Report on Websites Supporting the Work of the Regional Genetics Services

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September 2003
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This report is published by the Public Health Genetics Unit, a core facility of Cambridge Genetics Knowledge Park. The project was funded by The Wellcome Trust and the Department of Health.

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1 Introduction

This is an exciting and challenging time for all who are involved in the practice of clinical genetics—as professionals, patients or carers. The rapid roll call of “breakthroughs” raises hopes—and expectations—about the ability of clinical practice to deliver more and more effective care. It is a time of great opportunity but also risk—that the profession fails to keep abreast of developments through pressure of time, resources or lack of information; that the public expectations are not met; that the potential of the new medical knowledge fails to be fully exploited.

This report examines how practitioners in one area of the health profession—the regional clinical genetics services—are attempting to harness the considerable power of the online medium to add another layer of value to their activities. The report is based not simply on a survey of the sites from a distance, but also on a series of discussions with health professionals about how they go about their work, what their needs are and what might make an online service more or less useful. It does not seek to offer a template of how a website should be created, but by identifying elements of best practice, assessing some of the gaps in provision and considering some of the constraints that make the delivery of a service less effective, it is hoped that it might be a useful resource to any organisation in this field which is planning or developing its own online resources.

2 Context

The Public Health Genetics Unit (PHGU) was commissioned by the Wellcome Trust to identify the needs for education in genetics among healthcare professionals and to develop a national strategy to meet these needs. It is recognised that discoveries about the impact and influence of genetics are emerging at a speed that threatens to outstrip the health profession’s ability to keep abreast of developments. There is an urgent need to establish areas of weakness, to plan resources that help build knowledge and competencies and to find the best means of distributing them to healthcare professionals via formal training and as part of their continuing professional development. The objective is both to improve clinical practice and to raise general levels of genetics literacy.

3 Purpose

The purpose of this report is to support the work of strategy development by assessing the role of online resources in providing access to information, clinical support and education on genetics for health professionals, through a close study of the websites created by the Regional Genetics Services. The report will seek to identify the needs of health professionals, to identify the kinds of online content which best meet those needs, and to consider the gaps in the current provision. The report will try to draw some broad conclusions about the role of online resources in the future strategy.
4 Objectives

The objectives were:

- To review the websites of clinical medical genetics centres within the UK and in addition examine the role of websites for organisations representing groups of healthcare professionals such as the British Society for Human Genetics, the Royal College of GPs and the National Electronic Library for Health.
- To provide an overall summary and critique of these sites.
- To identify examples of current best practice.
- To consider how the sites might be developed to have a stronger role as an educational resource.
- To consider some of the issues in planning, resourcing, producing and maintaining effective online content.
- To consider the most effective relationship between locally produced online resources and centrally provided, generic material.

5 Methodology

A list of the Regional Genetics Services is available from a number of different sources such as the British Society for Human Genetics. Where no web address was given, a search via an established search engine such as Google was used to identify the site. Where this failed to produce results, a search of the website for the hospital or institution with which the Regional Genetics Service was associated was carried out.

In addition, some examples of sites supporting the work of related departments such as cancer services were examined to find further evidence of best practice and to assess how well the service was integrated with the work of the Clinical Genetics service.

As part of the wider work of strategy development, a number of working groups representing different professional disciplines have been conducted with a view to identifying needs and current provision of educational materials. In addition to the overall strategy work, these working groups have been opportunities to establish current usage, behaviour and need for online resources.

An in-depth user-study of the West Midlands site was conducted with a range of health professionals from the area in order:

- To look critically at their own and features of other websites.
- To consider the functions of the website for a broad cross section of professionals' needs.
- To consider how the site might be developed to meet educational needs.
- To consider some of the practical issues of planning, resourcing and maintaining such a site.

In addition a number of individual interviews were carried out with people responsible for planning and creating some of the other sites covered in this report.
These interviews covered the following sites: British Society for Human Genetics; National Electronic Library for Health; West of Scotland Regional Genetics Service (Institute of Medical Genetics, Yorkhill Academic Campus, Glasgow); East Anglian Medical Genetics Service (Addenbrooke’s NHS Trust).

6 Existing sites

The provision of online material about the Regional Genetics Services ranges from the non-existent, or merest mention of the service on a hospital site, through basic descriptions of the physical services offered, to more extensive sets of resources designed to support and improve clinical practice. Approximately 50% of the services have either no web presence, or a passing mention; 25% have basic sites with simple content; 25% have more ambitious, deeper content. This report will concern itself with the last two groups. It should be noted, however, that no survey of sites could be comprehensive. The proliferation of online banners announcing “site under construction” is evidence that more sites are being planned and developed. The thinking behind some of the sites planned for the future has informed this report. Furthermore, it is possible that some sites might not have been located at all, particularly some of those located on the NHS intranet. Although some were notified to us we found these difficult to search. The usual means of investigating and accessing the sites are described below.

7 Criteria

The websites were assessed according to the following criteria:

- Access
- Navigation, design, functionality
- Audience focus
- Range and quality of content, including links
- Potential to develop as an educational resource

8 Access –finding it

Lists of Regional Genetics Services are published on a number of websites aimed at a variety of audiences. Sample sources were assessed in the following groups:

- Professional bodies, societies and groups. Listings appear, with varying amounts of detail, on a number of such sites, including the British Society for Human Genetics (BSHG) website, the Royal College of General Practitioners website and the Oxford Primary Care Group. One helpful feature on the BSHG site was a clickable map offering access to information on services throughout the UK, with local maps for the location of each service.
- Special interest groups. These include the Genetics Interest Group (GIG), and a range of sites offering support to patients and families suffering from specific conditions, e.g. Contact a Family.
- Public interest sites e.g. the BBCi Health website
- Clinical Genetics sites. Some regional services publish full lists of comparable services in other parts of the country.
The number of sites listed varies between 20-30, depending on whether some of the supporting clinics and laboratories are included (a list of sites reviewed appears as an appendix). These lists generally give addresses and contact telephone numbers. Few lists include website addresses, and those which do are incomplete. None of the lists were up-to-date with new sites and it is possible that the same out-of-date information is being recycled between sites with none taking a lead on editing and updating the information.

A search via an established search engine like Google will usually feature one of these lists in the top ten results, although occasionally the individual site itself is higher in the search hierarchy.

Trying to find the site without knowing the web address (URL) beforehand can be difficult and time-consuming. This is often a reflection of complex organisational structures and hierarchies within the parent body, such as a hospital or university. Most, but not all, websites of the “parent” organisation have a search facility, but there is no overall consistent approach to “taxonomy” and navigation.

### Examples of Web Pages Listing Regional Genetics Services:

**Google** search for West Midlands Regional Genetics Service  
[www.google.com/search?hl=en&ie=UTF-8&oe=UTF-8&q=west+midlands+regional+genetics+service&btnG=Google+Search](http://www.google.com/search?hl=en&ie=UTF-8&oe=UTF-8&q=west+midlands+regional+genetics+service&btnG=Google+Search)

**British Society for Human Genetics**  

**Genetic Interest Group**  
[www.gig.org.uk/services.htm](http://www.gig.org.uk/services.htm)

**Contact a family**  
[www.cafamily.org.uk/gencentr.html](http://www.cafamily.org.uk/gencentr.html)

### 9 Design, navigation and functionality

All of the sites reviewed are designed for narrowband access. Some sites comprise a basic text document written for other purposes and transcribed for the web. Other sites were specially created for the web and offer a more sophisticated range of navigational tools.

Examples of the informational features and formats in common usage include:

- Hyperlinks within the site and to external sites
- Downloadable, reusable materials such as fact sheets, forms, posters, leaflets and frequently asked questions (FAQs)
- Graphics, still photography and simple flash animation
• Diagrams and flow charts
• Interactive maps
• Most, but not all, sites had a search function to help navigate the site.
• There were a few examples of directive structured content such as a “decision tree” that is designed to guide the user through a decision making process e.g. making a risk assessment or deciding whether or not to make a referral. There was no evidence of more sophisticated software to support these processes. It is, perhaps, unlikely that such facilities would be available on a public area of a site.

