Draft

A Strategy for Genetics in Wales

1. Background

Rapid recent advances in the Human Genome Project led to remarkable developments, which are taking place in the field of human genetics, and in particular their potential impact on the future provision of the health services. The impact on health and health care of the sequencing of the human genome has been well rehearsed[1]. Our understanding and categorisation of disease will be enhanced. Drug therapy may become safer and more effective as treatments are tailored to take account of individual responses to drugs. Genetic tests will increasingly be used to predict the risk of disease and initiate preventive action. Drug development will become faster and more efficient. Medicine had traditionally relied upon the careful observation of patients and the recognition of phenotypes, physically observable human traits or manifestations of disease, as the method by which diagnoses were made. With time, and greater scientific understanding, diagnosis will become more sophisticated, moving from a descriptive to a pathological and ultimately aetiological categorisation of disease. Classification of diseases will develop in the future based on genotype instead of one that depends on clinical and pathological characteristics.

Awareness that demand for services may exceed resources and capacity was behind the establishment by the Department of Health of the Genetics Commissioning Advisory Group. The group is developing mechanisms for evaluating and setting priorities for genetic technologies within the NHS.

As genetic testing becomes more widespread, molecular genetic and cytogenetic laboratories could provide a service similar to chemical pathology, accessed directly by clinicians[2]. All specialists will have to become familiar with the genetic factors underlying the diseases they see. General practitioners will need to acquire specific skills, including assessing genetic risk. Geneticists will continue to have a unique role because of their expertise in counselling and long term management of patients with genetic disorders.

Policy development must inevitably take into account issues wider than health and health services because the consequences of genetics for society reach beyond the boundaries of health services. The UK Government has also reviewed the advisory and regulatory framework for biotechnology. The Human Genetics Commission was therefore established in 1999 to look at developments in human genetics, promote informed debate about their implications for health and healthcare, and draw up strategic advice for ministers in an impartial forum. It has subsumed the functions of the Advisory Committee on Genetic Testing, the Human Genetics Advisory Committee and the Advisory Group on Scientific Advances in Genetics.

The Human Fertilisation and Embryology Authority (HFEA) on the other hand is a statutory body set up under the Human Fertilisation and Embryology Act (1990). Its duties include:

- to ensure that all UK treatment clinics offering in vitro fertilisation or donor insemination, or storing human eggs, sperm or embryos, conform to high professional and medical standards and are inspected regularly.

- to collect comprehensive data about these infertility treatments, and
- to provide detailed advice and information to the public.
- It also has the responsibility for licensing and monitoring of all human embryo research in the UK.

Another important professional committee is the Joint Medical Genetics Committee (JMGC) which was set up in 1998 to consider genetics services and their future, and to communicate with the DoH and the various Government advisory committees on issues concerning genetics. It represents the views of professional organisations and patient groups concerned with genetics: the British Society for Human Genetics, the Royal Colleges, the Faculty of Public Health Medicine, and the Genetic Interest Group. Its remit is

The Gene Therapy Advisory Committee (GTAC) is a non-statutory advisory body, which advises UK Health Ministers on developments in gene therapy research and their implications. It reviews and if appropriate approves individual protocols for gene therapy research. It works closely with the statutory body for medical products, the Medicines Control Agency (MCA), and with research ethics committees.

The Genetics and Insurance Committee (GAIC) has also been established as a non-statutory advisory body with a UK wide remit.

- to develop and publish criteria for the evaluation of specific genetic tests, their application to particular conditions and their reliability and relevance to particular types of insurance;
- to evaluate particular tests against those criteria and promulgate its findings; and
- to report to Health, Treasury and the Department of Trade and Industry Ministers, on proposals received by GAIC from insurance providers and the subsequent level of compliance by the industry with the recommendations of GAIC.

2. The vision for genetics in Wales

All fields of medicine will be utilising genetic advances in their practice, as well as in research. Therefore, genetics is viewed here not only from the speciality of clinical genetics and its activities, but also in relation to other specialties and to medicine overall. We should be aiming towards empowering health service professionals to be able to develop the genetic element of their health service and all other challenges that the new genetics may bring. Genetic educational strategy in the community, health service information strategy, the voluntary sector and patients organisations should ensure that patients are also empowered to deal with and understand their genetic disorders. The outcome should be well-informed patients, population and health professionals.

