

Specialised Testing for

Genetic Disorders

Part 1 - Recommendations for
Service Provision

Better Health Good Health Care

NSW  **HEALTH**

NSW HEALTH DEPARTMENT

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Table of contents

Executive Summary	1
Summary of Recommendations	2
Definition of Terms	4
1 Background	5
2 Need for change in policy direction	6
2.1 Improved testing techniques	6
2.2 Introduction of new tests	6
2.3 The broadening scope of DNA testing for genetic disorders	6
2.4 Research developments	7
2.5 Impact of pathology networking	7
3 Policy context and direction	8
3.1 Goals of the NSW Genetics Service	8
Healthier people	8
Fair access	8
Quality health care	8
Better value	8
4 Recommendations for the provision of specialised testing services for genetic disorders	9
4.1 Guidelines for clinical practice, genetic counselling and risk assessment	9
4.2 Guidelines to indicate appropriateness of specialised genetic testing as a diagnostic option	10
4.3 Request and consent forms	10
4.4 Collection and transport of specimens	11
4.5 Laboratory standards	11
4.6 Provision of new tests	12
4.7 Information, educational resources clinical and counselling services	13
5 Funding of specialised genetic testing services	14
5.1 Specialised genetics testing laboratories and pathology networking	14
5.2 Current funding position	14
5.3 Future funding options	15
5.4 Cost recovery/user pays	15
5.5 Requesting and paying for tests	16
5.6 Costs of tests	16
5.7 Access to specialised genetic testing	17
5.8 New Technologies	17
6 Conclusion	18

Executive Summary

A number of recent developments in specialised genetic testing make it timely to review the way services are provided. These developments include advances in DNA technology, simpler techniques, an increasing number of disorders for which specialised genetic testing is advantageous and networking of pathology services.

Specialised genetic testing may benefit individuals and families in a number of ways but it may also create dilemmas which need sensitive management. Each test has distinct advantages, disadvantages and limitations and should only be used after the individual being tested has given full consideration to the relevant issues.

The document is presented in two parts:

1. Recommendations for service provision

A policy statement with recommendations for best practice guidelines and for funding specialised genetic testing services.

2. Guidelines for Specialised Testing for Genetic Disorders

The second part of the document is in the form of a user manual to guide health professionals through the process of considering or using genetic testing as a diagnostic option. In order to achieve a high standard of service, it is important that health professionals are aware of the special issues concerning specialised testing for genetic disorders which differ from those associated with other medical interventions.

Key features of the recommendations and management guidelines include:

- Guidelines for genetic counselling and risk assessment associated with testing for genetic disorders to ensure individuals are fully informed about the advantages, disadvantages, limitations and other relevant issues before opting for testing.
- Guidelines to indicate appropriateness of specialised genetic testing as a diagnostic option.
- Request and consent form templates to foster best clinical practice and procedures for using specialised genetic testing.
- Guidelines for collection and transport of specimens.
- Repertoire of tests and laboratory locations.
- Laboratory standards for specialised genetic testing for specific disorders.
- Guidelines for assessing and introducing new technologies.
- Information on educational resources and locations of clinical and genetic counselling services.
- Recommendations for funding specialised genetic testing and consideration of the relationship between genetics services and pathology networks.

Summary of Recommendations

Guidelines for clinical practice, genetic counselling and risk assessment

- 1 *All testing should be carried out only after individuals have been fully informed about the advantages, disadvantages, limitations and other relevant issues concerning testing, and have consented to testing. Refer to Part II, Section 1 "Guidelines for Specialised Testing for Genetic Disorders".*

Guidelines to indicate appropriateness of specialised genetic testing as a diagnostic option.

- 2 *Existing guidelines for referral for specialised genetic testing endorsed by other bodies such as the NHMRC, Australian Cancer Network or Human Genetics Society of Australasia should be reviewed for adoption. As technology advances the NSW Genetics Service Advisory Committee, in consultation with relevant professionals and organisations, should monitor or develop new guidelines as appropriate.*
- 3 *The relevance of specialised genetic testing as a diagnostic option should be considered during the clinical and genetic counselling process, so that testing is only offered to individuals for whom it is appropriate or informative.*

