Guideline

Genetic Testing - Guidelines for prioritising genetic tests

Summary Many genetic tests provided by NSW public hospital laboratories are non-Medical Benefits Schedule items funded through NSW Health. The attached guidelines have been developed to assist clinicians and health services to prioritise genetic test requests based on clinical need, equity of access and within available funding levels.

Document type Guideline

Document number GL2007_013

Publication date 06 July 2007

Author branch Agency for Clinical Innovation

Branch contact 02 94644606

Review date 15 December 2017

Policy manual Not applicable

File number 07/6580

Previous reference N/A

Status Review

Functional group Clinical/Patient Services - Medical Treatment, Pathology

Applies to Area Health Services/Chief Executive Governed Statutory Health Corporation, Affiliated Health Organisations, Affiliated Health Organisations - Declared, Public Hospitals

Distributed to Public Health System, Divisions of General Practice, Health Professional Associations and Related Organisations, NSW Ambulance Service, Ministry of Health, Public Health Units, Public Hospitals, Private Hospitals and Day Procedure Centres

Audience Clinical;administration

Guideline

Department of Health, NSW
73 Miller Street North Sydney NSW 2060
Locked Mail Bag 961 North Sydney NSW 2059
Telephone (02) 9391 9000 Fax (02) 9391 9101
http://www.health.nsw.gov.au/policies/

Genetic Testing - Guidelines for prioritising genetic tests

Document Number GL2007_013

Publication date 06-Jul-2007

Functional Sub group Clinical/ Patient Services - Medical Treatment
Clinical/ Patient Services - Pathology

Summary Many genetic tests provided by NSW public hospital laboratories are non-Medical Benefits Schedule items funded through NSW Health. The attached guidelines have been developed to assist clinicians and health services to prioritise genetic test requests based on clinical need, equity of access and within available funding levels.

Author Branch Statewide Services Development

Branch contact Jennifer Blackwell 9391 9520

Applies to Area Health Services/Chief Executive Governed Statutory Health Corporation, Affiliated Health Organisations - Non Declared, Affiliated Health Organisations - Declared, Public Hospitals

Audience Clinical, administration

Distributed to Public Health System, Divisions of General Practice, Health Professional Associations and Related Organisations, NSW Ambulance Service, NSW Department of Health, Public Health Units, Public Hospitals, Private Hospitals and Day Procedure Centres

Review date 06-Jul-2012

File No. 07/6580

Status Active

Director-General

GENETIC TESTING
Guidelines for prioritising genetic tests

Introduction

Many genetic tests provided by NSW public hospital laboratories are non-Medical Benefits Schedule items funded through NSW Health. The charging policy for these tests is addressed in Policy Directive PD 2005_335. Further, the Policy Directive requires testing to be assessed and prioritised according to clinical necessity.

The attached guidelines have been developed to assist clinicians/health services to prioritise genetic test requests based on clinical need, equity of access and within available funding levels.

Background

Policy Directive PD 2005_335 (previously Circular 2003/86) *Charging Policy for Clinically Required Specialised Genetics Tests which are non-Medicare Benefits Schedule Items*, http://www.health.nsw.gov.au/policies/PD/2005/pdf/PD2005_335.pdf states that:

> *"Area Health Services are to meet the cost of testing from within their global budget allocation for clinically/medically required specialised genetic testing for non-Medicare Benefits Schedule items for:*
> - *admitted public patients*
> - *non-admitted public patients, and,*
> - *privately referred non-inpatients referred to a public sector specialist clinic*
>
> *Except where indicated, arrangements are to be consistent with "Principles for Funding of NSW Public Health Sector Pathology Services" PD 2005_533 (previously Circular 2001/113)*
> *http://www.health.nsw.gov.au/policies/pd/2005/pdf/PD2005_533.pdf*
>
> ***Specialised tests for genetic disorders*** *refers to tests which are non Medicare Benefits Schedule items performed by public hospital laboratories and funded by the NSW Health System. The costs of tests are generally in the range of $100 to $2000 per test, and more in rare instances. These tests are used to:*
> - *diagnose a genetic disorder, including a prenatal diagnosis*
> - *determine if a person is a mutation carrier for a disorder, or*
> - *detect an inherited predisposition to a genetic disorder.*
>
> *Local arrangements are to be negotiated concerning clinical responsibility for authorising testing as well as budget responsibilities for approving test requests. This would most appropriately rest with the head of a clinical genetics unit or delegated staff member. Referral to public sector genetics services will provide the patient with clinical geneticist expertise not generally available in the private sector. It will not guarantee testing, as it will need to be assessed and prioritised according to clinical necessity."*

Guidelines for prioritising genetic tests

To assist health services/clinicians prioritise genetic tests within available funding levels, the Genetic Services Advisory Committee (GSAC), NSW Department of Health, in association with Heads of Clinical Genetics Units has developed a priority system as a guide to appropriate genetic testing based on clinical need and equitable access. The charging of genetic tests is to be in accordance with the policy outlined in the above-mentioned policy directives.

High Priority

1. Prenatal Testing

- Where the confirmation of a clinical diagnosis by molecular testing will assist parents who may use the information in making reproductive choices.
- Where the confirmation of the clinical diagnosis will enable treatment options to be instituted which might be early in the newborn period.
- Where gonadal mosaicism is recognised to occur frequently (eg Osteogenesis Imperfecta with a risk of 3 – 4%)

2. Diagnostic Testing

- When confirmation of a clinical diagnosis will restore reproductive confidence in the family.
- When confirmation of a clinical diagnosis will lead to changes in management of an affected person.
- Where a diagnostic test can lead to predictive testing of other at-risk family members.
- To confirm a clinical diagnosis where it is relevant to screening for disease complications.
- To confirm a clinical diagnosis where it is relevant for funding purposes eg extra aid at school.

3. Carrier Testing

- Where the patient has had genetic counselling and is aware of a high likelihood of being a carrier based on family history or ethnicity and the patient has accepted the advantages and limitations of carrier testing.
- When there are prenatal diagnosis implications for a family because of a known family history.
- Where one partner is a known carrier of a recessive condition and carrier testing of the other partner may lead to the possibility of prenatal diagnosis and accurate reproductive counselling.

4. Presymptomatic and Predictive Testing

- Where there is a known family history of a disorder and mutation is known.
- Where there is definitive testing available and there is a family history of the disorder ie Huntington disease.

Low Priority

1. Prenatal Testing

- Where confirmation of a clinical diagnosis by molecular testing will not alter the reproductive choices or obstetric or perinatal care for the patient.
- Where there is only a low theoretical risk of gonadal mosaicism.
- Where there is a recessive condition and there is no need for carrier testing for the new partner who is at a low risk of being a carrier.

2. Diagnostic Testing

- Where the clinical diagnosis is confirmed by other means and genetic testing will not alter the patient's management or options.
- Where the test has been requested by the parents or health professionals and the geneticist thinks a diagnosis is unlikely or the test is not clinically indicated.
- Where confirmation of the clinical diagnosis by genetic testing will not influence whether prenatal testing is undertaken and or the type of test.
- Where the genetic test will not lead to confirmation or predictive testing of other family members eg no at risk relatives.
- Where confirmation of the clinical diagnosis will not alter screening of potential disease complications.

3. Carrier Testing

- Where the disorder is rare and there is no family history.
- Where the testing will not alter the lifestyle or health options for a person.

4. Presymptomatic and Predictive Testing

- Where there is no family history of the disorder.
- Where the only people to have predictive testing would be children for adult onset disorders.

Robert D McGregor AM
A/Director-General