staffing levels, waiting times, referrals and best clinical practice. The clinical genetics forum aims to agree quality standards and common ways of working across the regional genetics services.

Scottish Genetic Alliance UK

[Scottish Genetic Alliance UK](#) represents patients and families in Scotland to ensure that their voice is heard by decision makers such as NHS Scotland and the Scottish Government. They aim to improve the lives of people affected by genetic conditions by ensuring that high quality services are available to everyone who needs them. They aim to raise awareness of genetic conditions and campaign on policy issues.

Gengage

The Scottish Healthcare Public Engagement Network ([Gengage](#)) was established by the Scottish Government in 2008 following recommendations in the Calman Review, and its funding comes to an end in 2012. Its remit is to co-ordinate the efforts in Scotland to increase public awareness, dialogue and debate on issues to do with healthcare genetics. Gengage is funded by the Scottish Government and managed by the ESRC Genomics Policy and Research Forum at the University of Edinburgh.

Generation Scotland

[Generation Scotland](#) is a collaboration between the four Scottish Medical Schools (Aberdeen, Dundee, Edinburgh & Glasgow), biomedical research institutes and NHS National Services Division. It aims to create ethically-sound family and population-based infrastructure to identify the genetic basis of common complex diseases. Current projects include the Scottish Family Health Study ([GS:SFHS](#)), Genetic Health in the 21st Century ([GS:21CGH](#)) and the Donor DNA Databank ([GS:3D](#)). So far, over 30,000 participants have been recruited to these three projects.

PUBLIC HEALTH SCREENING

Screening is the process of identifying apparently healthy people who may be at an increased risk of a disease or condition. People who are identified as being at increased risk of a disease or condition during screening can then be offered information, further tests and appropriate treatment to reduce their risk and/or any complications arising from the disease or condition. Screening is associated with some complex terminology and these are explained in Table 6.

Table 6: Explanation of Screening Terminology

Term	Description
Cut-off level	Screening tests divide people into low-risk or high-risk groups. People in high-risk groups are offered further analysis. The cut-off point is the point defined by the programme to separate low-risk from high-risk
Detection rate / Sensitivity	A high detection rate or high sensitivity means that the test identifies as many people as possible with the condition. Detection rate / sensitivity is measured as the proportion of those with the condition, who have a positive (high-risk) screening result
False-negative result	People who have the condition being screened for but are given a negative (low-risk) result during screening
False-positive result	People who do not have the condition being screened for but are given a positive (high risk) result during screening
High-risk	People who are given a positive screening result
Low-risk	People who are given a negative screening result
Negative predictive value	The proportion of people with a negative (low risk) screening result who actually do not have the condition being screened for
Positive predictive value	The proportion of people with a positive (high-risk) screening result who actually do have the condition
Presumptive result	Screening tests divide people into high-risk and low-risk categories. They do not provide a definite answer. Screening results are called presumptive results until they are confirmed by diagnostic tests
Prevalence	The number of individuals in a population with the target condition
Screening	Testing people who do not have signs or symptoms of the condition being tested for with the aim of reducing risk of an adverse outcome or giving information about risk
Specificity	High specificity means the test has as few false positives as possible. It is measured as the proportion of those without the condition who have a negative result

Target condition	The condition that the screening programme is aiming to find, to reduce the risk of adverse effects from that condition
Target population	The target group who are offered screening
True-negative result	People who are given a negative (low-risk) screening result who do not have the condition being screened for
True-positive result	People who are given a positive (high-risk) screening result who do have the condition being screened for
Uptake	The proportion of people, who when offered a test, take it up

The easiest way to understand the screening process is to think of it as a kitchen sieve (National Screening Committee, 2012a). Screening programmes aim to identify individuals with or at risk of a particular condition or disease. This is known as the target condition. Screening programmes also have a target population. This is the group of people that the programme aims to screen. The target population are offered screening and those that uptake the invitation for screening are asked to “pass through the sieve”. Passing through the sieve is a metaphor for taking a screening test, such as having an ultrasound scan or giving a blood sample for laboratory analysis. In an ideal screening programme, the people without the target condition will pass through the sieve. People who pass through the sieve are deemed ‘low-risk’ and are not referred for further testing. These people are called true-negatives because the screening programme has classified them as ‘low-risk’ and they do not have the target condition. The people who do not pass through the sieve who do have the condition are called true-positives. They have been identified as ‘high-risk’ by the screening programme and further testing has confirmed that they do have the target condition.

However, the screening process is not that simple and there are some problems with the sieve model. Sometimes, people who do have the target condition can pass through the sieve. They are false-negatives because they have been given a ‘low-risk’ result by screening but they do actually have the target condition. Similarly, those who do not pass through the sieve who do not have the target condition are false-positives because they have been deemed ‘high-risk’ by the screening programme but further tests have shown that they do not have the target condition.

The results given by screening are called ‘presumptive’ results because screening does not provide a clear diagnostic result. Screening only categorises individuals into ‘high-risk’ or ‘low-risk’ categories. A ‘cut-off’ point is defined that separates low-risk results from high-risk results. The cut-off point is based on scientific evidence of likelihood that an individual above or below this point will have the target condition.