Test request and consent forms

- 4 *Template test request and consent forms included in Part II, Section 2 should be submitted to Institutional Medical Records Committees for approval for use.*
- 5 *Test request and consent forms should be completed for all individuals being tested. Refer to Part II, Section 2 for template forms. The request form includes a section to indicate whether clinical and genetic counselling guidelines have been followed.*
- 6 *A completed request form should accompany samples sent to the laboratory. Completed consent forms should be retained in the clinical unit. Individuals being tested should be provided with a copy.*
- 7 *Laboratories should adopt an educative role, so that where test requests do not comply with guidelines, referring health professionals can be informed of correct procedures for specialised genetic testing.*

Collection and transport of specimens

- 8 *All samples sent for testing should be collected according to the general guidelines in Part II, Section 3, and disorder specific requirements.*

Laboratory standards for specialised genetic testing

- 9 *Laboratories offering specialised genetic testing should meet standards set by NATA/RCPA accreditation, National Pathology Accreditation Advisory Council (NPAAC) and ISO/IEC Guide 25. (Part II, Section 4)*
- 10 *Standards for specific genetic disorders should be adopted or developed through consultation by Heads of Laboratories providing specialised genetic testing and other relevant organisations. (Part II, Section 4)*

Provision of new tests

- 11 *When technological developments make a new test possible, its potential for service provision should be investigated. The NSW Genetics Service Advisory Committee should coordinate this process, develop appropriate guidelines for testing, and advise NSW Health on the value of introducing a testing service and potential service locations.*

Information, educational resources, clinical and genetic counselling services

- 12 *Approved guidelines should be made available through mechanisms that make access effective, including the Internet. Information about educational resources (Part II, Section 5) and clinical and genetic counselling service locations (Part II, Section 6) should be updated regularly.*

Funding of specialised genetic testing services (non Medical Benefits Schedule items)

- 13 *Molecular genetics laboratories should be involved in the network of pathology services and adopt business unit practices including cost recovery for specialised genetic testing.*
- 14 *Existing laboratory funding should be retained in laboratories to support the changeover to user pays processes. Area Health Services should meet the cost of testing from within their population based resource share.*
- 15 *Cost recovery/user pays should be introduced for all specialised genetic tests which are non Medical Benefits Schedule items, with charges levied upon the facility requesting the service. Where the facility requesting the test is not in the patient's Area Health Service, the cost of testing may be referred back to the Patient's Area Health Service of residence provided a written memorandum of agreement has been negotiated between the Area Health Services.*
- 16 *Area Health Services should negotiate with clinicians who request specialised (non MBS) genetic testing to nominate a local authority/person to approve test requests, to approve payments and to ensure testing is offered to those for whom it is relevant and in accordance with guidelines in Recommendations 1 - 6.*
- 17 *Specialised genetics testing laboratories need to determine the true cost of tests, including staffing costs, consumables, capital costs, depreciation, other overheads and a percentage for continuing research and development, by June 2000.*
- 18 *Low volume/high cost/complex tests should each continue to be provided from a single laboratory for the State.*
- 19 *The NSW Genetics Service Advisory Committee should retain overview of developments in specialised genetic testing and advise NSW Health on mechanisms to meet the needs of the community for access to genetic testing.*

Definition of Terms

Genetic disorders result usually, but not always, from a change in the hereditary (genetic) material contributed by the parents at the time of conception. Symptoms may be apparent at birth or may occur later in life. Genetic factors make a substantial contribution to physical and intellectual disability, chronic ill-health, psychiatric illness and familial cancer. Because genetic disorders are family health problems, a diagnosis in one member has implications for other family members.

For the purpose of this document **specialised genetic testing** refers to tests which are funded by NSW Health through its public laboratories and which are non Medical Benefits Schedule items.

The tests are used to:

- diagnose a genetic disorder
- determine if a person is a mutation carrier for a disorder, or
- detect inherited predisposition to a genetic disorder.

using the following techniques or processes:

- molecular genetic (DNA) testing, including PCR based methods
- biochemical testing, including functional studies

It is to be noted that the scope of this definition does not include tests for non-inherited disorders which may use the same testing techniques, for example the diagnosis of bacterial, viral or malignant conditions for therapeutic purposes, or testing for multifactorial disorders, which are the result of an interaction of multiple genes with environmental factors.

1 Background

For the past 10 years, specialised testing for genetic disorders has been available through NSW public hospital laboratories in South Eastern Sydney, Central Sydney, Hunter, Northern Sydney and at the Royal Alexandra Hospital for Children. The laboratories are closely associated with clinical genetics units.