We should also be preparing for unprecedented changes in both the structures of health services and the way they are delivered. This includes hospital NHS specialties, primary care, and population screening programmes, pharmacogenetics and gene therapy.

3. The role of the Welsh Assembly (WE)

The WE should develop an overarching view of genetics in Wales that include NHS services, the private health sector, the voluntary sector, science and developments, the industry and the education sectors in Wales. The WE should coordinate all these governmental and extra-governmental agencies and ensure rapid translation of the genetic strategy into reality.

4. The Role of the Specialised Health Services Commission for Wales (SHSCW)

Commissioning at an all Wales level will continue, but it will include all genetic elements:

4.1.1. CGSW
4.1.2. Population Genetics Screening Programmes
4.1.3. Gene therapy
4.1.4. All genetics clinics based at different specialties

SHSCW will ensure that service delivery is equitable across Wales, with unified quality standards and outcome measures at all levels:

- Primary care
- Secondary care
- Tertiary care

SHSCW will ensure that Wales Strategy and services are in line with the UK policy and also part of Genetic Testing Network.

5. The role of Expert Clinical Genetics Service for Wales (CGSW)

Regional genetic centres are multidisciplinary, with clinical and laboratory services united or working closely together. Each centre is broadly similar, with a "hub and spoke" structure including specialist clinics and clinics in district hospitals and community facilities. Outreach staff from some centres may visit families at home
The clinical team comprises medical geneticists as well as counsellors whose background may be in nursing, science, or social work. Counsellors are becoming a recognised and registered profession, with formal postgraduate training in clinical practice and research.

Genetic laboratories would continue to offer analysis for monogenic diseases and, for rare disorders, to contribute to national and international networks for testing. They would lend their expertise to local partnerships, with other service providers developing high throughput systems for pharmacogenetic testing and population screening. This model of service is widely accepted and supported by the Joint Committee on Medical Genetics[3].

A core genetic service has been defined as (National Specialised Service Definition):
"An integrated clinical and laboratory service, provided for those with / concerned about a disorder with a significant genetic component, and their families (this includes inherited and sporadic genetic disorders) offering:

- Accurate clinical and genetic laboratory diagnosis
- Risk estimation (interpretative role)
- Genetic counselling (including pre and post predictive testing)
- Access to and overview of the latest science
- Accessible information for families/other health professionals and patient support groups (written and spoken)

- Interpretation and integration of complex genetic information
- Support to individuals and families (e.g. in decision making about future pregnancies)
- Prevention of a disorder or complications including family follow-up (e.g. anticipatory care, pre natal care and testing)
- Expert advice to other health professionals and commissioners
- Education and training for other health professionals including those providing genetic counselling within other specialised services (see below) and for undergraduate and postgraduate students
- Participation in research and clinical audit
- A family based approach where required
- Maintenance of confidential family records
- Maintenance of a DNA storage service
- Dealing with extended families long term

The concept of a regional centre serving 3 million people of Wales, has proved of great value and should be preserved and strengthened. The service will expand to accommodate increase in demand. Additional new responsibilities and roles are envisaged.

5.1. Consultant Geneticist Job portfolio is to be altered to include:

5.1.1. Developing interest with professional development in specified major areas of specialist genetics (neuromuscular, neurological and psychiatric genetics). Existing consultants already have one or more areas of interest and since these developments are gradual, the move towards increasing the interest portfolio should also be gradual and not prohibitive.

5.1.2. Together with the named specialist consultants and nurses, coordinate the genetic services delivered within these specialties.

5.1.3. Help in developing training syllabuses for medical and nursing staff within these specialties.

5.1.4. Develop a filtering system for high-risk families, by setting out referral guidelines to medical genetics in collaboration with the specialist consultant and nurse.