There are two main performance indicators for screening tests. The detection rate (or sensitivity) of a screening programme is the measure of all those with the target condition in the target population that screening identifies as ‘high-risk’. Specificity of a screening programme refers to the proportion of individuals without the target condition that are classified as low-risk by the screening programme. Screening programmes are therefore continuously monitored and reviewed by the National Screening Committee to ensure that the screening programme is effective.

CRITERIA FOR A SCREENING PROGRAMME

The [UK National Screening Committee](#), currently chaired by Scotland's Chief Medical Officer, Sir Harry Burns, advises the NHS and Health Ministers throughout all four UK countries about all aspects of screening and supports the implementation of screening programmes. The role of the National Screening Committee is to provide expert independent policy advice on what screening programmes to implement based on quality research.

Screening programmes have limitations and important practical and ethical differences from clinical practice as screening targets apparently healthy people. The potential harms of screening must be weighed up against the benefits. Screening aims to help them make better informed choices about their health. Screening can allow for early diagnosis of conditions and can therefore allow people to make informed choices or begin treatment early. The National Screening Committee aim to only implement screening programmes that do more good than harm. The criteria for a screening programme is detailed in Box 1.

Box 1: Criteria for a Screening Programme (National Screening Committee, 2012b).

The Condition

- The condition should be an important health problem and its epidemiology and natural history should be adequately understood
- There should be a detectable risk factor, disease marker or early symptomatic stage
- All cost-effective reasonable primary interventions must have been implemented
- If carriers of a mutation are identified during screening, the consequences should be understood including psychological implications

The Test

- The screening test should be simple, safe, precise and validated
- The test should be acceptable to the population
- The distribution of test values in the target population should be known and a suitable cut-off level defined and agreed
- There should be an agreed policy on the further diagnostic investigation of individuals with a positive test result and on the choices available to those individuals
- If the test is for mutations, the criteria used to select the mutations to be covered by screening, if all possible mutations are not being tested, should be clearly set out

The Treatment

- There should be an effective treatment for those affected by the treatment
- There should be evidence that early intervention will lead to better outcomes
- There should be agreed evidence-based policies covering which individuals should be offered treatment and the appropriate treatment to be offered
- Clinical management of the condition and patient outcomes should be optimised in all health care providers prior to participation in a screening programme