Most sites use basic, freely available software programmes. Some sites offer free downloads of the software needed to run some of the content, such as Acrobat for portable document files (pdf).

Most sites appeared to function within their own terms. There were, however, a number of examples of broken links. Most downloads were relatively fast, probably a reflection of the relatively simple design and software programmes incorporated.

All of the sites visited have open public access, although a small number have password-protected areas or links to an intranet site where deeper content can be published, more sophisticated software programmes such as Java script run, or more sensitive information handled. Typically, users of these password-protected areas might comprise specialists, GPs or students linking through to university course content.

10 Audience focus and needs

The websites address a range of audiences with different needs:

• Healthcare professionals who provide a service directly to patients or who act as gatekeepers to specialist services. These roles include doctors (GPs, non-genetic specialists, public health physicians), nurses, midwives and health visitors, dieticians and pharmacists. Some sites also serve professional bodies representing communities of interest.
• Those involved in education, both as teachers and students
• Those involved in the planning and management of NHS resources
• Patients and their families

Some sites make a clear statement of intent to cater exclusively for health professionals and adopt a level of language and knowledge deemed to be appropriate for this specialised audience. However, given the range of job functions encompassed, as well as the widely differing levels of competency in genetics even within the same professional area, exactly what constitutes an appropriate level of language and understanding is a matter of some debate.

Other sites acknowledge the open nature of the web and the hunger for information on the part of patients, their families and the wider public. These sites publish content which can either be used directly by members of the public, or can be
mediated to patients, for instance through a GP or nurse, to explain the services and choices available to them.

10.1 Needs of healthcare professionals

It is estimated that 90% of GP surgeries are computerised and that 60% of general practitioners use computers in the consultation (source: Oxford Primary Care Genetics Group: Topics in Primary Care). The working groups and the Birmingham user study confirmed the widespread use of the web and identified a number of core needs among healthcare workers in different disciplines to support best clinical practice:

- Background content that explains fundamental concepts like inheritance, supported with genetic conditions most commonly encountered in practice, such as familial cancers.
- Material which helps develop an understanding of the impact of genetics on many of the most common conditions.
- Health professionals need help to understand the paradigm shift which has seen genetics move from being a branch of medicine concerned with rare, single gene disorders to a discipline dealing with complex, multi-factorial influences underpinning many areas of medicine.
- Health professionals say they require accessible and fit-for-purpose material with a practical rather than theoretical approach. Too much material can be overwhelming and unhelpful. Instead of striving to be comprehensive it is better, as one respondent put it, to stimulate the practitioner to ask the question “can there be a genetic factor at work here?”
- Disease-based case studies covering a range of conditions likely to be encountered in clinical practice. Some conditions, such as cancer, are relatively well catered for with online resources but there is a dearth of contextual information in other areas.
- Explanation of basic genetics terms and concepts in an easy-to-use format.
- Easily accessible information on where to go to get help and support in making a diagnosis and planning interventions.
- Useful, up-to-date information on local services and contacts, processes and procedures involved in referral, consultation and testing.
- Sets of online resources that support clinical practice. These resources include: online “tools” that assist in compiling and recording family history; material that offers support in understanding, assessing and communicating the different categories of risk; “decision support tools” that allow for the effective planning of preventive strategies; a clear explanation of the guidelines for referral, including those at national and local level.
- Help with finding the most effective and reliable online sites from the vast number on offer.
- Information on conditions and processes that the professional can “mediate” to patients and their families to involve them in their own care and allow the patient to make informed choices.
- Material that addresses the wider social, legal and ethical issues arising from genetics such as confidentiality and consent, social impact on family members, issues of employment or insurance.
• Resources that allow the health professional to offer follow-through care beyond the first referral and ongoing support to patients and their families.

10.2 Needs of educators and learners

These were considered to be:

• Clinical Geneticists have an increasing role in educating fellow professionals. They need material which helps “demystify” genetics and increases general subject literacy
• Materials for formal education that integrates genetics into the curriculum of subjects in a way that avoids “curriculum overload”. Summaries of the core genetics competencies.
• Practically-focused, problem-based learning materials that allow professionals to update their knowledge as part of their continuing professional development
• “Just-in-time” information that tells people what they need to know to look after their patients without overloading them with facts and theory.

10.3 Needs of health service managers

These were considered to be:

• General education about the importance of genetics
• Information to help them to understand the language and concepts so that they can engage in the debate about service development and resource allocation.
• Material which helps them to understand the likely demand for resources and to plan accordingly
• Content which helps them manage the demand for services, for instance by publishing guidelines for appropriate referrals to support a GP in making a decision whether or not to refer.

10.4 Needs of patients and their families

These were considered to be:

• Information on a range of conditions, tests and therapies
• Lifestyle advice and information on preventive strategies
• A patient-focused explanation of the services, processes and support systems involved in their care, and information on management of their condition beyond the initial interventions.
• Information and contact details for support organisations, online or otherwise.
• The ability to contribute and to be involved in the process. The availability of information on genetics in the public domain, due largely to the growth of websites, has produced a further paradigm shift whereby the patient is often better informed about their condition than the health professional. There is the potential to involve the patient more proactively in the planning and management of their care.
• The emergence of genetics into the public domain has led some people to hope for a “miracle cure”. There is a need for material that can help manage expectations of what genetic medicine can and cannot yet achieve.

• Conversely, there are many anxieties about aspects of genetics which people find sinister, which may prejudice them against genetic medicine. There is a need for contextual materials that explain and reassure.

From all groups of users there is the demand that online content should be “at the right level”. Given the wide range of users it might appear difficult to find a common standard. However, in discussions with health professionals from a number of fields, and with patients representatives, a number of principles emerged:

• Don’t assume knowledge. Given the shortfalls in knowledge that the ongoing strategy work is exploring, it is better to err on the side of better basic information to build broad competencies than on the side of creating more and more content. More sophisticated, specialised content can be delivered via appropriate links.

• Language needs to be clear and accessible, and as jargon-free as possible. Online resources such as a glossary of genetics terms are useful here.

• Even content that is aimed at health professionals should feel accessible to patients. Content that can explain complex conditions and processes clearly and straightforwardly will both reinforce the health professional’s own knowledge and assist them in explaining those processes to patients.

11 Review of content

The following section deals in more detail with the kinds of content published on the sites under review. It seeks to identify common elements and features and to establish some examples of best practice. The content is organised and reviewed under the following broad headings:

• Description of the local Regional Genetics Service
• Information on the associated laboratories
• Information for health professionals
• Information for patients and their families
• Background on specific diseases and conditions
• Material related to social, legal and ethical issues
• Links to further resources
• Information on education and training
• Issues and future developments

11.1 Description of the Regional Clinical Genetics Service

Many Regional Genetics services do not have a website. The sites that do exist vary considerably in scope and quality, and this is often a reflection of local decisions to commit resources, or the availability of the enthusiasm or the necessary expertise within the unit. However the following useful features appear to be common to many of the sites:
• A simple definition of the purpose of the service. This might involve contextual information on the prevalence of genetic factors in many types of disease and a description of the services offered: E.g. “providing diagnosis, risk estimation, genetic counselling, management and support to individual patients and families with genetic disorders, through a family based diagnostic approach and specialised counselling skills” (West Midlands Regional Genetics Service).

• Information on accessing the service. This might include:
  - Contact details for named members of staff, with address and telephone numbers. Some sites also publish e-mail addresses for key staff: to do so implies a commitment to respond in a timely manner.
  - Information about members of staff. This ranges from functional information on areas of specialist interest and research expertise, to personal material like photographs that help to give the service a human face.
  - Details of the location of services. These might include click-able, interactive maps of local services and clinics within a region and simple site maps offering directions and transport details for patients.
  - Details of clinic hours and waiting times for appointments.

• The description of the kind of patient likely to be referred and common reasons for referral. This can be expressed in terms of the type of symptoms or disease suspected, the processes involved, or the background of the patient and their family.