Molecular and biochemical genetics laboratories have been supported through recurrent Department of Health allocations between 1987/88 and 1990/91 and through Area Health Service financial support during and since that time. Further funding has come from cross subsidisation and private sources.

Until recently there has been no private sector involvement in provision of DNA testing. With the exception of a newly created Medicare Item for haemochromatosis testing, there are no Medicare rebates for DNA tests for other genetic disorders.

Laboratories have operated under a consortium model where each laboratory tested a small number of genetic disorders but each genetic disorder was tested in only one laboratory. The rationale for NSW adopting a consortium approach rested on two arguments, the first being the rarity of individual genetic disorders is such that individual laboratories would see too few cases to maintain proficiency of diagnosis and interpretation. The second argument was an economic one, that specialised genetic testing services have low sample numbers but high consumable and labour costs. It was anticipated that a consortium model would be likely to minimise costly duplication of services.

Laboratory locations and their test repertoires are listed in Part II, Section 4. Laboratory Directors form the DNA Testing Working Party which reports to the NSW Genetics Service Advisory Committee.

When a new test becomes available the Working Party agrees on the most appropriate laboratory for service provision. A recommendation is then made from the NSW Genetics Service Advisory Committee to the Director-General of NSW Health.

2 Need for change in policy direction

The consortium approach has worked well in the past for rare disorders but the climate in which public hospital molecular and biochemical genetics laboratories are working has changed considerably over the last decade and is expected to change even more rapidly over the next ten years. Changes include:

2.1 Improved testing techniques

Changes in knowledge and methodology have simplified some forms of existing testing, creating opportunities for expansion in numbers of tests for specific disorders and for provision of these services by more than one laboratory.

As an example, cystic fibrosis (CF) carrier testing is one of the more frequently tested genetic disorders. Overall in NSW, 1 in 25 individuals is a carrier of a mutant CF gene. Carriers are not affected by CF but each pregnancy to a couple who are both carriers has a 1 in 4 risk of producing a child with CF. Each year in NSW about 35 children are born with the disease. There are over 700 mutations, the most common of which is present in 75% of CF carriers of Northern European descent. The next five most common mutations take the cumulative frequency of identifiable CF carriers to 82%. Kits are becoming available which detect an increasing number of mutations. The costs vary according to the kit supplier and specific mutations in the kit, but fall within the range of \$40 - \$80 per sample analysed. Further developments, coupled with increasing demand for testing are likely to produce a lower unit cost.

2.2 Introduction of new tests

Technological developments make it possible to use DNA testing for the diagnosis of an increasing number of genetic disorders, eg haemochromatosis, spinal muscular atrophy and Huntington disease.

Specialised genetic testing can be used as an alternative to more invasive forms of testing. For example, testing for familial retinoblastoma susceptibility reduces the need for regular examination, under anaesthetic, as a diagnostic method.

About 5% of breast and colorectal cancers involve an inherited predisposing mutation and it is now possible to offer predictive testing to family members at high risk where the mutation has been identified in the incident case family member. Using familial adenomatous polyposis as an example, individuals identified at high risk presymptomatically, can be closely monitored with early intervention when necessary. Where family members are found not to carry a mutation, they can avoid regular endoscopic examination from an early age.

2.3 The broadening scope of DNA testing for genetic disorders

DNA testing has been an integral part of a clinical genetics service. However, as technology develops it is being used more widely in the diagnosis of inherited disorders by other clinicians such as neurologists, oncologists, gastroenterologists, cardiologists or general practitioners. Some laboratories also wish to offer a wider range of tests.

For example, haemochromatosis is the most common inherited disorder in the population with 1 in 10 to 1 in 15 people carrying the mutation (heterozygosity). Three to 5 per 1000 people have the mutation in both copies of the gene involved (homozygosity). It is thought that many homozygous individuals never become symptomatic, particularly females because of their regular blood loss. However, it can result in skin pigmentation coupled with diabetes mellitus, arthropathy, impotence, fatigue, hepatic cirrhosis and predisposition to hepatoma and cardiomyopathy. DNA testing can confirm early diagnosis and where appropriate simple intervention by venesection can reduce associated organ damage and achieve near normal life expectancy. A single mutation accounts for between 85 and 100% of cases. DNA testing would provide a simple alternative to liver biopsy as a diagnostic tool.

2.4 Research developments

As developments cross the bridge from research to service delivery, more than one laboratory is likely to provide the service. Haemochromatosis testing is being offered by a number of molecular genetics laboratories and pathology services.