5.1.5. Ensure that quality standards and safeguards are developed for patients attending the all Wales genetics service are equally applied for other patients attending other sub-specialties

5.1.6. Work with these sub-specialties to develop audit and review of their parts of the service.

5.2. A consultant geneticist should develop an interest in primary care. He/she would liaise with the Primary Care Genetic Associates and identified GP trainers with special interest in genetics.

5.3. A consultant geneticist should develop an interest in population screening programmes and will liaise with the Screening Centre for Wales.

5.4. The genetic nurse specialists and associates will take on a more educational and coordinating role to link with specially trained nurses in other specialties and act as the first resource.

5.5. The number of consultants and genetic nurses is gradually increased to reflect the expected increase in demand for their clinical and laboratory services, and

also takes into account their new co-ordinating and training role for other specialties.

5.6. The Genetics Laboratory for Wales, with its molecular and cytogenetics arms is part of CGSW. The acquisition of equipment for high throughput DNA analysis will facilitate rapid genetic testing. DNA laboratories working for other specialties should coordinate their testing, which include utilising the facilities at CGSW. CGSW should develop in collaboration with other laboratories, standardised quality assurance measures and accreditation to be equally applied by all laboratories. The Wales Gene Park bid will facilitate such development.

To cope with the increasing demand for a much more diverse range of genetic tests, molecular geneticists have developed an informal network for genetic testing throughout the United Kingdom. This allows samples to be cross-referred between laboratories that offer different tests.

6. The Role of the proposed Wales Gene Park (WGP)

The establishment of WGP will enhance the development and the implementation of the all Wales strategy for Genetics in Wales.

6.1. Resources available will facilitate the implementation of the education and training, a key element of the strategy.

6.2. Horizon scanning element and technology transfer will facilitate the development and implementation of the strategy.

6.3. Large throughput DNA testing will be facilitated through this bid.

7. The role of hospital NHS Specialties (neurology, psychiatry, haematology…etc)

The consequences of the remarkable advances in molecular biology in recent years are likely to be experienced in every branch of medicine and medical practice. We should anticipate changes within every medical speciality and we have already seen some of these changes. Consequently, the nature of the roles and responsibilities assumed by clinical staff, physicians and nurses within these specialties will also change.

7.1. The speciality identifies one or two consultants who will develop the interest in genetics aspects of their speciality

7.2. Calman regulations should be reviewed and training arrangements put in place to enable a proportion of those training in each specialty to be accredited as a specialist with an interest in genetic medicine[4].

7.3. Joint specialist clinics with consultant geneticists are now frequent, notably in neurological, ophthalmic, cardiac and growth disorders genetics. Joint appointments between medical genetics and other specialties will accelerate[5].

7.4. Identifies a nurse (s) who will take-up the interest in genetics.

7.5. Both will liaise with a consultant geneticist and a genetic nurse at CGSW.

7.6. Both should be appropriately trained in genetics and counselling.

7.7. They will develop filtering system and referral guidelines with CGSW.

7.8. They should follow quality standards and develop audit and review of the service in collaboration with CGSW.

8. The role of Primary Care

Genetic counsellors (nurses/associates) are key to an integrated service. Based in the regional genetic centres with access to specialist resources, they already have responsibilities in outreach clinics, cancer and other units, and the community. As a development of the model they would be responsible for education and liaison between primary care teams and the regional genetic centre. There is a need to develop expertise in primary care to classify patients into risk groups from family history. Strong links with regional genetic centres will provide accurate information for the primary care team. In this model counsellors would serve as a first line of consultation, undertake some genetic counselling in the surgery, and facilitate referral of families at higher risk requiring specialist investigation and counselling.

9. The role of the 'Wales Screening Centre'

Population genetics screening programmes should involve CGSW. A consultant geneticist should be directly involved in the development and the delivery of these programmes. He/she should ensure that these programmes use the same service specifications, quality standards and outcome measures used for CGSW. He/she should be involved in the evaluation and review of these programmes. CGSW should ensure that genetic training packages are developed for staff delivering the service.

10. The education and training strategy

10.1. Specific genetics training programme should be set-up to accommodate the large number of health professionals who will need training.

10.2. To preserve and harness the existing genetic training resources and make the most of what we have, modular type courses are envisaged. Different professionals can chose different matrix of lectures to attend. A portfolio of relevant lectures is built into the programme for different specialties. Different lectures are run simultaneously in a designated building.