• A description of the kinds of diagnosis offered, for instance clinical diagnosis of genetic conditions, pre-symptomatic diagnosis of single gene disorders and prenatal diagnosis.

• A description of what happens during a consultation and what a patient might expect from the process.

• An explanation of the role of genetic counselling in following-up the diagnosis, for instance by providing medical information to patients about their condition; discussing risk to themselves and their children; explaining the testing and screening procedures available; helping patients to make informed choices; discussing with them strategies and support for managing their condition.

• An opportunity to state the values and approach which underpins the delivery of the service, e.g. family-based; non-directive; recognising the importance of consent and confidentiality; the commitment to deliver a high quality and timely service, and to communicate effectively with patients.

• An integrated service for the user. There is sometimes good and relevant content available from other departments, but a failure to create links to websites run within the same hospital. Examples of this organisational compartmentalisation or fragmentation include the absence of links between the work of some Regional Genetics units and Cancer Services. The most effective sites are those that align complementary resources to provide a seamless service to the user.
Examples of the features described above:

**West Midlands Regional Genetics Service**
www.bwhct.nhs.uk/clinicalgenetics/aMainour_services.htm
www.bwhct.nhs.uk/clinicalgenetics/amaingenticcons.htm
www.bwhct.nhs.uk/clinicalgenetics/amainhowtorefers.htm

**Liverpool Women’s Hospital**
www.lwh.org.uk/clinical_services/genetics/clinical/who_see.html
www.lwh.org.uk/clinical_services/genetics/clinical/map.html

**West of Scotland Regional Genetics Service**
www.gla.ac.uk/departments/medicalgenetics/nhs/medicalstaff.htm

**Medical Genetics Service for Wales**
www.uwcm.ac.uk/medicine/medical_genetics/service/patient.html

**Central Manchester and Manchester Children’s University Hospital**
www.cmht.nwest.nhs.uk/patient_info/waiting/results.asp?gpwaitRS=GEN

11.2 Information on Laboratories

Most Regional Genetics sites include information on the work of the associated Molecular Genetics and Cytogenetics laboratories involved in carrying out further tests on patients referred to the service. Much of the content is aimed at the healthcare professional and assumes a high level of medical and technical literacy. Some sites also include material useful for explaining the processes to patients. Examples of material relating to the work of the laboratories include:

- Explanations of the nature of the service (e.g. Molecular and Cytogenetics) and the purpose of the work carried out in the laboratory.
- A description of the services currently offered. This usually includes the kind of patient likely to be referred for testing; a definition of the conditions catered for and the type of tests carried out, with their limitations. Where tests are not available locally, some sites provide links to additional sources of information.
- Information on the local organisation of the laboratory services with contact details. Local guidelines on testing, information on procedures, detailed information on obtaining and treating samples, storage and delivery requirements and reporting times.
- Downloadable request forms and information on necessary accompanying documentation e.g. consent forms and clinical details about the patient.
- Whether the laboratory offers a service for collecting and storing DNA samples for future analysis, with request forms where appropriate.
Patient-focused resources e.g. downloadable information leaflets for patients in the form of Frequently Asked Questions

Some sites gather all this information together and publish it in an accessible form as a downloadable user manual.

Examples of Genetics Laboratory web pages:

West of Scotland Regional Genetics Service
www.gla.ac.uk/departments/medicalgenetics/nhs/molgen.htm
www.gla.ac.uk/departments/medicalgenetics/nhs/molspecs.htm
www.gla.ac.uk/departments/medicalgenetics/nhs/CVS_FAQ.htm

Liverpool Women’s Hospital
www.lwh.org.uk/clinical_services/genetics/molecular/services.html
www.lwh.org.uk/clinical_services/genetics/cytogenetics/form.html

Tayside University Hospitals NHS Trust, Dept of Human Genetics
www.humangenetics.org.uk/graphics/Cytomanual.pdf

11.3 Targeted information for healthcare professionals

This section examines some of the resources available to help health professionals develop and communicate their understanding of some of the key concepts involved with genetics. It identifies some of the online tools available to support clinical practice and some of the features designed to make service delivery more efficient.

It should be acknowledged that most of the resources for health professionals in the sites under review are targeted at GPs. There is very little material directly designed to support other roles such as nurses, midwives and health visitors, or any other health professional group. Where such material does exist, it is often to be found on specialist professional websites representing communities of interest.

It should further be pointed out that some of the features described below are to be found amongst online material relating to the work of specialist cancer services rather than the Regional Genetics services. This is probably a reflection of the fact that the links between cancer and genetics are better understood than in other conditions. Such material is included here because it is indicative of best practice.

The material reviewed falls under the following broad headings:

- Basic genetics literacy
- Professional support
- Explanation and tools for family history taking.
- Material to assist assessing and communicating risk and for deciding on appropriate strategies.
- Referrals guidelines, and processes.
Basic genetics literacy

- A number of sites provide glossaries of basic genetic terms. These provide a simple and quick way of reinforcing basic genetic literacy and building competencies for a range of professional needs. The level of detail varied considerably between different versions. It could be argued that such background content might best be provided from an authoritative central source rather than created locally, a process that duplicates effort and might lead to inaccuracies.

- Explanation of inheritance patterns. There appears to be very little good material offering a basic, accessible explanation of inheritance (this was identified as a priority need by the working groups).

Examples web pages

**West Midlands Regional Genetics Service**  
[www.bwhct.nhs.uk/clinicalgenetics/amainglossarys.htm](http://www.bwhct.nhs.uk/clinicalgenetics/amainglossarys.htm)

**South West Thames Regional Genetics Service**  
[www.genetics-swt.org/adar.htm](http://www.genetics-swt.org/adar.htm)

Professional support

- Some Regional Genetics Service sites offer dedicated areas for GPs. For instance, the site for South West Thames maintains a GP Forum, created by a GP, which brings together a range of useful material including: notes on referrals, a glossary of genetic terms, frequently asked questions, and a series of short factfiles dealing with such issues as autosomal dominant and recessive disorders, multifactorial inheritance, family history of different kinds of cancer and guides to breast examination.

- The British Society for Human Genetics website offers a Leaflets Index allowing members to exchange examples of clinically useful documents, for example, specimens of consent forms for children or adults in use with different medical services. Members are invited to comment on these and to suggest improvements. In general however, there are relatively few opportunities in the sites reviewed for health professionals to feed back to the sites and to share experience and examples of best practice online.

- In addition to the material available from professional bodies like the Royal College of GPs and the BSHG, there are sites that offer a perspective on the developing role of the primary care practitioner. For example, the Oxford Primary Care Genetics Group site, provides a discussion- with helpful links and further reading- of GP skills; the potential value and availability of computer programmes to support clinical practice in genetics; risk counselling; and emerging patient issues. However, such sites are not integrated with the Regional Genetics sites.
Examples of web pages

South West Thames Regional Genetics Service
www.genetics-swt.org/gp.htm

British Society for Human Genetics
www.bshg.org.uk/Leaflets/leafletintro.htm

Royal College of General Practitioners
www.rcgp.org.uk/rcgp/information/catalogues/example/11.asp

Oxford Primary Care Genetics Group
www.dphpc.ox.ac.uk/opcgg/topics.htm

Explanation and tools for Family History taking

The ability to build up a genetic profile of patients and their families through the systematic taking of a family history has been identified as one of the key requirements for the planning and effective delivery of care for genetics-related conditions. Many of the sites under review refer to the importance of compiling a pedigree for patients; a smaller number offer guidance and help with the process. Of the sites that do offer more developed resources, the following features are worthy of note:

- A clear explanation of the purpose and value of a family history in achieving a diagnosis, planning medical interventions and counselling patients and their families.
- A definition of pedigree relationships, and a diagrammatic explanation of the meaning of first and second-degree relatives.
- Examples of questionnaires that will prompt useful information from patients and assist in building up a genetic profile in order to establish the degree of risk for the patient and also other family members.
- Downloadable forms and templates that can be printed out and filled in. These forms are sometimes available via password-protected areas of a website, although sample forms might be published in the public areas of a site for guidance.
- Guidelines for drawing a pedigree such as: standardised symbols, conventions, and layout; the number of generations covered; a unique identifier for cataloguing and recalling the pedigree; standard information on the patient, the history taker, and the date of taking or amending the history; a record of actions taken; the opportunity to record detailed and descriptive information that might be relevant in building up a profile.
- Notes for specialist nurses. These nurses sometimes visit a patient prior to a referral being made. Guidance notes might cover the need to reassure the patient, to gather and check additional information for the pedigree, to advise on whether other family members might need surveillance or referral, and to secure appropriate consent e.g. for the taking and “banking” of DNA samples.
• Materials discussing the development of professional best practice, such as are available from the websites of professional bodies like the Royal College of GPs.