2.5 Impact of pathology networking

Most, but not all, molecular genetics laboratories have been incorporated into the administrative structure of pathology services and are subject to the NSW Health processes of pathology networking.

3 Policy context and direction

3.1 Goals of the NSW Genetics Service

Healthier people

To reduce the impact of genetic disorders and birth defects on affected individuals, individuals at risk and their families

Fair access

To enhance patient and family wellbeing through provision of conveniently located genetics services and access to specialised testing where appropriate

Quality health care

To maximise genetic health outcomes through provision of information, clinical, counselling and laboratory services

Better value

To support the appropriate development of clinical and laboratory services and implementation of beneficial new technologies

The recommendations of this document support the goals of the NSW Genetics Service and reflect commitment to NSW Health's Strategic Directions for Health 1998-2003.

A major aim of genetics services is to provide individuals and families with information, education, clinical, genetic counselling and diagnostic services and support to assist them where there is a disorder with an hereditary or genetic basis.

4 Recommendations for the provision of specialised testing services for genetic disorders

4.1 Guidelines for clinical practice, genetic counselling and risk assessment

Guidelines for testing for genetic disorders have been set out in NSW Health Department Circular 97/48 and in Part II, Section 1.

The rarity of individual genetic disorders, low sample numbers and high testing costs mean that specialised genetic testing is most appropriately offered to individuals with a positive family history, high risk or clinical indication.

Whilst genetic testing has acknowledged value, it also has limitations and the potential to raise complex personal issues. Therefore, it should only be used in the context of provision of full information about the implications of testing which include:

- the risk of developing a genetic disorder
- implications for other family members
- prognosis
- interventions
- privacy
- confidentiality
- insurance
- employment issues

Testing should be carried out only with informed consent of the person after they have had the opportunity to fully consider all the relevant issues. A sample consent form is included in Part II, Section 2.

Results should be made available to the patient by the referring practitioner with regard to all implications of genetic testing. It is important that all health professionals requesting tests are fully aware that the diagnosis of a condition in one family member will confer risks on other family members. Health professionals who have not been trained in genetics may need to transfer responsibility or consult with those with expertise in this area. Clinical and genetic counselling services are listed in Part II, Section 6.

Recommendations

- 1 *All testing should be carried out only after individuals have been fully informed about the advantages, disadvantages, limitations and other relevant issues concerning testing, and have consented to testing. Refer to Part II, Section 1 "Guidelines for Specialised Testing for Genetic Disorders".*

4.2 Guidelines to indicate appropriateness of specialised genetic testing as a diagnostic option

The repertoire of tests currently available in NSW and the laboratories providing them are listed in Part II, Section 4.

When the benefits of a test have been established, access should be in accordance with agreed principles of need, so that testing is available to those for whom it is relevant. Indications for testing would include, for example, positive family history, high risk or clinical indication.

In some instances guidelines have been developed and are available from organisations such as the NSW Genetics Service Advisory Committee, the Human Genetics Society of Australasia and the Australian Cancer Network, and are listed in Part II. Where guidelines do not exist it is appropriate to develop them through consultation with relevant organisations and health professionals.

Where improved techniques enable higher throughput for an existing test, it may be appropriate to alter priorities after giving consideration to cost/benefit issues such as increasing categories of people to whom that test should be available, or using efficiency gains to provide new tests.

Recommendations

- Existing guidelines for referral for specialised genetic testing endorsed by other bodies such as the NHMRC, Australian Cancer Network or Human Genetics Society of Australasia should be reviewed for adoption. As technology advances the NSW Genetics Service Advisory Committee, in consultation with relevant professionals and organisations, should monitor or develop new guidelines as appropriate.*
- The relevance of specialised genetic testing as a diagnostic option should be considered during the clinical and genetic counselling process, so that testing is only offered to individuals for whom it is appropriate or informative.*

4.3 Request and consent forms

Test request and consent forms have been developed to foster best clinical practice and procedures for collecting samples. Templates are included in Part II, Section 2. An appropriately completed request form will ensure laboratories are provided with essential information to support sample analysis. It will act also as a support tool to remind referring health professionals that optimal patient care in the clinical setting includes provision of relevant pretest information, genetic counselling and obtaining of consent.