10.3. Resources are required to run such a programme efficiently.

10.4. Courses should be validated and approval should be sought from the royal colleges and various professional bodies.

11. The role of pharmacogenetics

Pharmacogenetics aims at understanding how genetic variation contributes to variations in response to medicines. The pharmaceuticals industry is translating genetic science to produce an entirely new range of drugs as well as diagnostic tests to predict the effectiveness of their products or to identify those who might be susceptible to adverse side effects. The variation that exists in all genes causes different members of a population to express different forms of proteins, including those that metabolise drugs or are the sites of drug action. This can lead to different responses to these drugs. Measuring the DNA differences can thus predict the variation in response to the medicine. In the past, systematic research into the basis of adverse drug reactions has been hampered by the fact that these events are rare and individuals are difficult to trace and study while suffering a reaction.

Drug companies are responsible for developing most new medicines or devices, so it is important to consider how they can collaborate with academics to expedite new therapies. Pharmacogenetics is increasingly driven by industrial researchers, partly because of their ready access to clinical trial data on which pharmacogenetic research

can be carried out. The rigours of drug registration require a high level of data quality (and hence high cost) that few academic groups can afford.

Increasingly, the common needs for research tools of both industry and academic researchers are leading to joint activities. A successful recent example is the SNP Consortium, a grouping of 13 companies, five leading academic centres, and a charity (the Wellcome Trust) established to identify 300 000 single nucleotide polymorphisms in the human genome and make the information public as a research tool for all.

Collaboration between genetic services and research on one side and the therapeutic industry will ensure rapid transfer of technology to service and will also harness new era of effective use and sharing of available resources. The Gene Park bid will facilitate such development. Development of guidelines in the use of new drugs should be coordinated by the consultant geneticist and the relevant speciality consultant with genetic interest in the specified clinical area.

12. The role of gene therapy

CGSW is part of a multidisciplinary team involved in research and development of such therapies. They are also involved in the planning and implementations of such treatment programmes. The Gene Park bid is important to the development of gene therapy in Wales.

13. The Strategy Development Plan

13.1. Immediate (one year)

13.1.1. The Wales Gene Park bid is an excellent opportunity to implement the early stages of the strategy.

13.1.2. Staff recruitment and training should start as soon as possible.

13.2. Medium term (2-3 years)

13.2.1. Development of consultant portfolio to emphasise the new areas of interest and responsibilities.

13.2.2. Training packages are developed for various professional groups.

13.2.3. Premises identified and teaching faculty identified.

13.2.4. Genetic activities carried out within other specialties are mapped, identified and costed for a contract that is separate from core genetic services.

13.3. Long term (3-10 years)

This needs a lot of thinking and discussions with other stake holders.

14. The Principles and standards

14.1. To ensure that the strategy for Genetic Services in Wales is part and a continuum of the strategy for Genetic services in the UK. This will include commissioning of services and laboratory genetic testing network.

14.2. Although CGSW will not be directly involved in delivering genetic services out with existing core genetic services, it has an ethical and moral duty to ensure that patients with genetic condition are treated with similar standards of care whichever service they access.

14.3. SHSCW through contractual arrangement ensures that patients with genetic condition are treated with similar standards of care whichever service they access. Similar quality standards and quality outcome measures are applied for all services utilising genetic testing and dealing with patients and families with genetic disorders.

14.4. Identification and naming of geneticists, interested clinicians, and nurses, who will coordinate, deliver and evaluate genetic services within different specialties, primary care and population screening programmes. Accountability is more clearly defined.

14.5. SHSCW and the Welsh Assembly to plan for expansion in medical and nursing workforce in CGSW to deliver the Strategy for Genetic Services in Wales covering other specialties and including education.

