



The Royal College of Pathologists
Pathology: the science behind the cure

Code of Practice for Genetics Laboratories

August 2006

Unique document number	G003
Document name	Code of Practice for Genetics Laboratories
Version number	1
Produced by	Dr John Old, on behalf of the Specialist Advisory Committee on Genetics and Clinical Embryology
Date active	14 August 2006
Date for review	14 August 2009
Comments	Part of <i>Codes of practice for pathology services and departments</i> series, commenced May 2005.

This document was initially produced in June 2006 by Dr John Old, on behalf of the Specialty Advisory Committee on Genetics and Clinical Embryology, as part of the *Codes of practice for pathology services and departments* series commenced in May 2005. In accordance with the College's publications policy, it was placed on the Fellows' and Members' area of the College website for consultation from 16 June – 17 July 2006 and 22 people responded. Dr John Old and the Committee considered the feedback and amended the document accordingly. Please email publications@rcpath.org if you wish to see the responses to the feedback received.

Professor Carrock Sewell
Director of Publications

CODE OF PRACTICE FOR GENETICS LABORATORIES

This code of practice is intended as a supplement to the required standards of practice for Clinical Pathology Accreditation (UK) Ltd (CPA), which is now compulsory, and NHS clinical governance through the Commission for Health, Audit and Inspection (CHAI).

Throughout the document, bulleted lists provide references to guidance documents that have already been published by The Royal College of Pathologists and other organisations that give more detailed information. All College documents can be downloaded from the College website: www.rcpath.org/publications

Preface

The Codes of Practice for pathology services draw together, for each specialty, all relevant guidelines and standards issued by The Royal College of Pathologists.

Each Code of Practice comprises a coherent description of what is required for the provision of an effective, reliable and safe pathology service in that specialty. Thus, the codes and the documents cited within them can be used to assist in the appropriate delivery of pathology services and to help assess whether relevant standards for laboratory accreditation are being met.

Professor Adrian Newland

President

June 2006

1 Introduction

This code of practice provides guidance for the application of good laboratory practice in genetics laboratories, and is intended as a supplement to the standards of practice required for laboratory accreditation with Clinical Pathology Accreditation (UK) Ltd (CPA). All genetics laboratories in the UK must now be enrolled with CPA or another body accrediting to equivalent standard.

Clinical genetics laboratories consist of two closely interrelated disciplines: cytogenetics (the study of chromosome aberrations) and molecular genetics (the study of mutations at the molecular level), and several related subspecialties, such as biochemical genetics, immunogenetics and haemoglobinopathies. The disciplines make a particularly significant contribution to the area of preventive medicine through the screening for patients at risk because of genetic factors and the provision of prenatal diagnosis. Many laboratories are either located within an integrated and regionally based genetics centre, or have close links with clinical geneticists as well as other pertinent clinical specialties.

Clinical cytogenetics

Clinical cytogenetics is the diagnostic application of observed changes in chromosome number, structure and function. The main services offered by clinical cytogenetics laboratories are the prenatal diagnosis of chromosomal abnormalities from amniotic fluid, chorionic villi or fetal blood, and postnatal diagnosis from peripheral blood for a variety of referral reasons including congenital malformation, developmental delay, pregnancy loss and infertility. In addition, bone marrow and tumour samples are examined for acquired chromosome abnormalities, which may have diagnostic and prognostic value in the investigation of malignancy. Some laboratories are also using newly developed molecular cytogenetic techniques to detect gene copy number changes and micro-deletion syndromes.

Clinical molecular genetics

Clinical molecular genetics is the diagnostic application of molecular genetic technology. The main services offered by molecular genetics laboratories are the diagnosis or exclusion of single gene disorders, including prenatal diagnosis. The reasons for referral include a high prior risk of genetic disease, developmental delay and the investigation of diseases that may have a genetic aetiology such as some cancers and neurological conditions.

Biochemical (metabolic) genetics

Genetics laboratories also exist that specialise in biochemical (metabolic) genetics. Biochemical (metabolic) genetics is the study of inborn errors of metabolism, usually single gene disorders that are diagnosed by characteristic metabolite profiles or by reduced enzyme activity. The techniques are also used as a basis for prenatal diagnosis. A well developed network of 17 'stakeholder' laboratories exists in the UK.

- *Standards for the Medical Laboratory*. Clinical Pathology Accreditation (UK) Ltd., 2003

2 Head of department

The head of a regional or large sub-regional genetics laboratory is usually an appropriately experienced and qualified consultant grade clinical scientist (Career Pathway Stage 9) or occasionally a medically qualified clinical cytogeneticist or molecular geneticist. Either should normally be members of The Royal College of Pathologists (MRCPATH or FRCPath).

The head of department has overall responsibility for all aspects of the laboratory service, including pre-analytical, analytical and post-analytical decisions, authorisation and interpretation of results, and the provision of diagnostic expertise and clinical advice.

Clinical liaison is an essential element of this role. Management responsibilities include implementation of statutory regulations (e.g. health and safety), quality assurance, policy making, strategy planning, finance and resource control, workforce planning and training and clinical audit, as well as an involvement with business planning and contracting. Research and the development of new techniques are also under the direction of the head of department.