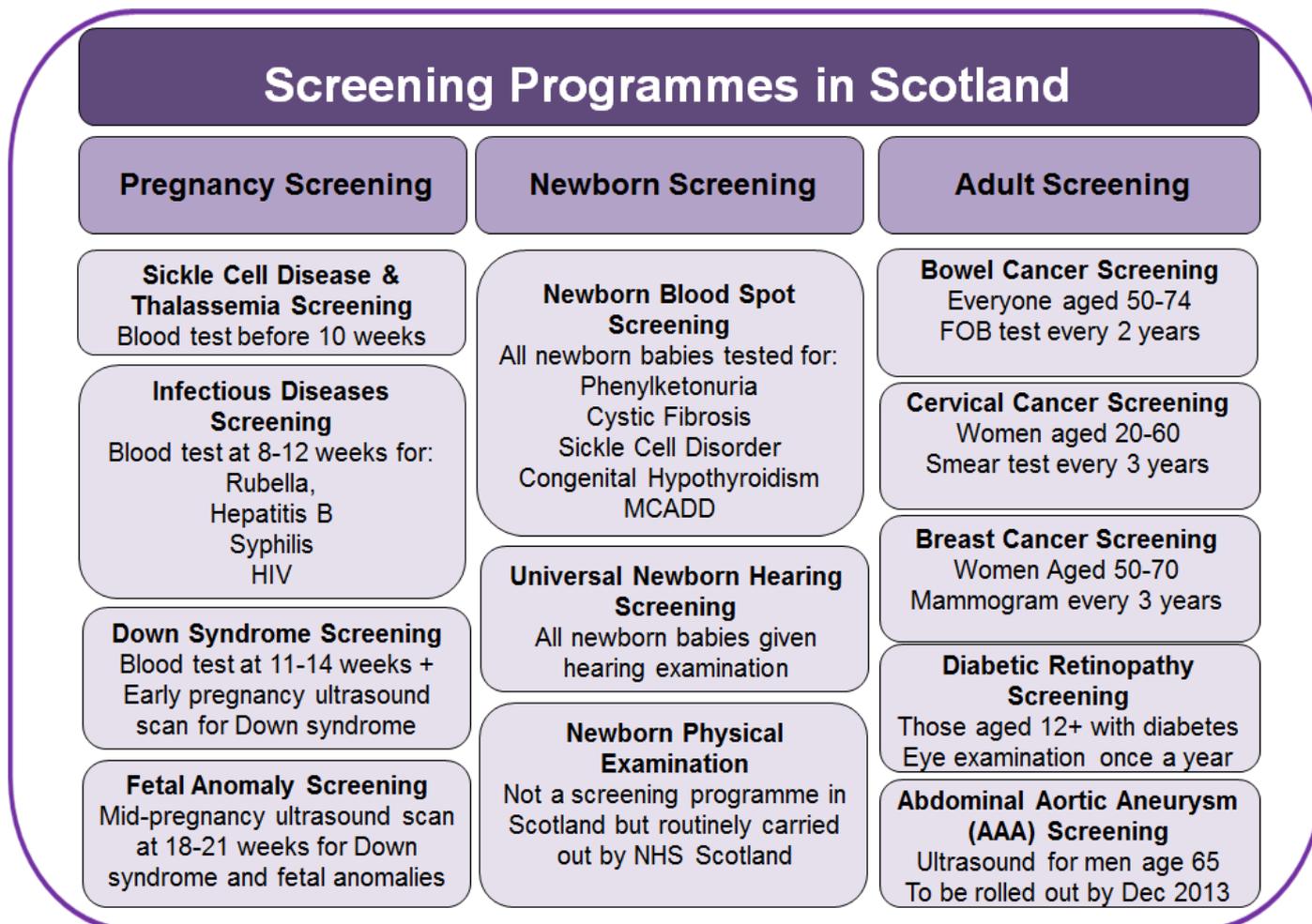
The Screening Programme

- The complete screening programme (test, diagnostic test, treatment) is clinically, socially and ethically acceptable to health professionals and the public
- There should be evidence from control trials that the screening programme is effective in reducing mortality or morbidity. Where screening is aimed at providing information to allow the person being screened to make an informed choice (e.g. Down syndrome screening), there must be evidence from trials that test accurately measures risk.
- The information that is provided about the test and its outcome must be of value and readily understood by the individual being screened
- The benefits of the programme should outweigh the physical and psychological harm
- The cost of the entire programme should be economically balanced in relation to health care spending as a whole. It should offer value for money.
- All other options should have been considered to ensure that no more cost effective intervention could be introduced
- There should be a plan for managing and monitoring the screening programme and an agreed set of quality assurance standards
- Adequate staffing and facilities should be available prior to the programme starting
- Information explaining the consequences of testing, investigation and treatment should be available to potential participants to allow them to make an informed choice
- Public pressure for widening the eligibility criteria & reducing the screening interval and for increasing sensitivity of the testing process should be anticipated. Decisions about these parameters should be scientifically justifiable to the public
- If screening is for a mutation, the programme should be acceptable to people identified as carriers and to other family members

SCREENING PROGRAMMES IN SCOTLAND

In Scotland, there are 3 streams of public health screening programmes: pregnancy screening, newborn screening and adult screening (Figure 3).

Figure 3: Public Health Screening Programmes in Scotland



PREGNANCY SCREENING

There are several national screening programmes available to pregnant women (Figure 3). These are routinely explained, discussed and performed by a midwife or a doctor. In some more complicated cases where there is a family history of a genetic disorder, the woman might be invited for genetic counselling.

Sickle Cell and Thalassaemia Screening

Pregnant women are offered a blood test within the first 10 weeks of pregnancy to screen for sickle cell disorder (SCD) and thalassaemia disorders. SCD affects the shape of red blood cells (the cells which carry Oxygen around the body). The change in shape means that the cells get stuck in small blood vessels and can cause great pain and damage to the baby's body. It can sometimes be fatal. Once detected though, antibiotics and immunisations can help prevent serious illness. Thalassaemia disorders are a group of genetic disorders that affect the body's ability to create new red blood cells. Since red blood cells are needed to carry Oxygen around the body, people with certain types of thalassaemia are very anaemic. They need regular blood

transfusions (every 4-6 weeks) and need injections and medicines for the rest of their lives (NHS Choices, 2012a).

Babies can only inherit a thalassaemia disorder if they inherit the altered form of the gene from both parents. Both parents need to be 'carriers' of the disease i.e. have one copy of the normal gene and one copy of the altered gene. Anyone can be a healthy carrier. It is only when two copies of the altered gene are inherited that it becomes problematic. People are more likely to be 'carriers' of an altered thalassaemia gene if they are from South Asia, East Asia, the Middle East, South America or the Mediterranean. This is because being a 'carrier' in these countries is common as it helps protect against malaria (NHS Choices, 2012a). Polish people also have a high carrier frequency as many people who migrated to Poland many generations ago were from places where malaria was common. Some NHS boards take a family history survey to see if the baby is likely to be at risk of a sickle cell or thalassaemia disorder.

If the baby is found to be at high risk of SCD or thalassaemia, a diagnostic test (amniocentesis or chorionic villi sampling) can be offered to provide a conclusive diagnosis. This is intended to provide the parents with the information to allow them to make informed choices about the pregnancy.

Infectious Diseases Screening

Screening for infectious diseases during pregnancy is routinely offered as a blood test at 8-12 weeks. The infectious diseases screened for are described in Table 7.

Table 7: Infectious Diseases Pregnancy Screening (National Services Division, 2012d).

Infectious Disease	Description
Rubella	Rubella is generally a mild illness. Most people are immunised against rubella infection during childhood. However, some people have not been immunised and if they are infected with rubella during the first 20 weeks of pregnancy, it can lead to deafness in the baby. Early detection and simple treatment can reduce risks to both mother and baby.
Hepatitis B	Hepatitis B is a virus that affects the liver and can be carried in the blood for many years without any signs or symptoms. Hepatitis B infection can be passed from mother to baby at birth. Early detection can prevent babies being infected with the virus which can prevent serious liver disease in adulthood.
Syphilis	Syphilis is a rare sexually transmitted infection that can damage the health of the baby. Early detection during pregnancy can be treated quickly and simply using antibiotics.
HIV	HIV is the virus that can lead to AIDS. HIV damages the immune system and destroys the body's defences against infection and disease. Infected mothers can pass the virus to their babies during pregnancy, birth or via breastfeeding. Women identified as HIV+ during pregnancy will be referred for specialist treatment & medication that can reduce the chance of HIV being passed to the baby.