Figure 1
MEDICAL GENETICS SERVICE FOR WALES (MGSW)

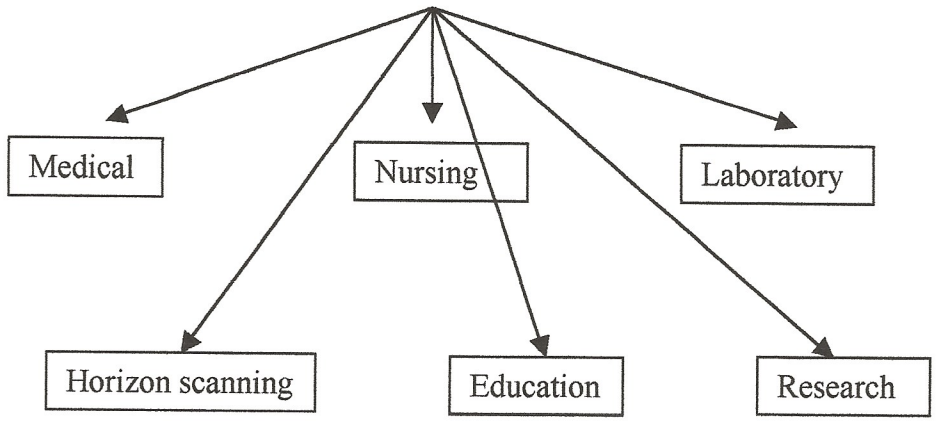