Recommendations

- Template test request and consent forms included in Part II, Section 2 should be submitted to Institutional Medical Records Committees for approval for use.*
- Test request and consent forms should be completed for all individuals being tested. The request form includes a section to indicate whether clinical and genetic counselling guidelines have been followed.*

- 6 *A completed request form should accompany samples sent to the laboratory. Completed consent forms should be retained in the clinical unit. Individuals being tested should be provided with a copy.*
- 7 *Laboratories should adopt an educative role, so that where test requests do not comply with guidelines, referring health professionals can be informed of correct procedures for specialised genetic testing.*

4.4 Collection and transport of specimens

General guidelines have been developed based on NATA guidelines to ensure that appropriate specimens are taken and are received by the laboratory in good condition (Part II, Section 3). There may be disorder specific requirements and these are available from clinical units and the testing laboratories listed in Part II, Section 4.

Recommendations

- 8 *All samples sent for testing should be collected according to the general guidelines in Part II, Section 3, and disorder specific requirements.*

4.5 Laboratory standards

Tests which are rarely requested or difficult to perform and tests which require reasonable sample numbers to maintain specialist knowledge and appropriate interpretive skills should continue to be undertaken in one laboratory.

Testing should be carried out in laboratories either with or working towards NATA/RCPA accreditation. Laboratories should meet standards set by the National Pathology Accreditation Advisory Council (NPAAC) and ISO/IEC Guide 25 with regard to appropriate staff training, staff levels, facilities, health and safety, specimens, equipment and instrumentation, methods, quality management, reporting and records.

In addition to NATA/RCPA requirements, laboratories should meet specific criteria for specialised testing for genetic disorders endorsed by relevant organisations such as the Human Genetics Society of Australasia and the NSW Genetics Service Advisory Committee. These criteria include: ability to provide, either individually or in collaboration, the complete suite of tests to the level accepted as the standard of care for the specific disorder they are testing; quality assurance standards for minimum throughput; test results; turn around time and outcome reporting; technical expertise; validated methods and procedures; quality assurance and appropriate provision of results.

Testing should only be undertaken on specimens received in appropriate condition and accompanied by the completed request form and family pedigree, where appropriate.

Result reports should contain:

- collection date and identifying information
- indication for testing
- method used (including unpublished modifications)
- molecular data
- interpretation of the raw data in clear and concise text which is appropriate for a health professional who is not a geneticist

- an indication of potential implications for other family members
- details of further tests or information that may be required
- a statement regarding the possibility of inaccuracy, for example due to non-paternity.

Results should be made available to patients via the referring practitioner. There should be close liaison between laboratory staff and clinicians concerning the interpretation of results. See *Genetic counselling and risk assessment Part 2, Section 1*.

Recommendations

- 9 *Laboratories offering specialised genetic testing should meet standards set by NATA/RCPA accreditation, National Pathology Accreditation Advisory Council (NPAAC) and ISO/IEC Guide 25. (Part II, Section 4)*
- 10 *Standards for specific genetic disorders should be adopted or developed through consultation by Heads of Laboratories providing genetic testing and other relevant organisations. (Part II, Section 4)*

4.6 Provision of new tests

With technological advances, the range of tests potentially available is rapidly increasing. The NSW Genetics Service Advisory Committee will continue to develop guidelines and make recommendations to NSW Health on the appropriateness of introducing new services with regard to:

- clinical indications for testing
- which laboratory/ies should or could offer which tests

Evidence based pathways should be developed which provide rigorous and robust criteria for offering tests. Issues to be considered include testing capabilities, criteria for test availability, sensitivity and specificity of tests, costs and benefits, risk categories for access, numbers in the community for whom testing is appropriate, pretest genetic counselling requirements, quality control measures in laboratories and recommended throughput, and whether it is appropriate to offer a particular test from one or more laboratories.

In general tests which are rarely requested or difficult to perform and tests which require reasonable sample numbers to maintain specialist knowledge and appropriate interpretive skills should be undertaken at limited number of sites. It may be appropriate for more than one laboratory to provide testing where there is higher throughput or simpler methodology. For some of the rarer diseases it may be appropriate that the services of interstate or overseas laboratories are used.

Testing should be available for medically indicated conditions only. Sex determination of a fetus should only be performed for health or medical benefit purposes, eg sex linked conditions. Alternative testing arrangements are available for paternity testing and relationship testing.

The role of private laboratories falls outside the scope of this document. However, in the interests of quality testing and appropriate service delivery, they are encouraged to consider the recommendations herein.