Down Syndrome Screening

Down syndrome is a genetic disorder that is characterised by an extra copy (i.e. 3 copies) of chromosome 21. It is characterised by learning disabilities and distinct physical features. However, people with Down syndrome, like everyone else, vary a lot in appearance, personality and ability. The severity of the condition will also vary from individual to individual.

Down syndrome can occur in pregnancies of women of any age. However, the older the mother is during pregnancy, the higher likelihood she has of having a child with Down syndrome. For example, a pregnant woman aged 22 has a 1:1450 chance of having a baby with Down syndrome whereas a pregnant 45 year old woman has a 1:45 chance (Barts and The London Medical School, 2012). Down syndrome can also be inherited in a way that all children of a couple will be affected, however this is extremely rare.

During pregnancy, a blood test at 11-14 weeks is routinely taken to look at levels of different proteins in the blood. Levels outside the 'normal' range can indicate a positive screening result for Down syndrome. A second screen for Down syndrome during pregnancy is the nuchal translucency ultrasound scan (early pregnancy ultrasound scan). This scan measures the amount of fluid at the back of the baby's neck and is then analysed by a computer programme. A higher nuchal translucency measurement indicates a positive result for the screening and these pregnancies are deemed high-risk. The early pregnancy ultrasound scan can also detect genetic conditions such as Edwards syndrome¹ and Patau syndrome²; unfortunately however, most Edwards & Patau pregnancies will result in miscarriage.

Pregnant women identified as high risk during screening for Down syndrome will be offered information about Down syndrome and a diagnostic test (see below). If a diagnostic test confirms Down syndrome, the women (and partner) will be able to discuss their options with their midwife. They may also be advised to contact [Down Syndrome Scotland](#), a support group for everyone affected by the condition and for new parents of a Down syndrome child.

Fetal Anomaly Screening

The mid-pregnancy ultrasound scan for fetal anomalies takes place between 18-21 weeks. The aim of fetal anomaly ultrasound screening is to identify:

- Anomalies that are not compatible with life;
- Anomalies associated with high morbidity and long-term disability;
- Fetal conditions with the potential for intrauterine therapy; and
- Fetal conditions that will require postnatal investigation or treatment (National Screening Committee, 2005).

Fetal anomaly screening can detect abnormalities including cleft lip, Patau syndrome, Edward's syndrome, lethal skeletal dysplasia, open spina bifida and anencephalies (NHS Scotland, 2010). Screening has been found to detect 44.7% of all fetal anomalies, although detection rates vary for different anomalies (NHS National Screening Committee, 2005).

¹ Edwards syndrome (Trisomy 18) is caused by an extra copy of chromosome 18 (like Down syndrome which is caused by an extra copy of chromosome 21). It usually results in miscarriage. In cases where live birth is given to a baby, the baby will suffer from severe internal organ problems and the average life expectancy is 2 months (Patient.co.uk, 2011a).

² Patau syndrome (Trisomy 13) is caused by an extra copy of chromosome 13. It usually results in miscarriage. In cases where live birth is given to a Patau syndrome baby, the baby is likely to suffer severe physical abnormality and the median survival rate is 2.5 days (Patient.co.uk, 2011b).

Diagnostic Testing

Those identified during screening as being at 'high risk' can be offered a diagnostic test to confirm if the unborn baby could develop, or has developed an abnormality or serious health condition. Diagnostic testing can be either by amniocentesis or chorionic villi sampling (CVS). These procedures are summarised in Table 8.

Table 8: Comparison of Amniocentesis and CVS as prenatal diagnostic procedures

	Amniocentesis	Chorionic Villi Sampling (CVS)
The procedure	A needle is injected into the womb to take a sample of amniotic fluid (fluid that surrounds the fetus)	A sample of chorionic villi cells are taken from the woman's placenta for analysis
Miscarriage rate as a direct result of the procedure	1 in 100	1 in 50
Time at which procedure can be carried out	15-16 weeks of pregnancy	10-14 weeks of pregnancy

Although CVS carries a higher risk of miscarriage than amniocentesis, CVS can be performed earlier, which would result in a less traumatic termination of pregnancy should a woman (and partner) opt to terminate the pregnancy. The midwife, genetic counsellor or other health professionals will discuss the advantages and disadvantages of both diagnostic procedures with women (and partners) of high-risk pregnancies, should they decide to have a diagnostic procedure.

Pre-implantation Genetic Diagnosis (PIGD)

PIGD is an option for couples who are both carriers of a sickle cell disorder, but do not want to pass this disorder onto their baby. It is similar to in vitro fertilisation (IVF). IVF is a method that helps infertile couples conceive by removing an egg from the woman's ovaries and fertilising it with a man's sperm in a laboratory. Similarly, PIGD removes eggs from a woman's ovaries and these are fertilised with her partner's sperm. The fertilised embryo is then tested for sickle cell disorder and if the results are negative, the embryo is then implanted into the woman's womb. There is no guarantee that PIGD will be successful as there is only a 1 in 5 success rate (NHS Choices, 2012b). In most cases, PIGD is only carried out by private clinics and would cost around £8000 for a course of treatment (NHS Choices, 2012b).

