Department of Health

Rheynn Slaynt

Clinical Recommendations Committee

Recommendation 03/13

Genetic tests approved by the United Kingdom Genetic Testing Network and listed on the NHS Directory of Molecular Genetic Testing are considered as High Priority by the Isle of Man Department of Health. All referrals for genetic testing must meet the Isle of Man genetic testing criteria and follow the genetic testing pathway. http://ukgtn.nhs.uk/

Approved by the Minister on 27 August 2013

Clinical Background Information

Rapid developments in medical science have facilitated an expanding repertoire of tests for disorders likely to have an underlying genetic basis.

New technologies provide an enhanced understanding of genetic disorders and the complex biological pathways underpinning many common diseases. These advances have generated increased expectations and demands amongst patients, relatives and the general public. There is a need to ensure that patients, often with complex clinical needs, and their families have equitable access to the advantages resulting from progress in genetic science.

The UK Genetic Testing Network (UKGTN) commissioned the Foundation for Genomics and Population Health (PHG Foundation) to write a discussion paper on the impact of genetic testing for mainstream specialties in order to consult with a variety of organisations. The UKGTN consultation took place in February 2010. The report stimulated much debate within the 'genetics' community resulting in a broad endorsement of integrated working between medical specialties and clinical genetic services. The need to embed genetics into mainstream services, together with the widespread use of genetic tests, was recognised to be consistent with the recommendations of the House of Lords report into Genomic Medicine.

The views expressed to the UKGTN were:

- There is a general consensus in favour of the continued existence of clinical genetics as a specialist commissioned service
- There is a need for the further development of pathways for testing involving primary, secondary and tertiary care
- There is support for the use of genetic tests by relevant specialities as part of agreed clinical and diagnostic pathways
- There is support for the strengthening of existing good models of care (e.g. specialty clinical networks, joint and multidisciplinary clinics)

UKGTN Approved Testing

The UKGTN assesses whether providing a test as an NHS service is likely to be of benefit to patients. Approved tests have been evaluated for clinical validity and utility.

All approved tests are listed in the NHS Directory of Molecular Genetic Testing.
Not all requests for new tests are accepted by UKGTN. There can be a variety of reasons for this, including: unclear scientific understanding; the test is still in a research phase; or the test does not add anything new to the diagnosis or management of the patient.

The Gene Dossier Process

The Gene Dossier process was developed by the UKGTN in 2004 as a tool to evaluate whether a proposed laboratory genetic test for a specific genetic disease is to be recommended for inclusion on the NHS Directory of Genetic Testing. Once a test is accepted for inclusion in the Directory it is recommended to be considered for funding under local specialised commissioning arrangements. The Directory lists tests for diseases, which have been agreed as appropriate for clinical use. The tests actually provided and the laboratories providing them are available from the online database on the UKGTN website. The purpose of the Directory is to allow equity of access to genetic testing across the UK NHS. The process ensures that the decision regarding the recommendation of a test is explicit, transparent and based on evidence.

A gene dossier must be submitted to the UKGTN for any new genetic test that a UKGTN laboratory member wishes to provide and to have listed on the NHS Directory of Genetic Testing. For gene dossier purposes, a genetic test is defined as any test for NHS service provision by a UKGTN member laboratory which will require funding by specialised commissioning arrangements as supporting provision of clinical genetics services as defined in the Specialised Services National Definition Set (SSNDS) for Medical Genetics.

Those diseases for which testing has been recommended by the working group and endorsed by the UKGTN Steering group are submitted to NHS commissioners with the recommendation that they should be considered for funding as an NHS service

The diagnostic care pathway can be optimised by the use of new genetic tests, often reducing the need for other conventional diagnostic methods, improving the patient experience and providing efficiencies to the NHS.

GENE DOSSIER EVALUATION FRAMEWORK

The gene dossiers evaluation framework is based on the ACCE framework and includes the following details:

1. seriousness of the condition
2. prevalence of the condition
3. purpose of the test – diagnosis treatment, prognosis and management, pre-symptomatic testing, risk assessment
4. technical details of the test
5. context in which the test is to be used - defined population groups
6. characteristics of the test – the clinical sensitivity, specificity and predictive value
7. clinical utility of the test – how it adds to patient management and the availability of alternative diagnostic procedures

8. ethical, legal and social considerations
9. price of the test
10. multidisciplinary and transparent (gene dossiers/testing criteria available on website)

Quality, Innovation, Productivity and Prevention (QIPP) Quality

UKGTN provides a trustworthy source of information for patients and the public.

