

Guidelines for the development of genetic testing in the NHS

a. Key Regulations

Human Genetics
Committee (HGC)
12th September 2007

Recording of 29th HGC plenary meeting, Durham, 12/09/07

www.hgc.gov.uk/Client/Content.asp?ContentId=789

Addresses HGC's response to the Government's proposal for a Single Equality Bill. The Commission also discussed the oversight of private genetic tests, and the provision of genetic tests on the NHS.

Organisation for
Economic Co-
Operation and
Development
(OECD)
2007

Guidelines-Quality Assurance in Molecular Genetic Testing

www.oecd.org/dataoecd/43/6/38839788.pdf

The Guidelines address genetic testing for variations in germ line DNA sequences or products arising directly from changes in heritable genomic sequences that predict effects on the health, or influence the health management, of an individual.

World Health
Organisation (WHO)
2004

25 recommendations on the ethical, legal and social implications of genetic testing

ec.europa.eu/research/conferences/2004/genetic/pdf/recommendations_en.pdf

The recommendations presented in this document are focused on issues that must be addressed when implementing genetic testing in healthcare systems and using genetic testing as a research tool. Topics covered include: population screening, genetic counselling, issues related to gender and ethnicity, the use of "biobanks," and the collection of biological material. An Expert Group was invited by the European Commission and existed of representatives from industry, NGOs, scientists and representatives of different specialities: law, philosophy, ethics and medicine. The recommendations address three key areas: General framework; Implementation of genetic testing in healthcare systems; and Genetic testing as a research tool.

Clinical Molecular
Genetics Society
(CMGS)
*To be ratified.
Publication
anticipated early
2008.*

Practice guidelines for the Interpretation and Reporting of Unclassified Variants (UVs) in Clinical Molecular Genetics.

Increased demand for molecular genetic testing has caused a marked change in the scale and sensitivity of molecular genetic analysis within the service environment. This has led to a rapid increase in the detection of novel sequence variations of unknown pathogenicity. As routine diagnostic service labs have limited time- and budget resources, a set of agreed standards are essential in the determination of the clinical significance of variants identified in routine testing. The standards outlined in this document have been drawn up as a guide to assess variants of unknown clinical significance for situations where there is likely to be a clinical benefit.

Genetics
Commissioning
Advisory Group
(GenCAG)
25th May 2005

Terms of reference

www.dh.gov.uk/en/Policyandguidance/Healthandsocialcaretopics/Genetics/Geneticsgeneralinformation/DH_4117687

The Genetics Commissioning Advisory Group was set up to take a strategic national overview of genetics in healthcare delivery. It aims to provide advice to commissioners of genetics services to enable them to provide appropriate services for NHS patients and their families. Members are drawn from relevant professional bodies and groups and Royal Colleges, an umbrella patient support group (GIG) and from the specialized commissioning groups which commission these services in the NHS.

EuroGentest
4th November 2007

Definitions of Genetic Testing – 3rd Draft

EuroGentest is an EU-funded Network of Excellence (NoE) with 5 Units looking at all aspects of genetic testing – Quality Management, Information Databases, Public Health, New Technologies and Education. Through a series of initiatives EuroGentest encourages the harmonization of standards and practice in all these areas throughout the EU and beyond. This document intends to present different definitions of Genetic testing (or related definitions) and to identify trends, differences and ideas presented by national, international, health, patient and genetics associations.

EuroGentest
1997-2006

Best Practice Guidelines for Molecular Genetic Testing

www.eurogentest.org/web/info/public/unit1/eqa/molecular_genetics/guidelines.xhtml

Collection of international guidelines on Charcot-Marie-Tooth Disease, Cystic Fibrosis, Cystic fibrosis Carrier Screening, Faktor V Leiden, Fragile X Syndrome, Friedreich Ataxia, Haemoglobinopathies, Haemophilia A, Haemophilia B, Haemoschromatosis, Hereditary Breast and Ovarian Cancer, Hereditary Motor and Sensory Neuropathies, Hereditary Nonpolyposis Colorectal Cancer, Huntington Disease, Mitochondrial DNA Disease, Muscular Dystrophy Duchenn/Becker Type, Prader-Willi Syndrome/Angelman Syndrome, Retinoblastoma, Spinal Muscular Atrophy and Y Chromosome Microdeletions.

Institute for
Prospective
Technological
Studies (IPTS)

The IPTS is one of the seven scientific institutes of the European Commission's Joint Research Centre (JRC). The work undertaken by IPTS is mainly done at the request of other Directorates General of the European Commission. The IPTS Life Sciences Section programme performs evaluation of the consequences, opportunities and challenges of modern biotechnology for Europe, in terms of economic, social and environmental aspects. This is complemented by assessment of new bio-medical technologies such as tissue engineering, pharmacogenetics or genetic testing, and specially their deployment across Europe, their quality standards and the optimal use of bio-banks as they condition the progress in European biomedical science.

Joint Committee on Medical Genetics
April 2006

Consent and confidentiality in genetic practice

This report from the JCMG is a response to requests from genetics professionals for clarification of issues of consent and confidentiality in clinical practice, particularly with regard to the requirements of the Data Protection Act 1998 and the Human Tissue Act 2004. Although the report specifically addresses clinical practice in the genetics services, its discussions and recommendations are believed to be of value for all specialties.

As well as clarifying issues surrounding consent and confidentiality in genetic practice, the report identifies key practice points and lists documents which may be helpful when seeking consent.

On some issues the report makes recommendations for practice.

The report covers:

- general aspects of consent as applied to genetics
- the sharing of information with other family members and between professionals
- genetic investigations performed on stored material
- the Human Tissue Act 2004, consent and DNA analysis
- the Data Protection Act 1998 and the processing of medical genetic information.