NEWBORN SCREENING

Two screening programmes exist for newborn babies in Scotland. These are the newborn blood spot test and the universal newborn hearing screening programme.

Newborn Blood Spot Test

National Services Division is responsible for monitoring and evaluating newborn blood spot screening for all newborn babies (formerly known as the 'Guthrie test'). National Services

Division has commissioned the Biochemical Genetics Department at the West of Scotland Regional Genetics Service (Southern General Hospital, Glasgow) to operate the Scottish Newborn Screening Laboratory. Its remit is:

- To screen blood samples of all babies born in Scotland (currently around 60,000 per year);
- To ensure the reporting of all results to the proper authorities and the prompt referral of all positive cases for treatment;
- To provide data on incidence of the conditions as required; and
- To review new technology with a view to the incorporation of new tests/methods into the screening programme. (National Services Division, 2012e).

This test identifies babies who may have rare but serious conditions. Early treatment can improve health and prevent severe disability or death. The blood spot test is offered to all newborn babies in Scotland and tests for the genetic conditions set out in Table 9. The test is usually performed by a midwife who will prick the baby's heel to obtain a few drops of blood that is then sent to the Scottish Newborn Screening Laboratory for analysis.

Table 9: Genetic conditions tested in newborn blood spot test (National Services Division, 2012e)

Genetic condition	Incidence for babies born in Scotland	Features
Phenylketonuria (PKU)	1 in 8000	PKU is a rare genetic disorder whereby the baby cannot digest phenylalanine (which is found in most foods). PKU can be treated with a very strict diet that is low in phenylalanine.
Cystic Fibrosis (CF)	1 in 2500	Around 1 in 25 people in Scotland have the altered version of the CF gene. CF occurs when a baby inherits altered copies of the CF genes from both parents. It affects the lungs the most and causes chest infections and problems digesting food.
Sickle Cell Disorder (SCD)	1 in 2500	SCD affects the shape of red blood cells (the cells which carry Oxygen around the body). The change in shape means that the cells get stuck in small blood vessels and can cause great pain and damage to the baby's body. It can sometimes be fatal. However, once detected, antibiotics and immunisations can help prevent serious illness.
Congenital Hyperthyroidism (CHT)	1 in 3500	CHT means that the baby can't produce enough of the thyroxine hormone. This hormone is needed for mental and physical development. Early detection means the baby can be treated with throxine medicine.
Medium Chain Acetyl-CoA Dehydrogenase Deficiency (MCADD)	1 in 10,000	This is a genetic condition where the baby cannot break down certain fats to make energy. This can be problematic when a baby has an illness and can be fatal when a baby has an infection and cannot eat food as the baby cannot burn stored fat.

In 2011/12, the Scottish Newborn Screening Laboratory performed newborn blood spot tests for 58,686 babies. Of the 58,686 screens, there were 8 presumptive positive results for PKU, 41 for CHT, 32 for CF, 5 for MCADD and 9 for SCD and other haemoglobinopathies. These 94 presumptive positive cases were given referral for diagnostic testing. In the same year, screening was declined by parent/guardians in 21 cases for PKU, CHT and MCADD with 23 screens declined for CF and 27 screens declined for SCD (National Services Division, 2012f).

Newborn Hearing Screening

The [Universal Newborn Hearing Screening Programme](#) is a hearing test for newborn babies. It is usually performed before a baby leaves hospital. Hearing loss in one or both ears affects around 1 in 1,000 babies and most of these babies are born into families with no family history of hearing loss. Results are usually given at the time of the test (National Services Division, 2012g).

Newborn Physical Examination

In England, the [NHS Newborn and Infant Physical Examination \(NIPE\) Screening Programme](#), offers parent/guardians the opportunity to have a physical head to toe examination of the baby to check for any abnormalities at 72 hours after birth and then again at 6-8 weeks of age (National Screening Committee, 2012c). It includes a detailed examination of the baby's eyes, heart, hips and testes in boys. In Scotland, newborn and infant physical examination is not a national screening programme but it is routinely carried out throughout NHS Scotland (National Screening Committee, 2012d).

ADULT SCREENING

There are five adult screening programmes in Scotland. These are for bowel cancer, breast cancer, cervical cancer, diabetic retinopathy and abdominal aortic aneurysm.

Bowel Cancer Screening

Bowel cancer (sometimes known as colorectal cancer) is the third most common cancer in Scotland after lung and breast cancer and is the second most common cancer in men in Scotland. It is a major public health concern as Scotland has a higher rate of bowel cancer than any other Western country (ISD Scotland, 2012a). The [Scottish Bowel Screening Programme](#) offers a bowel screen to everyone in Scotland, every 2 years, between the ages of 50 and 74. In England and Wales, the equivalent programmes are offered to those aged 60 to 74 and the Northern Ireland equivalent is offered to those aged 60 to 69 (National Screening Committee, 2012e). Younger people who have certain genetic conditions or who have a family history of bowel cancer may be offered screening at an earlier age.

Screening for bowel cancer looks for early signs of cancer in otherwise healthy people who have no symptoms of the disease. Screening for bowel cancer is relatively straightforward and requires only small samples of bowel motions (faecal occult blood (FOB) testing). These can be taken at home using the kit sent by the NHS board and can be posted back free of charge. For every 1,000 people that take the FOB test, around 20 will have an abnormal result. They are offered a colonoscopy for diagnosis and of these people, around 2 will be diagnosed with bowel cancer (Cancer Research UK, 2012). Regular bowel screening has been shown to reduce the risk of dying from bowel cancer by 16% (Cancer Research UK, 2012).