UKGTN aims to promote equity of access to genetic testing to NHS patients

In addition to the molecular laboratories in the Regional Genetics Centres and the specialist laboratories that became members in 2003, all the cytogenetic laboratories that are part of a Regional Genetics Centre are now UKGTN members. All member laboratories meet accreditation standards

The UKGTN website is a valuable resource for the public, healthcare professionals and laboratory scientists wishing to gain access to genetic testing information available in the NHS

Innovation

The available evidence clearly shows that array CGH testing achieves an increased diagnostic yield compared with standard karyotyping and UKGTN recommends that it should be funded as a first-line cytogenetic test for learning disability or developmental delay for postnatal referrals throughout the NHS as soon as possible.

UKGTN and CMGS collaborate to develop a standardised national measure of workload that is transparent, flexible and easy to use: Molecular Units (MolUs)

UKGTN will work in synergy with NICE regarding genetic developments.

UKGTN works together in close collaboration with relevant clinicians and scientists to incorporate new technologies into the Gene Dossier process.

Productivity

The diagnostic care pathway can be optimised by the use of new genetic tests often reducing the need for other conventional diagnostic methods, thus improving the patient experience and providing efficiencies to the NHS

UKGTN monitors activity and costs through the Gene Dossier process

UKGTN is linking with various organisations to improve coding and classification, IT systems and the communication of test information for genetic conditions

The work undertaken by the UKGTN to monitor and develop the principles for commissioning a clinical genetic service has been acknowledged by the House of Lords.

The UKGTN and NGEDC are collaborating on a project to encourage the appropriate use of genetic testing among non-genetic specialists.

Prevention

UKGTN Testing Criteria provides guidance to professionals for appropriate testing which can prevent patients having other unnecessary diagnostic tests and procedures and can avoid admissions to hospital.

National Tariffs for Genetics

How is Pricing determined?

In England the Payment by Results system for funding NHS healthcare is based on tariffs that are nationally set with NHS Trust Market Forces Factor applied separately. The Department of Health Payment by Results team calculate national tariffs based on data collected from NHS Trusts.

The activity within a hospital has to be coded in order for the collection of data to be standardised. The organisation responsible for coding activity in the NHS is the Health and Social Care Information Centre. National tariffs have not currently been calculated for all NHS services including genetics. Therefore where national tariffs do not apply local prices are used. Different systems of funding apply in Scotland, Wales, Northern Ireland and the Isle of Man.

UKGTN National Tariff Exercise

The UKGTN was asked to carry out a piece of work to determine a methodology to derive tariffs for molecular genetic testing in order to inform the Department of Health Payment by Results team. The exercise was completed in 2006.

The report details the methodologies used to calculate testing tariffs and the data collected from the laboratories that took part in the exercise. Tests were categorised into simple, medium and complex and tariffs calculated for each category based on expenditure and activity in the baseline year 2004/05. From this initial work the report recommended that two further test categories should be included, 1) very simple and 2) highly complex.

An informal consultation with UKGTN membership was conducted in 2007. All those who responded felt it was important that UKGTN continues to be involved in developing national tariffs for genetics and in working collaboratively to develop a consistent model of commissioning. The UKGTN continues to work with the NSCG, the HSCIC and the Department of Health in the development of genetic national tariffs.

Genetic Testing on the Isle of Man

- The range of diagnostic genetic testing is funded from two areas of the Isle of Man Department of Health:
    - By Noble's Hospital Pathology Department who pay for (but are not funded for these tests) tests requested by on Island doctors – GPs and Consultants.
    - From the contract with the North West Specialised Commissioning Team (NWSCT) – this pays for patients who are referred to the consultant geneticist at LWH and are under their care, and is part of the package of care for which they have been referred.

The majority of samples taken at Noble's are sent to Liverpool Women's Hospital, with some more unusual tests being sent to other specialist laboratories in the UK.

Prior to 2010-11 Liverpool Women's hospital had not charged the IOM for carrying out tests. In 2010-11 they started invoicing for the service.

The service can be broken down into two areas:

- Tests on patients who have been referred to a LWH consultant and the responsibility for care rests with the LWH consultant. These tests and OP appointments are

invoiced to the NWSCT under our contract for Tertiary care and come from the Off Island treatment budget.