Examples of web pages

**West Midlands Family Cancer Strategy/West Midlands Regional Genetics site**
- [www.bwhct.nhs.uk/wmfacs/about/how/fhf.htm](http://www.bwhct.nhs.uk/wmfacs/about/how/fhf.htm)
- [www.bwhct.nhs.uk/wmfacs/clingen/pedigreerels.htm](http://www.bwhct.nhs.uk/wmfacs/clingen/pedigreerels.htm)
- [www.bwhct.nhs.uk/wmfacs/fhc/fhcnotes.htm](http://www.bwhct.nhs.uk/wmfacs/fhc/fhcnotes.htm)

**South West Thames Regional Genetics Service**
- [www.genetics-swt.org/fhist.htm](http://www.genetics-swt.org/fhist.htm)

**Clinical Genetics Society (via BSHG site)**
- [www.bshg.org.uk/Society/CGS/CGSPedigree.htm](http://www.bshg.org.uk/Society/CGS/CGSPedigree.htm)

**Royal College of General Practitioners**
- [www.rcgp.org.uk/rcgp/clinspec/genetics_group/familyhist.asp](http://www.rcgp.org.uk/rcgp/clinspec/genetics_group/familyhist.asp)

*Risk assessment*

There is a stated need for content that facilitates the understanding, analysis and communication of the concept of risk. A better understanding of risk can enable health professionals to provide more timely and effective care for patients, plan preventive strategies, offer reassurance and counselling and target resources more effectively. These are general observations on available content:

• Although the specific criteria for assessing risk will vary from condition to condition, there is surprisingly little information at a conceptual level to assist in understanding general principles that will allow clinicians to build on the work of family history taking or explain risk to patients and their families.

• There are examples of some broad definitions about the working categories of risk, i.e. low (or “population level” risk), medium and high risk, but relatively little help in calculating or interpreting a statistical assessment of risk.

• The most effective content dealing with risk is found in relation to cancer services, perhaps because the relationship between familial risk and clinical condition is better understood in this area. Here, there are many examples of risk profiles developed for specific conditions against which patients can be assessed. The clinician can then link these to guidelines for referral in order to plan appropriate measures and to offer counselling to assist the patient in making informed decisions about their lifestyle or the clinical steps available to them.
Guidelines on referral

This is another key area where online resources are useful and important. There is a need for accessible information on national guidelines relating to the management of different genetic conditions, and for accurate and up-to-date information on local policy and practice. As before, the most detailed and rigorous online guides for referrals occur in the area of cancer treatment rather than directly through the Regional Genetics Service. Two sites were examined in detail, the Cancer Information and Support Service within Central Manchester and Manchester Children’s University Hospitals, and the West Midlands Family Cancer Strategy, linked to the West Midlands Regional Genetics Service website. Features of best practice in the area of referrals include:

- **Manchester**
  - Integrating local practice within the national framework, with links to guidelines published online by the Department of Health.
  - A downloadable version of these guidelines presented as a wall chart for ease-of-reference
  - Notes for GPs setting out the process of referral and stating the responsibilities of the GP and of the service provider (in this case, the Hospital Trust) to act within a target timeframe.
  - Detailed referral criteria for a range of types of cancer based on the analysis of symptoms and linked to the age and the lifestyle of the patient
  - A downloadable form for urgent referrals.

- **West Midlands**
  - This site has developed a flow chart or “decision tree” designed to give an overview of the referrals process and to offer a step-by-step guide through the available courses of action.
  - It helps distinguish between patients in need of immediate referral and those needing surveillance, and identifies actions to plan an appropriate strategy for dealing with each group, linked to local service provision.
  - For high-risk patients who are judged to be in need of referral, it takes the carer step-by-step through the process of arranging appointments in a framework of ongoing counselling and support. It provides background information on genetic testing for patients who have been referred to the next level of care, and discusses some of the implications of testing.
  - For patients judged to be in need of surveillance, it identifies appropriate measures, such as reassurance for patients, close examination of the family tree, information and advice on lifestyle and self-examination techniques, and if necessary, the banking of DNA samples. The flow chart also covers a course of action for dealing with patients who are not
judged to be in need either for referral or surveillance and provides useful information sheets for them.

**Examples**

**Central Manchester and Manchester Children’s University Hospital**
- www.cmht.nwest.nhs.uk/cancerinfo/referralguidelines.asp
- www.cmht.nwest.nhs.uk/cancerinfo/hsccologuide.asp
- www.cmht.nwest.nhs.uk/cancerinfo/hscgpnnotes.asp
- www.cmht.nwest.nhs.uk/cancerinfo/hsccoloform.asp
- www.doh.gov.uk/cancer/referral.htm
- www.doh.gov.uk/pub/docs/doh/wallchart.pdf

**West Midlands family Cancer Strategy**
- www.bwhct.nhs.uk/wmfacs/about/how/algorithm1.htm
- www.bwhct.nhs.uk/wmfacs/guidelines/referral.htm

**West of Scotland Cancer Genetics Service**
- www.gla.ac.uk/departments/medicalgenetics/cancer/brcaguidelines.htm

**Liverpool Women’s Hospital Cancer Genetics Service**
- www.lwh.org.uk/clinical_services/genetics/clinical/cancer_service.html

**11.4 Resources for patients and their families**

The open nature of the online medium makes it a primary source of information for people seeking information about disease, therapies and lifestyle issues concerning themselves and their families. An observation that was frequently made in the workshops and interviews was that, in an area of emerging knowledge like genetics, the traditional relationship between the professional and the patient, in which the former is the fount of knowledge, and the latter the passive recipient of care has been changed, and sometimes reversed. Due to the availability of vast amounts of information via the web, the patient is often as well informed about their condition as the health professional. This presents the health professional with the opportunity to involve the patient more proactively in the management of their care and more involvement in important choices. There are caveats of course. Many patients find it more reassuring to entrust their carer with telling them what they need to know without encumbering themselves with information that might be confusing or alarming. Furthermore, the sheer volume of available websites means that quality control is a real issue.

Although the sites under review are largely aimed at healthcare professionals, many also publish content that is designed to be used by the patient and their families, either directly, or perhaps in consultation with their GP or practice nurse. Such material, by adopting an appropriate clarity, language and tone, has an important secondary potential, which is to reinforce the understanding and competencies of health professionals themselves.
Examples of content presented to patients and families include:

- A fact sheet on genetics and inheritance. This is published by the Institute of Child Health and is accessed via the Great Ormond Street Hospital site. The high profile nature of the hospital's work means that there is likely to be substantial public interest, and this is acknowledged in the website, which has sections both for health professionals and for patients and their families. The fact sheet covers such basic questions as:
  - What are genes?
  - How can genetic disorders be inherited?
  - Is it always possible to make a genetic diagnosis?
  - How can genetic abnormalities be detected?

It is available under “Information for families”. Given that the working group identified the need for background information on inheritance, this kind of material might also be a useful introductory resource for health professionals in different functions and different levels of specialty.