In Scotland, uptake of bowel cancer screening from those eligible over the 2 year period from November 2008 – October 2010 was 53.7%. Females are more likely than males to uptake bowel cancer screening with 57.2% of eligible females taking the test compared to only 50.0% of males during the same two-year period (ISD Scotland, 2012c). Uptake of bowel cancer screening is also affected by social factors. Only 38.7% of eligible males living in the most deprived areas in Scotland took part in the screening programme (ISD Scotland, 2012c). Of everyone screened, 2.3% were given a positive result and were referred for further diagnostic testing. The screen positive rate varies geographically across Scotland with the highest bowel cancer screen positive rates being from NHS Lanarkshire and NHS Greater Glasgow & Clyde. The lowest screen positive rates are in catchment areas of NHS Fife, NHS Grampian, NHS Tayside and NHS Dumfries and Galloway. The screening programme detected twice as many cancers in men than it did in women. Of the people who were screen positive, diagnostic testing confirmed that 8% of the men and 6.4% of the women had a form of bowel cancer (ISD Scotland, 2012c). Bowel screening can also detect small growths in the bowel called polyps, which can have the potential of becoming cancerous.

Breast Cancer Screening

Breast cancer is the most common cancer diagnosis for women in Scotland, although it is the second most common cause of cancer death in women (after lung cancer). In Scotland, over 4000 women are diagnosed and around 1,000 women and 10 men die of breast cancer each year (Breast Cancer Care, 2011).

The Scottish Breast Screening Programme is provided by 6 breast screening centres in Scotland (Aberdeen, Dundee, Edinburgh, Glasgow, Inverness and Irvine) supported by 19 mobile units for remote areas (National Services Division, 2011). Currently, around 80% of the screening takes place on the mobile units (National Services Division, 2011). Women aged between 50 and 70 are invited for a mammogram every 3 years. Women over the age of 70 are not routinely invited for screening, but can request a screen. Younger women with a family history of breast cancer may also be invited for regular screening.

Figures for 2010-11 show that the Scottish Breast Screening Programme detected around 1,700 cases of breast cancer and uptake of screening for those eligible is around 74.9% (ISD Scotland, 2012d). In 2010-11, 9.3 cancers were detected per 1000 screens performed. In total, the screen identified around 1,700 cases of breast cancer (ISD Scotland, 2012d).

Cervical Cancer Screening

Cervical Cancer is the most common form of cancer for women under the age of 35 in Scotland (NHS Greater Glasgow & Clyde, 2011). The Scottish Cervical Screening Programme offers screening (pap smear test) to all women in Scotland between the ages of 20 and 60 every 3 years. The smear test aims to detect abnormal changes in a woman's cervix which if left untreated could develop into cervical cancer. It is estimated that cervical screening saves around 5,000 lives in the UK each year and prevents 8 out of 10 cervical cancers from developing in the first place (Health Protection Scotland, 2012).

In 2010/11, uptake of cervical cancer screening within the past 3.5 years was 73.6% of eligible women across Scotland. Of the 379,355 tests processed in 2010/11, 97% were of a satisfactory quality. Of satisfactory results, 90.8% were deemed low-risk (negative result) with the remainder referred for further diagnostic testing (ISD Scotland, 2012e).

The human papillomavirus (HPV) can cause cervical cancer. It is a very common virus and can spread via sexual intercourse. Because it is so common, most people will be infected by HPV at

some point of their life (Immunisation Scotland, 2012). There are different types of HPV and it is two special types of HPV that cause around 70% of cervical cancers (Immunisation Scotland, 2012). The HPV vaccination was introduced in 2008 for girls aged 12-13 years (S2 High School girls). Girls need three doses over a period of six months to provide the best protection. As of September 2012, the vaccination will protect against the two most dangerous HPV strains that cause around 70% of cervical cancers, whilst providing protection against another two types of HPV and genital warts. However, the vaccination does not provide protection against all types of HPV and therefore regular screening is important (Immunisation Scotland, 2012).

The most recent figures show that in 2010-11, uptake of HPV vaccination for girls in S2 at High School was 91.8% for the first dose, 90.2% for the second dose and 81.0% for the third dose (ISD Scotland, 2012f). Data collected from 2008 show that a number of girls complete the immunisation programme after the end of the school year in which they were offered the vaccine. Therefore, uptake rate is likely to be higher than published as some girls will not get their third dose until 2012-13 when they are in S3 at school (ISD Scotland, 2012f). Updated figures are to be published by Information Services Division in September 2012.