Recommendations

- 11 *When technological developments make a new test possible, its potential for service provision should be investigated. The NSW Genetics Service Advisory Committee should coordinate this process, develop appropriate guidelines for testing, and advise NSW Health on the value of introducing a testing service and potential service locations.*

4.7 Information, educational resources, clinical and counselling services

The approved guidelines should be made available to health professionals requesting specialised genetic testing. Where necessary, laboratories could adopt an educative role in promulgating the guidelines to referring health professionals.

The NSW Genetics Education Program produces a variety of resources to assist with health professional and patient education. These and other resources are listed in Part II, Section 5. The Genetics Education Program lists all currently available information on the Internet: <http://www.genetics.com.au>.

Clinical genetics and genetic counselling services are available in a number of locations in NSW. Health professionals are encouraged to make use of the expertise of their local clinical genetics units which are easily contacted by phone or fax. Details of clinical genetics, genetic counselling, cancer genetics and prenatal diagnosis services are listed in Part II, Section 6.

Recommendations

- 12 *Approved guidelines should be made available through mechanisms that make access effective, including the Internet. Information about educational resources (Part II, Section 5) and clinical and genetic counselling service locations (Part II, Section 6) should be updated regularly.*

5 Funding of specialised genetic testing services

The following discussion and recommendations relate to specialised genetic tests which are funded by NSW Health through its public laboratories and which are non Medical Benefits Schedule items.

5.1 Specialised genetics testing laboratories and pathology networking

The networking of pathology services has substantially advanced in NSW. Networking means that service units work in a more coordinated way, both within and across health service boundaries to enhance the efficient and effective provision of services.

The restructure of pathology services as business units has incorporated user pays/cost recovery processes. The user pays process has been accepted as the option for providing new genetic tests with the introduction of familial colorectal cancer DNA testing.

The potential exists for specialised genetics testing laboratories to become part of pathology networks. All these services are based within teaching hospitals. With the exception of the Department of Molecular Genetics within the Western Sydney Genetics Program at RAHC and the Familial Cancer Service Laboratory at Westmead, they have practical relationships with pathology hubs. The incorporation of molecular genetics laboratories into pathology hubs is likely to achieve administrative and financial efficiencies. Discussions between the Peak Pathology Council and the NSW Genetics Service Advisory Committee indicate that this is feasible.

Recommendation

13 Molecular genetics laboratories should be involved in the network of pathology services and adopt business unit practices including cost recovery for specialised genetic testing.

5.2 Current funding position

At the present time each laboratory is funded by a variety of sources, to differing levels:

- 1 Departmental allocations make up approximately one quarter to one third of current molecular genetics funding. This funding was allocated between 1987 and 1991 to Area Health Services under Program 2.3 to enhance laboratory services. The proportion of Statewide funding in individual laboratories varies significantly.
- 2 Area Health Service Funding
- 3 Cross subsidisation within laboratory services where molecular genetics testing is supported by revenue from other (Medical Benefits Schedule rebatable) laboratory services
- 4 Private funding
- 5 Research funding

5.3 Future funding options

Several funding options for the future have been considered:

- No change
- Pooled Funding for Statewide specialised genetic testing
- Cost recovery/user pays

The first two options are not recommended. Concerning the current funding position, initial enhancement funding was effective in the early development of specialised genetic testing. However, growth in demand has outstripped laboratories' capacity to provide testing. To some extent this growth has been met by Area Health Services, but there are inequities where a single laboratory provides a statewide service.

Pooled funding would require an accurate estimate of service use by Area Health Services with funds put aside in an identifiable pool. As volume is difficult to estimate at a single point in time, services may be over or under funded. Allocations are likely to lag behind increases in testing causing budget overruns or delays in service. Both options have no control on test requests at the clinical level and all costs are borne by the laboratory.

5.4 Cost recovery/user pays

The NSW Genetics Service Advisory Committee in consultation with NSW Health and the Peak Pathology Council supports the option of cost recovery/user pays. It is seen as a more flexible option which would be more responsive to change.

Under this model each Area Health Service would use resources from its population based resource distribution share, to provide or purchase specialised genetic testing for its residents in conjunction with appropriate clinical and genetic counselling services.

As the proportion of initial Departmental funding is a small component of total requirements and it is difficult to identify and separate from other funds, this funding should be retained in laboratories to provide support for laboratory services during the change over to user pays processes.

Charging should be introduced for all specialised genetic tests which are non Medical Benefits Schedule items with the exception of newborn screening and some biochemical tests which are funded separately.