- Family support materials on Regional Genetics sites. An example from the Liverpool Women’s Hospital site includes information designed to guide people through the referrals process. Often the patient is unsure why they are being referred. This is even more the case when tests or pedigree taking have revealed the need to refer close family members who may not, themselves, be aware of an existing or potential condition. The tone and language need to be simple, clear and free of technical terms or jargon. There is an explanation of the support available through genetic counselling and a description of the different steps in the process beyond referral. The approach puts the patient at the centre of the process with the intention of helping them make informed decisions. It is explained to the patient that they will be contacted by phone or home visit by a genetic associate and they are asked to gather family details ahead of this in order to make the process of family history taking more efficient. They are also offered the option of an interpreter or signer. Finally there is a list of links to supporting organisations (links are discussed more fully below).

- Dedicated areas of content. Some sites have clearly signposted areas of content aimed at patients and their families as an easy-to-find summary of information they might need to know relating to their appointment at a genetics clinic. Often this takes the form of frequently asked questions covering such issues as why they might have been referred, how a diagnosis is arrived at and types of test available. This format has the further useful supportive function of providing the patient with an overview of the kind of service they should expect and a list of questions they should ask their clinician to get the maximum informational value from the consultation. Some sites also take the opportunity to provide a “checklist” of important information or documentation it would be helpful for the patient to bring with them in order, for example, to build up a family history, if this has not been done prior to the appointment. Even sites whose overall purpose is to serve a professional
community e.g. the BSHG website, recognize the value in also publishing accessible information for patients.

- **Downloadable care cards.** One example encountered was a printable care card for Myotonic Dystrophy published by the Scottish Muscle Network. This is designed to be printed out, folded and carried by the patient in a plastic wallet. It contains a summary of the ways the condition affects the patient’s health and assists them in monitoring their symptoms. It also contains a diary of clinic visits; tests carried out and contact details. The card also has an important role in alerting medical services to the nature of the problem should a patient be suddenly incapacitated.

- **Use of patients to help build communication and best practice.** The website for the Scottish Muscle Network, linked to the West of Scotland Regional Genetics Service website had an imaginative example of using patients and carers as a means of building informational networks amongst health professionals in order to extend their knowledge and improve the management of neuromuscular disorders. Patients who use the site are invited to send details of their care teams to the website so that the health professionals can be invited to educational and management meetings. The talks given by patients and carers at these meetings also provide the opportunity for professionals to hear different perspectives on the effectiveness of the service provided.

- **Opportunities to feedback.** It is, perhaps, surprising how few sites offer the opportunity for the patient to tell the service what they think of the information offered and what they need. Clearly, any such opportunity implies a commitment both to monitor the feedback in a timely manner and to take account of it in improving the site. Both of these have resource implications.

- **Invitations to take part in research.** In a few cases, websites invite patients and their families to take part in programmes of research into genetic disorders. Given that many patients are highly knowledgeable and motivated, it is surprising that more websites do not tap into this potential resource.
### Online material on specific disorders

This section deals in more detail with online content relating to specific diseases. It examines the contextual and support materials relating to some of the more common diseases where there may be a genetic factor at work. As noted before, the best and largest amount of material is in the area of cancer care, and these resources are examined in some detail. It should be said that in many cases these materials are created and published by other departments and services, and that their underlying editorial principle and navigational structure is defined by the condition – “cancer” - not the broader subject of “genetics”. They are included here because they provide some excellent examples of material that helps to build up an understanding of the impact of genetics on the onset and development of the condition and on the various tests and therapies used to manage it. It is not always the case, however, that this material is adequately linked to material created by the Regional Genetics Services, even within the same overall website. An appropriate overview of relevant content would overcome this compartmentalisation and provide a more integrated service to the user.

A further point of note is that very little material was found describing the impact of genetics on other common conditions, such as heart disease. Developing knowledge of these medical and causal links will enable the creation of better support materials, but at present there appears to be a significant gap in provision.

**Content about cancer and genetics**

Among the sites that do publish a significant amount of material on cancer and genetics, the following three were looked at in detail:
• West of Scotland Cancer Genetics Service. This site is integrated with the work of the West of Scotland Regional Genetics Service and is run through the Institute of Medical Genetics at the Yorkhill Hospital, Glasgow. It offers the following features:
  o A brief but useful introduction to cancer genetics explaining the difference between mutation and genetic predisposition
  o Background information on breast, ovarian and colorectal cancer
  o A description of the work of the Cancer Family History Clinic, in the form of questions and answers, and a diagrammatic explanation of a family tree *Who is who in your family.*
  o Brief referral guidelines for the three types of cancer described above.

• West Midlands Family Cancer Strategy. This is a very substantial site that complements that of the West Midlands Regional Genetics Service. It is rather dense and complex to navigate, but offers many useful features, some of which have already been covered in some of the other sections. These include:
  o Information on the different types of cancer, its incidence and the impact of inheritance
  o Advice and factsheets on self-examination and healthy living
  o Protocols for surveillance and referral
  o Family history taking, with supporting documentation
  o The provision and management of different resources and services throughout the region.

• Central Manchester and Manchester Children’s University Hospital Cancer Information and Support Service. This site is linked to the Cancer Genetics Service (family history) within the regional genetics service based at St Mary’s Hospital, Manchester. Some features of the site e.g. referrals guidelines have already been considered above. Overall the site is noteworthy for the thoroughness of its content and its relevance to health professional and patient alike. It includes:
  o An explanation of cancer, its incidence and the difference between benign and malignant tumours
  o Advice on healthy living, with active links to external sites offering more help; guidance on self-monitoring for symptoms
  o Information on many different types of cancer linked to the services and procedures operating locally.
  o Referral guidelines
  o An overview of the different approaches to treatments and their outcomes
  o Advice for patients and their families on living with cancer where effective treatment might not be available, such as palliative care, support groups and spiritual and emotional care.
  o Question-and-Answer resources that take a patient step-by-step through the processes and issues involved in their treatment
  o Extensive links to other sites offering further support.
  o An opportunity for users to feedback comments and information about their needs to assist with the development of the site.
Examples of Web Pages:

**West of Scotland Cancer Genetics Service**
- [www.gla.ac.uk/departments/medicalgenetics/cancer/index.htm](http://www.gla.ac.uk/departments/medicalgenetics/cancer/index.htm)
- [www.gla.ac.uk/departments/medicalgenetics/cancer/breastcancer.htm](http://www.gla.ac.uk/departments/medicalgenetics/cancer/breastcancer.htm)
- [www.gla.ac.uk/departments/medicalgenetics/cancer/whoiswho.htm](http://www.gla.ac.uk/departments/medicalgenetics/cancer/whoiswho.htm)

**West Midlands Family Cancer Strategy**
- [www.bwhct.nhs.uk/wmfacs/inherit/breastov/breastovintro.htm](http://www.bwhct.nhs.uk/wmfacs/inherit/breastov/breastovintro.htm)
- [www.bwhct.nhs.uk/wmfacs/surveillance/lifestyle.htm](http://www.bwhct.nhs.uk/wmfacs/surveillance/lifestyle.htm)
- [www.bwhct.nhs.uk/wmfacs/inherit/quickguide.htm](http://www.bwhct.nhs.uk/wmfacs/inherit/quickguide.htm)

**Central Manchester and Manchester Children’s University Hospitals Cancer Genetics Service**
- [www.cmht.nwest.nhs.uk/cancerinfo/aboutcancer.asp](http://www.cmht.nwest.nhs.uk/cancerinfo/aboutcancer.asp)
- [www.cmht.nwest.nhs.uk/cancerinfo/typesofcancer.asp](http://www.cmht.nwest.nhs.uk/cancerinfo/typesofcancer.asp)
- [www.cmht.nwest.nhs.uk/cancerinfo/genetics.asp](http://www.cmht.nwest.nhs.uk/cancerinfo/genetics.asp)
- [www.cmht.nwest.nhs.uk/cancerinfo/treatments.asp](http://www.cmht.nwest.nhs.uk/cancerinfo/treatments.asp)
- [www.cmht.nwest.nhs.uk/cancerinfo/livingwithcancer.asp](http://www.cmht.nwest.nhs.uk/cancerinfo/livingwithcancer.asp)

*Information on other disorders*

As mentioned above, there is little evidence of online information about the relationship between genetics and many common conditions such as heart disease, diabetes and psychiatric illness. Examples of material on conditions other than cancer include:

- The genetics of cystic fibrosis. The South West Thames Regional Genetics Service has brief background information on this condition and also looks into gene testing and the potential for screening.
- The Scottish Muscle Network, linked to the West of Scotland Regional Genetics Service has a considerable amount of material on neuromuscular disorders designed to improve communication among professionals and to support patients.
- The West of Scotland service also provides some information on Downs Syndrome and Spina Bifida in the context of explaining pre-natal screening.
### Examples of Web Pages:

**South West Thames Regional Genetics Service**  
www.genetics-swt.org/cf.htm

**Scottish Muscle Network**  
www.gla.ac.uk/muscle

**West of Scotland Regional Genetics Service**  
www.gla.ac.uk/departments/medicalgenetics/nhs/biogenpopscreen.htm

#### 11.6 Dealing with genetics in society

Some of the workshops identified a need for material that sets genetics in its wider social context and allows health professionals and patients to understand and navigate some of the issues raised when medical decisions have to be weighed against complex legal, ethical, social and cultural considerations. A number of these issues have already been clearly identified, such as those concerned with genetic testing and screening. Other issues are still emerging as our understanding of genetics develops, such as the relationship between inheritance and lifestyle, and the predisposition of some communities and races to suffer from certain kinds of genetics-related condition. Some of these issues are addressed by organisations like the Genetics Interest Group but overall there is relatively little information and guidance directly available through the Regional Genetics Services at present. Some of the examples of such material that were encountered include:

- A brief explanation of some of the implications of genetic testing, for instance, for other members of the family, for insurance, employment and education, and its psychological impact (West Midlands Family Cancer Strategy)
- A paternity testing service offered through the University of Glasgow. This was an unusual example, but clearly the genetic data available from a DNA test of this kind might have very direct social and emotional implications and it seems important that these should be addressed within the context of the service delivery.
- Statements on matters of public policy. The British Society for Human Genetics, for example, publishes documents such as a Joint Statement on Genetics Research and Insurance representing the position of various interested bodies. Also many sites link to external websites of bodies like the Human Genetics Commission or the Department of Health where further pronouncements on public policy can be found.
- There were a very few examples of dealing with genetic considerations specific to particular racial groups, such as the higher susceptibility of Ashkenazi Jews to breast cancer and ovarian cancer. However, there was little evidence of material that was culturally specific and which addressed, for instance, the health and lifestyle issues and needs of particular groups in a multicultural context.
society. None of the regional genetics sites examined had material in any language other than English.

- There were a few examples of printable consent forms for some laboratory tests, but little material that set out the principles and practices governing consent and confidentiality in the wider context that could be used, for instance, to inform and reassure patients and their families.

- Some sites provide a “News service” of press releases. This has the potential to keep both health professionals and public alike abreast of developments in science, medicine and public policy. The website for Great Ormond Street, for example, has an extensive Press Release Archive and, given the high profile and news value of such an institution, there is a potential benefit in helping to build genetic literacy amongst journalists and public commentators as well. However, the resource implications of such a service should not be underestimated. One regional site which claims to offer a “News and Events in Genetics” facility appears to have last updated its content in May 2001. Overtly dated content undermines the credibility of a site.

### Examples of Web Pages:

**West Midlands Family Cancer Strategy**  
www.bwhct.nhs.uk/wmfacs/genetic/implications.htm  
www.bwhct.nhs.uk/wmfacs/inherit/breastov/ashkenazi.htm

**West of Scotland Regional Genetics Service**  
www.gla.ac.uk/paternity/

**British Society for Human Genetics**  
www.bshg.org.uk/Official%20Docs/Insurance/jointstatement.htm

**Great Ormond Street Hospital/Institute of Child Health**  
www.gosh.nhs.uk/pressoffice/pressarchive/genes_03_04_02.html

**South West Thames Regional Genetics Service**  
www.genetics-swt.org/news.htm

**Sheffield School of Health and Related Research**  
www.shef.ac.uk/~scharr/publich/research/genetics/workplac1.html

### 11.7 Links

All of the sites reviewed offered links to further resources. Some of these were cursory, others very extensive. The kind of links on offer fall under a number of broad headings:

- Sites related to primary care. These include the websites of professional bodies like the BSHG, the Clinical Genetics Society and the Royal College of General Practitioners. There are also links to specialist registers and databases which
assist in the work of diagnosis like FIDD (The Frequency of Inherited Disorders database) or the Human Gene Mutation Database, maintained by the Institute of Medical Genetics at Cardiff, and OMIM (Online Mendelian Inheritance in Man).

- Sites that deal with matters of Public Health, Policy and Governance and the bigger scientific picture such as the Department of Health, the Human Genetics Commission, and the Human Genome Project sites.
- Sites that offer general educational materials and information on, for instance, computer software programmes.
- Medical journals and resources relating to specialist research.
- Sites dealing with the social, ethical and legal implications of genetics, and the public understanding of science, such as the Genetics Interest Group website.
- A very large number of sites that offer information and support to patients suffering from specific conditions such as Contact-a-family and Cancer Back-up.

It is impossible to quantify the online resources available or to examine the linked sites in detail. It is useful, perhaps, to try to establish a few broad principles about links to make sure that they’re effective, useful and not downright harmful:

- The content needs to be validated for accuracy, authority and appropriateness.
- Links need to be regularly reviewed, quality-controlled and maintained up-to-date.
- Ideally links should be content-specific, i.e. they will bring the user immediately to the relevant material rather than to a home page where another laborious search might be required.
- Broad headings help the user find the sites most relevant to them. Annotated commentaries on the links are also useful, for instance, a brief summary of the kind of material contained on the site (and whether it’s any good).
- Feedback facility that allows the users to “add value” by submitting suggestions for links, or adding their opinions of the links on offer. This is the way that commercial sites like Amazon rapidly grow traffic and build repeat usage.
- Overall there is the need for the role of a “trusted guide”, a source of reliable, timely and useful information to make sense of the bewildering plethora of content available online.
Examples of Web Pages:

**British Society for Human Genetics**  
[www.bshg.org.uk/links.htm](http://www.bshg.org.uk/links.htm)

**The Oxford Primary Care Genetics Group**  
[www.dphpc.ox.ac.uk/opcgg/links.htm](http://www.dphpc.ox.ac.uk/opcgg/links.htm)

**The Royal College of General Practitioners**  
[www.rcgp.org.uk/rcgp/clinspec/genetics_group/links.asp](http://www.rcgp.org.uk/rcgp/clinspec/genetics_group/links.asp)

**Medical Genetics Service for Wales**  
[www.uwcm.ac.uk/study/medicine/medical_genetics/internal/genet_links.html](http://www.uwcm.ac.uk/study/medicine/medical_genetics/internal/genet_links.html)  
[http://archive.uwcm.ac.uk/uwcm/mg/fidd/](http://archive.uwcm.ac.uk/uwcm/mg/fidd/)

**West of Scotland Regional Genetics Service**  
[www.gla.ac.uk/departments/medicalgenetics/cancer/links.htm](http://www.gla.ac.uk/departments/medicalgenetics/cancer/links.htm)

**Genetics Interest Group**  
[www.gig.org.uk/links.htm](http://www.gig.org.uk/links.htm)

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**Educational resources**

Among the key roles for a clinical geneticist, in addition to providing a service to patients, is to be a conduit for expertise and information about genetics for health professionals. Increasingly there is an opportunity and an urgent need for clinical geneticists to fulfil the role of active educator in the meaning and importance of genetics in healthcare that will support continuing professional development, inculcate the right attitudes and behaviours and help develop and disseminate best practice among a wide range of health professionals. Many clinical geneticists are already involved in talks, lectures and seminars for health professionals. This section looks at some of the online resources concerned with training and professional development available from the sites under review.