Diabetic Retinopathy Screening

There are approximately 150,000 people in Scotland with diabetes and around 5-10% of diabetics have a sight-threatening retinopathy (Facey *et al.*, 2002). Retinopathy is a condition that can cause severe damage to the eye and can result in blindness. Diabetic retinopathy is the biggest single cause of blindness and visual impairment in Scotland amongst people of working age (Facey *et al.*, 2002). The rising prevalence of diabetes means that it is likely to remain a major health and economic problem in Scotland. If diabetic retinopathy is detected and treated early, damage can be minimised (National Services Division, 2012h). The [Diabetic Retinopathy Screening Programme](#) offers screening to everyone over the age of 12 with diabetes. They are invited for an eye screen once a year. In 2010-11, uptake of diabetic retinopathy screening from those eligible was 84.8%, despite a 4% increase from the previous year in the number of diabetics in Scotland aged 12 and over (NHS Scotland, 2011). Of the 238,383 people screened in 2010-11, 3.9% were referred to ophthalmology for further examinations (NHS Scotland, 2011)

Abdominal Aortic Aneurysm (AAA) Screening

In July 2012, the Scottish Government announced its intention to roll out an AAA screening programme by 2013 for all men aged 65. An AAA is an enlarged part of the aorta that is in the abdomen. It usually has no symptoms unless it bursts, in which case it can be fatal. AAA is very common in Scotland with around 284 men aged 65 or older dying from AAA each year, with around 60% of these deaths preventable by screening (Scottish Government, 2012b). Screening will only be offered to men age 65 as 95% of ruptures occur in this group (Scottish Government, 2012b). The screening will use ultrasound to measure the size of the aorta. It is estimated that once the new screening programme is fully established it will prevent around 170 deaths by AAA each year (Scottish Government, 2012b). The aim of this screening programme is to:

- Reduce the mortality rate associated with the risk of aneurysm rupture in men aged 65 years and older; and
- Shift the balance of care from reactive emergency management of rupture to elective management through early diagnosis (Healthcare Improvement Scotland, 2011).

Screening can identify men with AAA and for those with a large AAA, surgery or lifestyle changes can reduce the chance of the AAA rupturing. Those with a small or medium AAA will be called back for regular surveillance.

APPENDIX 1: PROFESSIONAL GENETICS ASSOCIATIONS

Organisation	Aims
<p>British Society for Human Genetics (BSHG)</p> <p>The BSHG is the independent umbrella society for UK professional organisations for human genetics. Constituent organisations of BSHG are the ACGS, AGNC, CGC, SGPPH and the CGG (all described below).</p>	<p>BSHG aims to:</p> <ul style="list-style-type: none">• Advance the science of human genetics;• Promote study and research into human genetics as it relates to health and disease;• Promote public awareness of human genetics as it relates to health and disease;• Support, guide and inform the professions contributing to applications of genetics in the health care systems of the UK;• Offer informed opinion on issues of public interest in relation to genetics;• Co-ordinate and assist as appropriate the activities of the Constituent Groups (BSHG, 2012).
<p>Association of Clinical Scientists (ACS)</p> <p>The ACS is an association for clinical scientists and trainee clinical scientists throughout the UK. The main role of the ACS is to assess trainees as a preliminary to registration with the HPC.</p>	<p>ACS aims to:</p> <ul style="list-style-type: none">• Promote, advance and encourage the study and practice of the application of science in the prevention, diagnosis and control of illness, disease and disability;• Establish, uphold and improve the standards of qualification, training, competence and conduct of Clinical Scientists in the United Kingdom;• Work with official bodies including Health Professions Council (HPC) on matters relating to the above;• Liaise with relevant professional bodies to set standards for training and training centres to include availability of appropriate resources, regular assessment, pastoral care and mechanisms for addressing students training issues (ACS, 2009).
<p>Association for Clinical Genetic Science (ACGS)</p> <p>The ACGS is a new professional body that is in the</p>	<p>ACGS aims to:</p> <ul style="list-style-type: none">• Promote and advance the application and practice of clinical genetic science such that the discipline best serves the needs of patients and their families;• Collaborate with other organisations with similar objectives to the Association,

process of being firmly established. It is being formed as a result of a merger between the Association for Clinical Cytogenetics (ACC) and the Clinical Molecular Genetics Society (CMGS). This new professional body has been formed following the recognition that as genetics and genetic technology is a rapidly advancing field, the specialities of molecular genetics and cytogenetics are moving ever closer together (ACGS, 2012a). The ACC and the CMGS are in the process of being legally dissolved whilst the ACGS appoints a new chair and writes a constitution.

- both within the United Kingdom and elsewhere, to advance practice;
- Develop and publish professional standards and national best practice with regard to clinical genetic science;
- Promote research, development and innovation in clinical genetic science and genetic testing and facilitate translation into practice to benefit patients and their families;
- Promote public awareness of clinical genetic science as it relates to health and disease;
- Provide informed opinion to Government and other external bodies on issues relating to the application and practice of clinical genetic science;
- Actively promote appropriate training, education and continuing professional development for members of the Association;
- Provide a unified voice on issues that concern the Association and its members;
- Maintain the financial viability of the Association in order to support these objectives (ACGS, 2012b).

Association of Genetic Nurses and Counsellors ([AGNC](#))

AGNC is the society that represents genetic nurses, counsellors, associates and other non-medical staff working within clinical genetics whose work involves contact with families or individuals affected by or at risk of a genetic condition.

AGNC aims to:

- Provide support for professionals working in the field;
- Provide forums for education and scientific meetings;
- Represent the views of the profession;
- Prescribe good standards of clinical practice;
- Liaise and collaborate with other clinical and scientific colleagues in the field;
- Provide channels of communication within the profession (AGNC, 2012).