Figure 2

LINKS WITH THE NHS IN WALES

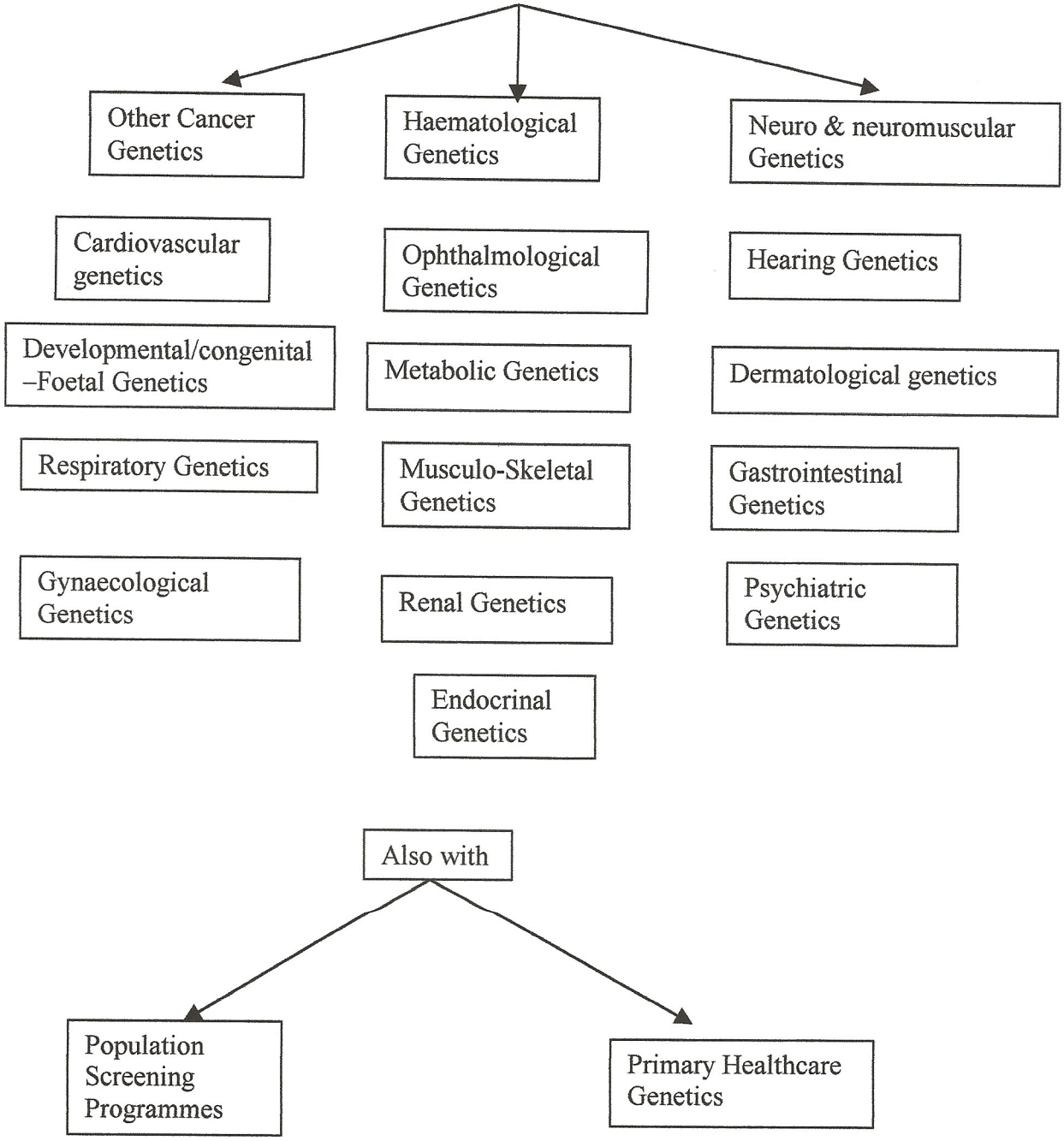