An Area Health Service would either provide specialised genetic testing or become the purchaser on behalf of its residents for services not provided within its Area. Areas may choose to negotiate service agreements using:

- Block contracts
- Price per case contracts
- Price and volume contracts

Care will be needed to ensure that access to specialised genetic testing is equitable regardless of Area of residence.

Recommendations

- 14 *Existing laboratory funding should be retained in laboratories to support the changeover to user pays processes. Area Health Services should meet the cost of testing from within their population based resource share.*

5.5 Requesting and paying for tests

Specialised genetic tests are mainly requested by clinical geneticists but are also requested by other clinicians, specialists and general practitioners.

Charges for tests should be levied upon the facility requesting the service. Negotiations at a local level would identify which facility or clinicians would have authority to request testing, to ensure there is an appropriate budget, to ensure testing is made available to people for whom it is appropriate (see referral guidelines for specific disorders Part II, Section 4), to approve payments, and to ensure guidelines are met, where relevant, for:

- Providing appropriate genetic information about the advantages and limitations of testing prior to testing
- pre test counselling
- risk assessment
- consent for testing
- completing laboratory request forms
- post test counselling

Recommendations:

- 15 *Cost recovery/user pays should be introduced for all specialised genetic tests which are non Medical Benefits Schedule items, with charges levied upon the facility requesting the service. Where the facility requesting the test is not in the patient's Area Health Service, the cost of testing may be referred back to the Patient's Area Health Service of residence provided a written memorandum of agreement has been negotiated between the Area Health Services concerned.*
- 16 *Area Health Services should negotiate with clinicians who request specialised (non MBS) genetic testing to nominate a local authority/person to approve test requests, to approve payments and to ensure testing is offered to those for whom it is relevant and in accordance with guidelines in Recommendations 1 - 6.*

5.6 Costs of tests

Under the previous funding system it has not been necessary for laboratories to identify all costs associated with testing or determine costs of individual tests. Adopting a cost recovery/user pays system, will require costs for tests to be determined, based on all components of service provision. Test prices should be based on real costs for specific tests and not subsidised by other testing.

Recommendation

- 17 *Specialised genetics testing laboratories need to determine the true cost of tests, including staffing costs, consumables, capital costs, depreciation, other overheads and a percentage for continuing research and development, by June 2000.*

5.7 Access to specialised genetic testing

Funding processes should be structured to ensure the continued provision of, and access to, specialised testing for genetic disorders which are currently or potentially diagnosable provided benefits are monitored.

Service provision should be on a collaborative and cooperative basis. Essentially this is likely to mean that for low volume/high cost/complex testing where guidelines and throughput indicate a disorder should be studied in one laboratory only, it would remain in the laboratory where it is currently and where the expertise lies.

A more competitive approach would be possible for higher throughput tests where guidelines indicate appropriateness of undertaking testing in more than one laboratory.

Concerning the position of the laboratories outside the hub network, they have the option of joining a hub network or developing their own business unit approach and cost recovery processes for services which fall outside their patient base.

With the exception of haemochromatosis testing, currently there is no fee-for-service work of this type through the Pathology Services Table of the Medicare Benefits Schedule. Negotiations will be progressed with the Commonwealth through the Medical Services Advisory Committee.

Recommendations

- 18 *Low volume/high cost/complex tests should each continue to be provided from a single laboratory for the State.*
- 19 *The NSW Genetics Service Advisory Committee should retain overview of developments in specialised genetic testing and advise NSW Health on mechanisms to meet the needs of the community for access to genetic testing.*

5.8 New Technologies

The financial impact of new technologies cannot be estimated. It is likely that within the next five years, technology will simplify and automate some types of testing. However, the cost of this technology is not known and therefore cannot be factored into test prices.

6 Conclusion

Although genetic disorders are rare individually, collectively they contribute a significant proportion of the burden of ill-health. Due to the rarity of specific genetic disorders it is likely that there will be a continuing need for specialised genetic testing services.

It is possible and appropriate to retain specialised testing services, at the same time as allowing responsibility for appropriate use of tests to rest with the users; ie clinicians on behalf of the patients they serve; and, Area Health Services who are responsible for health care services for their residents.

Where appropriate, the recommendations of this document have been adopted and reproduced in Part II of this document in the form of a user manual to guide health professionals through the process of considering or using genetic testing as a diagnostic option.