- **Websites of professional bodies: defining the role.** As one would expect, there is a substantial set of material published on the websites run by organisations supporting the work of groups of professionals. The British Society for Human Genetics and the Clinical Genetics Society publish a number of documents that help define and develop the emerging role of the clinical geneticist and identify key areas where consistency and best practice need to be established, such as: an approach that extends support and care to the extended families of patients with genetic disorders; the need to prioritise and manage effectively the demands for educational resources; the maintenance of genetic registers; and the need to preserve the opportunity to engage actively in research. The BSHG also conducts and publishes surveys of
the ways in which its members are currently involved in communicating genetics to the public.

- **Establishing core competencies and setting professional standards.** The BSHG has published a document that seeks to establish a core curriculum of genetics for the teaching of undergraduate medical students and influence the provision of training and teaching at medical schools. This document identifies the essential knowledge and skills required for an understanding of basic genetics, and states the key skills and competencies, attitudes and behaviour involved in best clinical practice. In each case it helps to define the sets of learning outcomes to be achieved in training.

- The BSHG also publishes an **appraisal template** for competency-based assessment of trainees in Clinical Genetics.

- **Information on courses and training.** The sites of professional bodies give some background information on training. For instance, the Association of Genetic Nurses and Counsellors has a short paper –“**Training to be a Genetic Counsellor**”- and a directory of courses in clinical/medical genetics and genetic counselling throughout the UK. The BSHG publishes details of courses, conferences and other events for Professionals. These include workshops, study days and short course. However, the information is not very substantial.

- There is a dearth of information about courses and training opportunities directly available from the Regional Clinical Genetics sites, and this appears to be an important omission. Some sites do feature information on locally available courses. The Department of Medical Genetics at Wales, for example, publishes details of a number of courses, ranging from a full Masters Degree in Genetic Counselling to short courses in Public Health Medicine and Medical Genetics. In each case there is a short definition of course content, entry requirements and contact details. Some other regional sites associated with universities offer links to academic resources within the university, although these are sometimes password-protected. Others publish undergraduate lecture notes and slides, and reading and reference lists.

- **Direct provision of teaching and training materials.** Although much of the online content reviewed in this report has an educational purpose, there is little evidence of the systematic use of materials as part of a programme of education and training that can be delivered directly via a computer or as a blend of online, face to face and practically-based learning. The Royal College of General Practitioners has invited expressions of interest in the provision of distance learning courses in medical education, although it is not clear, as yet, whether this will result in accredited online course content for clinical genetics. Elsewhere, in the course of this report one other mention was made of distance learning materials in a bid for funding to develop computer based learning modules for the Regional Genetics site in Edinburgh. Again, there is no evidence yet that this has been developed further.

- The potential for **structured online learning** using short modules, immersive, case-based scenarios and an area of a website dedicated to ongoing

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1 haitez, N et al. Teaching Medical Genetics to Undergraduate Medical Students, 2002. Document agreed by the British Society for Human Genetics and Joint Committee on Medical Genetics available at [www.bshg.org.uk/Official%20Docs/UNDERG~1.doc]
professional development (e.g. a “Learning Zone”) has yet to be exploited by any of the sites reviewed.

Examples of Web Pages:

**British Society for Human Genetics**
www.bshg.org.uk/Official%20Docs/clingenrole.htm
www.bshg.org.uk/trainees/AppraisalAndAssessment/competencies.htm
www.bshg.org.uk/Events/events.htm

**Association of Genetic Nurses and Counsellors**
www.agnc.co.uk/training.htm
www.agnc.co.uk/course.htm

**Medical Genetics Service for Wales**
www.uwcm.ac.uk/study/medicine/medical_genetics/study/courses/

### 12 Summary and conclusions

This section will offer a brief overview of the results of the website report. It will summarize the key features identified that might be used to plan the provision of future sites, and will explore the relationship between a centrally created site and local sites.

#### 12.1 Summary of main findings

The main findings were as follows:

- Provision of websites is patchy, with many regional omissions.
- There is no common standard for what constitutes “fit for purpose”. Design, navigation and content differ widely, as a reflection of local organisational structures, practices and resources. In some cases, where there is good content relating to clinical genetics, it can be hard to find through the lack of effective integration within the host site.
- There is often a lack of clarity about the objectives of the site and on the target audiences. This often concerns the balance between content aimed at the health professional and content that is designed to be used by the public. However, there are also significant gaps in provision to some professional groups e.g. Nurses.
- Although many sites offer basic descriptions of local services, there is a clear need for more contextual, educational material to build the understanding of basic genetics and of the impact of genetics on individuals, their families and society.
• Maintenance is an issue. There were examples of inaccurate and out-of-date information being published, for instance, where guidelines on local practice had been changed without the site being amended accordingly.
• Many examples of good content do exist. Some areas of practice, e.g. cancer genetics, are well served: other areas of disease are less well provided for. There are a number of examples of excellent downloadable, reusable support materials; tools and guidelines; online documents to facilitate the delivery of services; developing communities of interest to share best practice; clear, useful background material. However, there is no coordination of content between sites, which sometimes results in duplication.
• Many sites make effective use of the web to direct users to further sources of information. However, there is a need for an authoritative, “trusted guide” through the sheer volume of content available.

12.2 Summary of key features

The key features were as follows:

• Information on delivery of local services: location and times of clinics and services offered; contact details; guides to local processes and protocols; online “user manuals” for specialist services.
• Local and national referral guidelines
• Service integrated with overall host website, with opportunity to search and navigate with “Genetics” or “Clinical Genetics” as the guiding principle.
• Downloadable proforma documents that expedite the delivery of a service e.g. online referral request forms or requests for laboratory testing.
• A checklist of questions that might help to identify possible genetic factors at work, help with prevention, diagnosis and treatment. Decision support tools e.g. for making a risk assessment or deciding whether to make a referral.
• Basic information on genetics, such as explanation of Inheritance and contextual material that builds an understanding of how genetics can impact on health in many different ways.
• Glossaries of genetics terms.
• Tools to support good clinical practice e.g. taking a Family History
• Easy-to-use informational resources for patients and professionals e.g. downloadable factsheets, FAQs and online fact files on health and lifestyle issues; guides to the referral and consultation process; explanations of the implications and outcomes of genetics testing; explanation of consent; counselling procedures; resources that help the patient with the management of their condition e.g. downloadable care cards. This content can be aimed directly at the patients and their families, but can also be “mediated” through the health professional.
• Targeted information for specific user groups e.g. nurses and Genetics Counsellors.
• Information on specific sets of diseases where there is a genetic factor at work.
• Downloadable marketing materials that distribute information and offer a quick guide about the services on offer e.g. posters and leaflets (which might be seen in a surgery or sit on a GPs desk).
• Content that helps the professional to engage with the social, legal and ethical issues arising, and the patient to make informed choices.
• Opportunities for professionals to exchange ideas and share best practice, and for patients and their families to feedback their own knowledge and experience.
• Validated, annotated links to further online and offline resources. Timely and accurate information about developments in genetics as they impact upon service delivery. Specialised information e.g. on areas of particular expertise or research interest, that might support a decision to refer.
• Information on courses and knowledge-building activities for different groups of professionals.
• A multi-disciplinary approach that addresses areas of collective need and builds on collective strengths among health professionals.

12.3 Centralised provision versus local websites

The National electronic Library for Health has announced plans to develop a “branch library” on Genetics. The development of GenePool ([www.nelh.nhs.uk/GenePool](http://www.nelh.nhs.uk/GenePool)) is being led by the British Society for Human Genetics ([www.bshg.org.uk/nelh.htm](http://www.bshg.org.uk/nelh.htm)). It will deliver “an online genetics resource…which is aimed at non-geneticist clinicians, patients and their families”. The site will offer:

- **Briefings**: peer-reviewed articles on genetic diseases and basic concepts
- **Webliography**: annotated database of information and further reading
- **News**: updates on developments in genetics research, treatment and policy
- **Services**: referral guidelines, decision support tools and details of genetics services

Other sections will include discussion groups, software tools (including risk assessment tools), support group information and details of editorial and administrative staff.