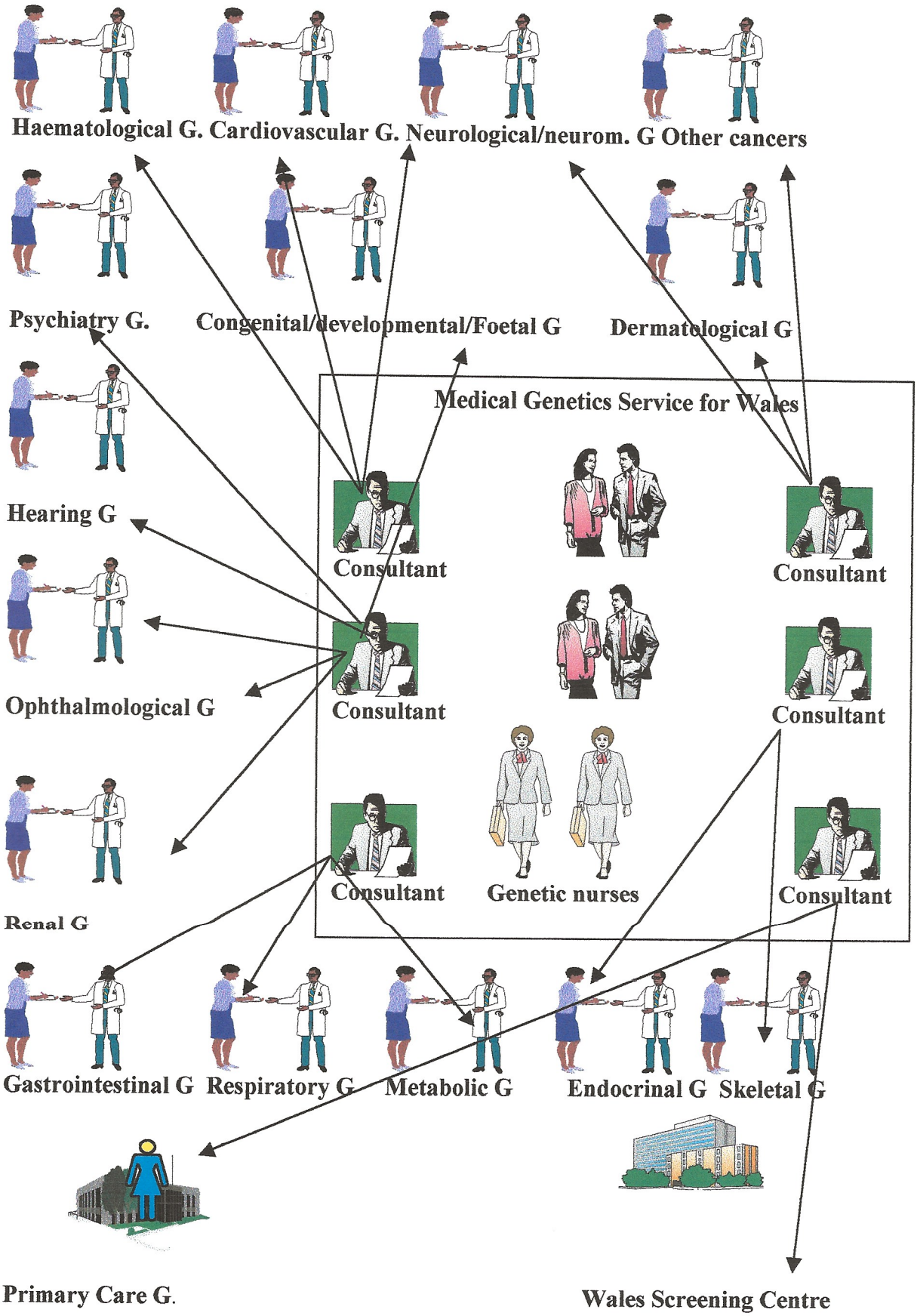
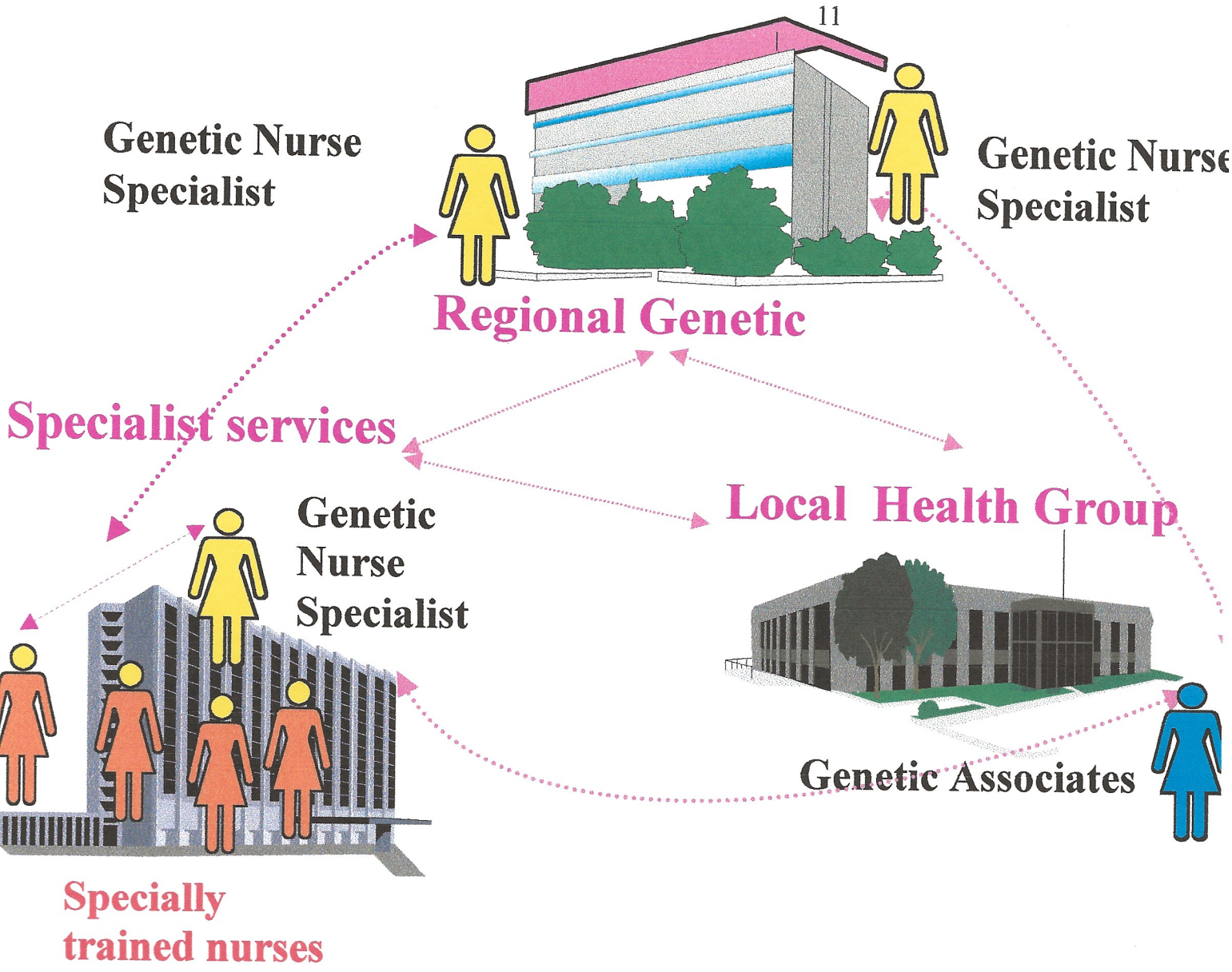