UK Genetic Testing Network (UKGTN)
15th August 2005

Testing criteria Molecular Genetics Tests
www.ukgtn.nhs.uk/gtn/UKGTN-information/dossier/dossier.html

The UKGTN is a collaborative group of laboratories and their clinicians, commissioners and patient representatives who adhere to quality criteria and standards, working within clinical governance arrangements. The UKGTN is involved in the provision of genetic tests for inherited disorders in order to promote access to high quality molecular genetic testing services for patients across the UK.

b. Policy and Review

Department of Health 24 th June 2003	Our inheritance, our future: realising the potential of genetics in the NHS www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_4006538
The aim of this White Paper is to set out a vision of how patients could benefit in future from advances in genetics, and to raise awareness of the potential of genetics in healthcare; includes a £50m three-year plan of implementation.	
DH January 2007	Commitments in the Genetics White Paper: progress to date www.dh.gov.uk/en/Policyandguidance/Healthandsocialcaretopics/Genetics/Geneticsgeneralinformation/DH_4138863
This document sets out progress against each commitment made in the 2003 Genetics White Paper "Our inheritance, our future – realising the potential of genetics in the NHS".	
Department of Health 9 th February 2007	3-year progress review of the genetics White Paper www.dh.gov.uk/en/Policyandguidance/Healthandsocialcaretopics/Genetics/Geneticsgeneralinformation/DH_4138863
EuroGentest 4 th November 2007, 3 rd draft	Definitions of Genetic Testing en.eurogentest.org/files/public/unit3/DefinitionsGeneticTesting-3rdDraf18Jan07.pdf
The document iwritten by a European Commission Expert Group (25 Recommendations on the Ethical, Legal and Social Implications of Genetic Testing, 2004) establishes the need to define explicitly what is meant by <i>genetic testing</i> , whenever the term is used, and the need to develop a consensus definition that is globally applicable. It suggests that a definition should be developed globally by several public and private bodies involved (including WHO, OECD, EC, IFHGS and ICH) and states that the EC should take initiative.	
The 2003 Genetics White Paper committed to a review after 3 years to see what additional action was needed to improve the NHS genetic services.	
NHS Executive and the Human Genetics Commission 2002	Laboratory Services for Genetics www.phgfoundation.org/policydb/4010/
Evaluation of NHS laboratory genetic services, identification of future developments and their potential implications.	

c. Related Programmes and Pathways

Database of Chromosomal Imbalance and Phenotype in Humans using Ensembl Resources (DECIPHER)	www.sanger.ac.uk/PostGenomics/decipher/
--	--

Array-CHG allows detection of submicroscopic chromosomal imbalance across the entire genome. CHG is used for diagnosis of developmental delay/learning disability and/or multiple congenital anomalies which can not be identified by routine karyotyping. DECIPHER allows for visualization of chromosomal locations analysed by CGH, genes and expressed-sequence tags, identify syndromes, record and compare phenotypes

Diagnostic Mutation Database (DMuDB)	ngri.man.ac.uk/
--------------------------------------	--

The DMuDB provides a resource for high quality (rare) mutation data from UK diagnostic laboratories, sharing of mutation data between UK diagnostic laboratories to support genetic testing services for patients.

ENSEMBL	www.ensembl.org/index.html
---------	--

The European Molecular Genetics Quality Network (EMQN)	http://www.emqn.org/emqn/index.html
--	---

EMQN is a not-for-profit organisation promoting quality in molecular genetic testing through the provision of external quality assessment (proficiency testing schemes) and the organisation of best practice meetings and publication of guidelines.

Ensembl is a joint project between EMBL-EBI and the Sanger Institute and constitutes a database producing and maintaining automatic annotation on selected eukaryotic genomes.

Gene Dossier	www.ukgtn.nhs.uk/gtn/UKGTN-information/dossier/dossier.html
--------------	--

The Gene Dossier Evaluation Process is a specialist commissioner route designed by the UKGTN to integrate new genetic test in the NHS Directory of Molecular Genetic Testing. *Submissions are made to the UKGTN by UKGTN laboratories for tests funded through the Specialist Commissioner route (meeting assessment criteria and adopted by GenCAG/National Specialised Commissioning Advisory Group).*

Genotype-To-Phenotype: A Holistic Solution (GEN2PHEN)	<i>To commence early 2008</i>
---	-------------------------------

GEN2PHEN is 60 month collaborative project of 18 bioinformatics groups, part of the EU framework programme 7. The project aims to unify human and model organism genetic variation databases towards increasingly holistic views into genotype-to-phenotype data, and to link this system into other biomedical knowledge sources via genome browser functionality. Part of its remit is to develop key standards for the G2P database field, create database components, services and integration infrastructures, populating G2P databases, building a major G2P internet portal and to undertake a whole-system utility and validation pilot study.

National Institute for Biological Standards and Control (NIBSC)	www.nibsc.ac.uk/
---	--

NIBSC is part of the National Biological Standards Board, responsible for safeguarding and advancing public health by assuring the quality and safety of biologicals. NIBSC provides independent testing of biological medicines for the UK market before release, but may also carry out tests following particular problems after release. *NIBSC collaborates with the National Genetics Reference Laboratory (NGRL) in Manchester to develop and WHO certify reference materials for genetic testing.*

d. Nowgen Events and Comment:

Nowgen Seminar on Future Trends in NHS Genetic Testing, November 15th 2007.

In the years to come, emphasis within Genetics will be on:

- Bioinformatics; applicable to pre-analytical, analytical and post-analytical stages; Informatic Databases
- Genome Sequencing
- Comparative Genome Hybridisation (CGH) Array
- Non-Invasive pre-natal diagnostics/foetal sexing
- Microfluids