Clinical Genetics Society ([CGS](#))

The CGS is the society for medically qualified doctors who specialise in clinical genetics and other professionals involved in the care of individuals and families with genetic conditions.

CGS aim to:

- Advance and promote the science and practice of clinical genetics;
- Bring together workers who have a common interest in clinical genetics;
- Understand, prevent, cure and alleviate conditions with a genetic aetiology;
- Publish and disseminate reports, statements, and research findings
- Promote and facilitate education for the genetics community, other health care professionals, those outwith the profession, and the wider public;
- Encourage high standards of training for professionals within clinical genetics;

- Facilitate research into basic human genetics and genetic disorders;
- Maintain excellent links with patient groups;
- Continue constructive dialogue with government and other politicians on genetics-related issues (CGS, 2012).

Society for Genomics, Policy and Population Health ([SGPPH](#))

SGPPH aims to discuss and raise awareness of the policy, ethical, legal and social issues raised by genomic science and technologies (SGPPH, 2012).

SGPPH is a forum for those with an interest in genetics and molecular science and its impact on health. Its membership includes lawyers, philosophers, social scientists, public health professionals, policy makers, geneticists and molecular scientists.

Cancer Genetics Group ([CGG](#))

CGG aims to improve the quality of care of patients and their families with any condition resulting in hereditary tumours.

Membership of CGG is open to those with an interest in hereditary cancers.

APPENDIX 2: KEY SUPPORT ORGANISATIONS FOR WOMEN WITH 'HIGH RISK' PREGNANCIES

Organisation	Support available
<u>Antenatal results and choices (ARC)</u>	Provides support and information to expectant and bereaved parents throughout and after the antenatal screening and testing process.
<u>Contact a Family Scotland</u>	Provides information, support and advice to parents and carers of children with any special need or disability. Campaigns to improve the circumstances of families with disabled children and for inclusiveness.
<u>Down Syndrome Scotland</u>	Provides information, services and support people with Down syndrome, their families and carers.
<u>Positively Women</u>	Offers peer support, advice, information and advocacy for HIV positive women and provides special support for HIV positive women who are pregnant.
<u>Scottish Spina Bifida Association</u>	This is an organisation that provides a support service to parents of children with spina bifida and related conditions. It aims to empower those affected to make informed choices and decisions.
<u>Sickle Cell Society</u>	Provides information, counselling and care for people with sickle cell disorders and their families.
<u>SOFT UK</u>	Helps those affected by Edward's and Patau syndrome, chromosomal abnormalities and related disorders through prenatal diagnosis, termination of pregnancy / birth of the baby, caring for the baby and bereavement.
<u>UK Thalassaemia Society</u>	Provides information, education and research to those affected by, or working with thalassaemia.
<u>Waverley Care</u>	Provides support to people living with HIV and Hepatitis C, and to their partners, families and carers. They also aim to raise awareness of these conditions and their prevention.
<u>Healthtalkonline</u>	Provides video interviews and descriptions of people's experiences of health procedures, tests and conditions. Includes experiences of pregnancy screening, diagnostic testing, termination of pregnancy following a diagnosis of fetal abnormality and experiences of living with different conditions.
<u>Health in my language</u>	Website that provides information on a wide range of health topics and health services in Scotland in different languages.

APPENDIX 3: KEY SUPPORT ORGANISATIONS FOR NEWBORN SCREEN POSITIVES

Organisation	Support available
<u>The National Society for Phenylketonuria (NSPKU)</u>	NSPKU provides help and support to people with PKU, their families and carers and campaigns.
<u>Cystic Fibrosis Trust</u>	Provide practical support and advice to people with Cystic Fibrosis and their families. Support individuals to ensure they have the best level of care.
<u>Sickle Cell Society</u>	Provides information, counselling and care for people with sickle cell disorders and their families.
<u>British Thyroid Foundation</u>	Support people with thyroid disorders and helping their families and people around them to understand the condition.
<u>Children Living With Inherited Metabolic Diseases (CLIMB)</u>	Provide confidential support, advice and information about MCADD and other metabolic disorders and promote awareness of them.
<u>National Deaf Children's Society (NDCS)</u>	Dedicated to removing barriers for deaf children and young people. Provide a support network to new parents and carers of deaf children.
<u>Contact a Family Scotland</u>	Provides information, support and advice to parents and carers of children with any special need or disability. Campaigns to improve the circumstances of families with disabled children and for inclusiveness.

APPENDIX 4: KEY SUPPORT ORGANISATIONS FOR ADULT SCREEN POSITIVES

Organisation	Support available
<u>Bowel Cancer UK</u>	Provide practical support and advice about bowel cancer, raise awareness about the disease and campaign for best treatment and care.
<u>Breast Cancer Care</u>	Provide information and support for anyone affected by breast cancer.
<u>Jo's Cervical Cancer Trust</u>	Provide information about cervical cancer and support and friendship to women of all ages affected by cervical cancer.
<u>Macmillan Cancer Support</u>	Provide practical, medical and financial support to people affected by cancer and push for better cancer care.
<u>Diabetes UK</u>	Cares for, connects with and campaigns on behalf of all people affected and at risk of diabetes in the UK.
<u>Circulation Foundation</u>	Promote research into causes, treatment and prevention of vascular disease in the UK. Provide information and support for patients and families affected by vascular disease, including abnormal aortic aneurysms.

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