Such a service might fill many of the gaps identified in this report, but there are also issues:

- It has the potential value of a single, one-stop-shop destination, a portal for a very rich set of resources for many different audiences. On the other hand such a proposition will require substantial marketing to drive widespread use and might begin by having to compete with well-established local service sites (for instance the local hospital) or with well-known sites serving special interest groups (e.g. GIG). Branding will be a key consideration: the National electronic Library for Health is unlikely to become a compelling proposition for the public.
- The creation of a suite of resources is a substantial editorial and production undertaking which will take time (and budget) to deliver and there is a risk of further duplication of content that already exists locally.
- Such a service needs to be responsive to local needs and practice.

On the other hand, as this report has explored, local provision can be inadequate. There are some excellent local sites, often down to the enthusiasm, expertise and
commitment of individuals or groups within an organisation. These local sites are often themselves constrained by resource and maintenance issues. For example some sites find it almost impossible to update or amend their content: control of their content is managed through the overall hospital site, which can be unresponsive and slow to meet the needs of maintenance, resulting in inaccurate information continuing to be published. Other sites are vulnerable to shifts in local priorities that might see resources being reallocated elsewhere.

In practice, the way forward is probably a “mixed economy” where local content is supported by centrally produced resources. The local services would build on local practice, be responsive to local needs, knowledge and initiative and achieve local ownership from patients and professionals, without being over-regulated or over-standardised. A central service could provide more in-depth resources, drive up quality standards, and achieve wider marketing and distribution. Such a partnership between, for example, GenePool and an emerging network of local sites would see a central service responsible for:

- Planning, coordinating, commissioning and quality controlling background content
- Identifying and addressing gaps in provision e.g. by subject area or user group
- Holding funds for specific projects which could be “bid for” by local services
- Identifying and circulating best practice
- Developing more sophisticated tool kits and software for general use
- Taking the role of “Trusted Guide” to external online sites: validating, annotating and recommending sites and researching specific content links. The same applies to offline resources.
- Advising on what is “fit for purpose” in particular cases and designing templates or a “kit of parts” that can be used by local sites to plan and publish their content according to their needs and the level of resources available.
- Acting as a portal to the whole network of local sites, an additional entry point to the local web address.
- Providing an overview of developments, news and policy issues, and offering content that addresses the broader social, ethical and legal issues.
- Sponsoring multi-disciplinary projects
- Establishing and disseminating technical standards and providing a resource of technical support. A more ambitious model might be that GenePool should be the hosting service for the network of local content, although this might over-extend the internal links to local hospital sites and make navigation more difficult.
- Offering deeper layers of more specialised content that could be available as a subscription service.
- Taking a more proactive role in developing educational materials, for instance, maintaining a database of relevant courses across the country or, more ambitiously, developing online modules that can build a user’s understanding of particular subject areas in their own time and at their own pace.
## Appendix

### List of websites

Those marked * are regional sites where there is a significant body of material that has been examined in detail

#### Regional Genetics Services

**Aberdeen** (North Scotland Regional Genetics Service) *
- [www.abdn.ac.uk/medgen/](http://www.abdn.ac.uk/medgen/)

**Belfast** (part of Queen's University site, Dept of Medical Genetics)
- [www.qub.ac.uk/cm/mg/](http://www.qub.ac.uk/cm/mg/)

**Birmingham** *(West Midlands Regional Genetics Service, West Midlands Family Cancer Strategy, West Midlands Regional Genetics Laboratory)*
- [www.bwhct.nhs.uk/clinicalgenetics/index.htm](http://www.bwhct.nhs.uk/clinicalgenetics/index.htm)
- [www.bwhct.nhs.uk/wmfacs/index.htm](http://www.bwhct.nhs.uk/wmfacs/index.htm)
- [www.bwhct.nhs.uk/regionalgenetics/index.htm](http://www.bwhct.nhs.uk/regionalgenetics/index.htm)

**Bristol** (Clinical Genetics Department)
- No website found

**Cambridge** (East Anglian Regional Genetics Service Addenbrooke's Hospital, CIMR)
- [www.addenbrookes.org.uk/serv/clin/women/medgenet/clingenet1.html](http://www.addenbrookes.org.uk/serv/clin/women/medgenet/clingenet1.html)
- [www.addenbrookes.org.uk/serv/clin/women/medgenet1.html](http://www.addenbrookes.org.uk/serv/clin/women/medgenet1.html)
- [www.cimr.cam.ac.uk/medgen/](http://www.cimr.cam.ac.uk/medgen/)

**Cardiff** (Institute of Medical Genetics) *
- [www.uwcm.ac.uk/uwcm/mg/](http://www.uwcm.ac.uk/uwcm/mg/)

**Chester**
- No website found

**Dublin** (National Centre for Medical Genetics)

**Dundee** (Tayside University Hospitals NHS Trust Ninewells Hospital and Medical School) *
- [www.humangenetics.org.uk/clingen.htm](http://www.humangenetics.org.uk/clingen.htm)

**Edinburgh** (South East Scotland Clinical Genetics Service)
- [www.genetics.med.ed.ac.uk](http://www.genetics.med.ed.ac.uk) (no content currently active)
Exeter (Devon and Cornwall Regional Genetics Service)  
No website found

Glasgow (West of Scotland Regional Genetics Service)  *  
www.gla.ac.uk/departments/medicalgenetics/nhs/

Inverness (Regional Genetics Service)  
No website found

Leeds (Yorkshire Regional Genetics Service)  *  
www.leedsteachinghospitals.com/services/lab_radio_pharm/genetics.html

Leicester (Leicestershire Clinical Genetics Service)  
www.le.ac.uk/iog/members/clinicalgenetics.html  
www.le.ac.uk/ge/rt7

Liverpool (Liverpool Women’s Hospital)  *  
www.lwh.org.uk/clinical_services/genetics/clinical/index.html

Liverpool (Mersey Regional Clinical Genetics Service, Alder Hey Children’s Hospital)  
No website found

London  
Great Ormond Street Hospital/Institute for Child Health/North East Thames Regional Genetics Service  *  
www.ich.ucl.ac.uk/clinserv/clingen.htm  
www.ich.ucl.ac.uk/units/cmgu/bindexJAN.htm

North Thames Regional Genetics Service  
No website found

North West Thames Regional Genetics Service  
No website found

South East Thames Regional Genetics Service  
www.guysandstthomas.nhs.uk/page627.htm

Kings College Division of Medical and Molecular Genetics, Clinical Genetics Centre  
www.kcl.ac.uk/ip/ebitimiigbaseimokumo/clinicalgenetics.html

South West Thames Regional Genetics Service  *  
www.genetics-swt.org/
Manchester
North Western Regional Genetics Service
No website found

Central Manchester and Manchester Children’s University Hospital Cancer Information and Support Centre *
www.cmht.nwest.nhs.uk/cancerinfo/

Newcastle (Northern Region Genetics Service)
No website found

Institute of Human Genetics, University of Newcastle
www.ncl.ac.uk/ihg/

Nottingham (Clinical Genetics Service)
www.ncht.org.uk/servicelist.html#genetics

The Division of Genetics, Nottingham University
www.nottingham.ac.uk/genetics/division/

Oxford (Regional Genetics Service)
No website found

Sheffield (North Trent Genetics)
www.sheffield.nhs.uk/hospitals/childrensgenetics.htm

Southampton (Wessex Clinical Genetics Service) *
www.som.soton.ac.uk/research/geneticsdiv/wessexregionalgeneticsservice.htm
Professional bodies

British Society for Human Genetics  
www.bshg.org.uk/

Royal College of General Practitioners  
www.rcgp.org.uk/

Clinical Genetics Society  
www.clingensoc.org/

Oxford Primary Care Genetics Group  
www.dphpc.ox.ac.uk/opcgg/

Association of Genetic Nurses and Counsellors  
www.agnc.co.uk/

Clinical Molecular Genetics Society  
www.cmgs.org/

NB These sites represent those that were publicly accessible following thorough searching of the web during late Autumn 2002. They may not be comprehensive and specifically do not include sites on NHS Net, some of which were notified to us subsequently. We acknowledge, therefore, that the list will not be comprehensive, but it does represent what a competent health professional might easily be able to access on a national basis.