Figure 3


Haematological G.
Cardiovascular G.
Neurological/neurom. G
Other cancers
Psychiatry G.
Congenital/developmental/Foetal G
Dermatological G
Medical Genetics Service for Wales
Hearing G
Consultant
Consultant
Ophthalmological G
Consultant
Consultant
Renal G
Consultant
Genetic nurses
Consultant
Gastrointestinal G
Respiratory G
Metabolic G
Endocrinal G
Skeletal G
Primary Care G.
Wales Screening Centre



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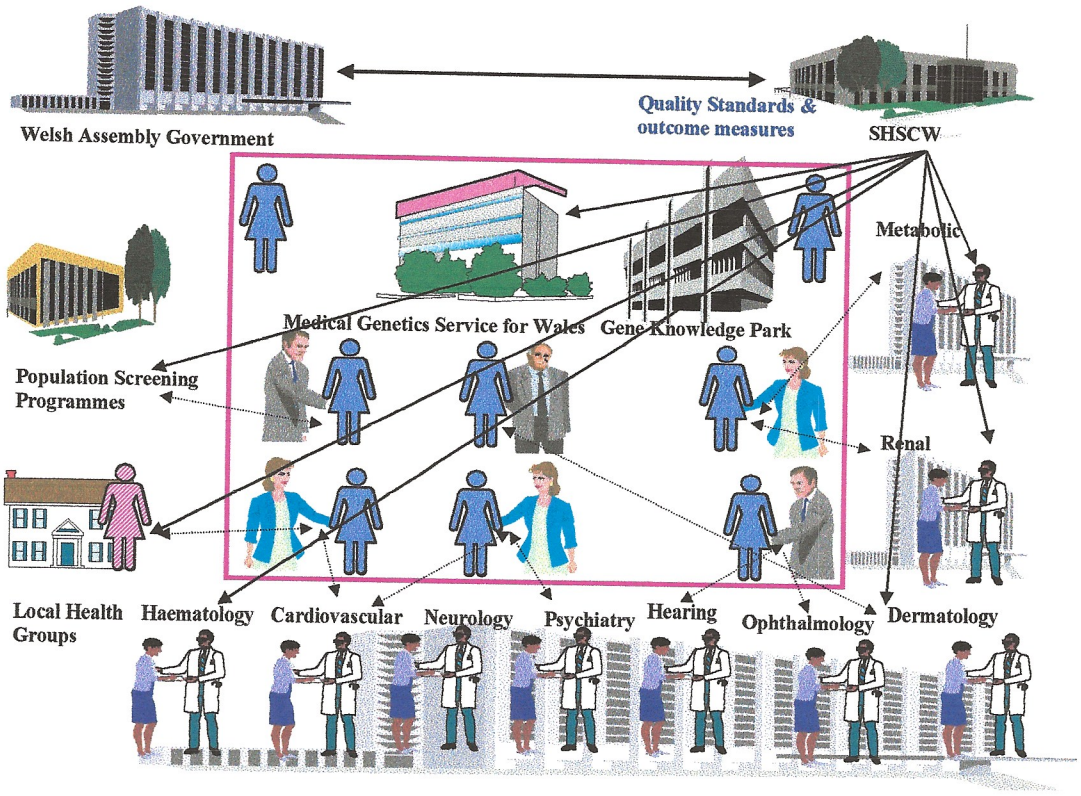
[4] Harper PS, Hughes HE, Reburn JA. Clinical genetics services into the 21st century. A report from the clinical genetics committee of the royal college of physicians of London, 1996.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Dermatology