- Tests requested by doctors (GPs and Noble's Consultants) on the IOM – patient remaining under the care of the IOM doctor. These samples are all sent via the Pathology Laboratory at Noble's Hospital and (although payment for these tests is met from the pathology budget there is no additional funding for these tests) the funding comes from the Pathology Laboratory budget. Whilst previously funded under a block contract, these are now charged on a cost-per-test basis.

These costs came as an additional pressure on both these budgets.

Breakdown of referrals to Liverpool

Source of Referrals for Outpatients to Liverpool Women's 01/04/2010 to 28/02/2011, Genetics, Isle of Man Patients

	Referrer	Number
	GENERAL MEDICAL PRACTITIONER	22
	CONSULTANT, other than in an Accident and Emergency Department	48
	Self-referral	6
	Other - initiated by the CONSULTANT responsible for the Consultant Out-Patient Episode	1
	Other - not initiated by the CONSULTANT responsible for the Consultant Out-Patient Episode	4
		81

The data in the above table comes from the Liverpool Genetic Testing Centre. From this source it appears that over 25% of off Island referrals for Genetic tests have been made by a GP.

Breakdown of tests taken at Noble's and sent away for testing.

Approximately 10% of tests taken on Island are requested by GPs.

Source of Referrals for Genetic tests to the Pathology department, Noble's Hospital - March 2011 – July 2011 (5 months)

	Referrer	Number
	GENERAL MEDICAL PRACTITIONER	3
	CONSULTANT, other than in an Accident and Emergency Department	9
	Self-referral	0
	Other - initiated by the CONSULTANT responsible for the Consultant Out-Patient Episode	3
	Other - not initiated by the CONSULTANT responsible for the Consultant Out-Patient Episode	3
		18

Problems with Genetic Testing Referrals on the Isle of Man

A number of avoidable problems have arisen from referral requests made by Isle of Man referrers, leading to rejection of the referral by the testing unit and/or a requirement for retesting. In particular:

- Referrals have not included all the required information.
- The wrong samples have been taken.
- Samples, which are time sensitive, have been taken at the wrong time and have deteriorated before they were tested.
- Referrals have been made to the wrong testing unit.

Criteria/Pathway

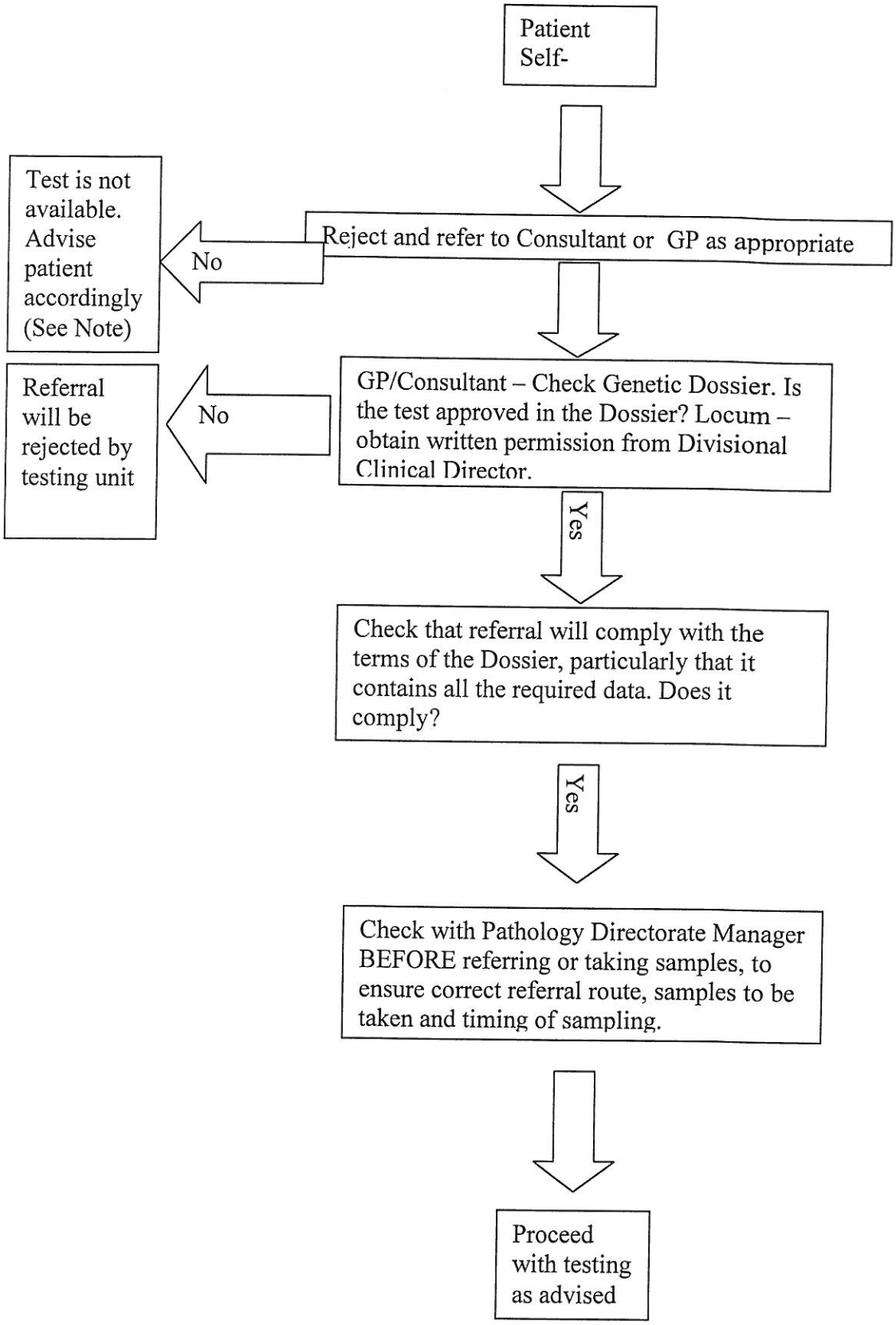
Recommendations:

- Only tests that have been approved by the UKGTN will be funded by the IOM Health Service – that is only tests that have a Gene Dossier and are coded Blue or Green on the UKGTN List.
- Because it undergoes regular revision, referring doctors must check the UKGTN list before making a referral.
- Referrals for Genetic testing must comply with the terms of the Gene Dossier and, in particular must contain all the required data as, otherwise the receiving genetic testing unit will not accept the referral.
- Referring doctors must check with the Pathology Directorate Manager before referring or taking samples. This is so that they can be advised on the correct referral route and, most importantly, which samples should be taken and when they should be taken.
- Locum Consultants can request genetic tests following written permission from the Division's clinical director.
- The Isle of Man will not fund patients who self refer.
- Any request for a test that is not on the UKGTN approved list or from a clinician who does not fulfil the referring criteria **will not** normally be tested or funded.
- Any request for funding outside this policy must be presented to the CRC Exceptions Committee in advance.

If approved, this policy must be communicated to all GPs and Consultants on the Island.

The Genetic Testing Centre at Liverpool Women's Hospital **MUST** be advised of the above rules so that they know who they can accept referrals from, and under what circumstances.

ISLE OF MAN GENETIC TESTING POLICY FLOWCHART



Note: Any request for funding outside this policy must be presented to the Clinical Recommendations Committee Exceptional and individual Funding Request Panel (EIFRP) in advance.

Evidence (NICE/SIGN)

UKGTN collaboration with NICE

The UKGTN has developed formal links with the diagnostic team at the National Institute for Clinical Health and Excellence (NICE). This is to promote joint working in areas of mutual interest, and regular meetings have occurred. To date areas under discussion are 'array CGH' (a technique which detects very small genetic variations) and its application to all conditions related to developmental delay, a pre-assessment of arrays in particular for cancer and companion diagnostics when evaluating pharmacogenetic developments. The NICE diagnostic team has also confirmed that UKGTN should continue with the evaluation of new tests for rare genetic conditions through the gene dossier process. As these tests affect small numbers they would not meet the criteria for selection for NICE assessment.

UKGTN will work in synergy with NICE regarding genetic developments.

Financial Resources

This recommendation was approved by the Minister to be funded from **within** the current Department of Health budget allocation.

Acknowledgements

Prepared by Mr Norman McGregor Edwards, Director of Health Care Delivery for the CRC, in conjunction with Mr Tony McMaster, Pathology Directorate Manager, Noble's Hospital, Mrs Erica Kermode, Tertiary Care Manager, Noble's Hospital, Mrs Sue Rafferty, Medical Records Manager, Noble's Hospital.

References

"Supporting Genetic Testing in the NHS" (second report of the UKGTN) November 2010