- *The Clinical Scientist in Pathology*. The Royal College of Pathologists, 2005

3 Staffing and workload

The staff structure of a genetics laboratory is a mix of clinical scientists, genetic technologists (formerly known as medical technical officers), biomedical scientists and laboratory support staff, both technical and clerical. Clinical scientists are employed at differing grades reflecting their roles and responsibilities. The senior clinical scientists in charge of the day-to-day running of the laboratory are healthcare scientists at Career Stage 8, principal clinical scientist level. Senior clinical scientists with a management or other specialist responsibility are expected to be members of The Royal College of Pathologists (MRCPATH). Many laboratories also employ trainee clinical scientists, who are unregistered and must be supervised by a registered staff member in all aspects of their work. On completion of training, all clinical scientists must be registered with the Health Professionals Council.

The number of staff and the skill mix within a genetics laboratory should be adequate and appropriate for the workload and repertoire of the laboratory. Genetics is a very fast-moving discipline, with a rapid pace of scientific advance allowing an increasing number of genetic disorders to become amenable to molecular investigation. The balance of staff should take into account such factors as changing referral patterns, the application of new genetic tests and the introduction of new techniques and technology.

- *Medical and Scientific Staffing of National Health Service Pathology Departments*. The Royal College of Pathologists, 1999.

4 Management of services

The role of the head of department has been described above, but is essentially the provision of a service at the highest standards possible within budgetary limitations. In addition to his/her role in departmental administration, the head of department should contribute to planning decisions at the relevant hospital, district and regional level. Consultant clinical scientists in genetic laboratories should ensure that their role is widely recognised. There is also a responsibility to ensure that the wide-reaching nature of these disciplines is recognised and the effects or potential consequences of any service developments or changes are appreciated.

Clinical scientists in genetic laboratories are called upon to develop, manage and support genetic testing networks and other initiatives at regional, national and international level for the optimal delivery of diagnostic services, including the Genetics National Reference Laboratories and UK Genetics Testing Network. Senior clinical scientists in genetics are also often required to work for various professional bodies and specialist societies, including The Royal College of Pathologists, Joint Committee on Medical Genetics, external quality assurance bodies (e.g. CPA), external quality assessment (e.g. National External Quality Assessment Service) and professional bodies (e.g. Association of Clinical Cytogeneticists [ACC], Clinical Molecular Genetics Society [CMGS] and British Society for Human Genetics). This should be recognised as a legitimate part of the clinical scientist's work and be practised to the same high professional standards as their clinical and laboratory roles. This should be recognised in the clinical scientist's job description and allowed for, where appropriate, in the planning of professional activity.

5 General procedures

The head of a genetics laboratory must ensure that users of the service are advised on the appropriateness, limitations and availability of the test required and that the requirements for handling and transporting the sample are met. Standard operating procedures should be in place for all aspects of the laboratory work (pre-analytical, analytical and post-analytical) and these should comply with those standards defined by the CPA.

Diagnostic procedures for genetic disorders should, wherever possible, follow the best-practice guidelines published by the ACC, CMGS, the European Molecular Genetics Quality Network or MetBioNet. Details can be obtained from the websites of these organisations.

- Association of Clinical Cytogeneticists: www.cytogenetics.org.uk
- Clinical Molecular Genetics Society: www.cmgs.org
- European Molecular Genetics Quality Network: www.emqn.org
- Metabolic Biochemistry Network: www.metbio.net

6 Pre-analytical procedures

Genetics laboratories must have written procedures for each stage of the pre-analytical process. Laboratories should follow best practice guidelines for ensuring patient consent and maintaining patient confidentiality. Upon receipt, specimens must be matched with requests and a unique accession number assigned to both specimen and request. There should be clear criteria for establishing adequate patient identification data and a policy for handling incorrectly labelled specimens. Staff should be trained in the safety aspects of handling biological specimens and the use of equipment.

Specimens should be stored in the appropriate conditions for the investigations required. Conditions and legal requirements and restrictions for the storage of tissues will be subject to the Human Tissue Act 2004. All laboratory personnel must therefore be fully informed of the appropriate guidelines and licensing requirements as published by the Human Tissue Authority (2006).

- *Interim Guidelines on the Use of Clinical Samples Retained in the Pathology Laboratory*. The Royal College of Pathologists, 2005.
- Human Tissue Authority: www.hta.gov.uk
- *Consent and confidentiality in genetic practice. Guidance on genetic testing and sharing information*. A report of the Joint Committee on Medical Genetics. Royal college of Physicians, Royal College of pathologists, British Society for Human Genetics 2006.

7 Analytical procedures

Clinical scientists are responsible for determining the repertoire of diagnostic procedures provided by the department. Although other healthcare scientists may perform some elements of a diagnostic test, registered clinical scientists are responsible for the final outcome of any diagnostic test, the interpretation of the results and the provision of appropriate clinical advice. All diagnostic tests must have a written procedure that is validated and reviewed at regular intervals.

8 Post-analytical procedures

Clinical scientists are responsible for ensuring the results are reported in a timely manner, with the inclusion of relevant interpretative comments and clinical liaison as appropriate. Only appropriately qualified and trained clinical cytogeneticists and molecular geneticists may interpret results and authorise reports unsupervised. Guidelines for target reporting times for cytogenetic tests have been established over a number of years. They are reviewed periodically by a Professional Standards Committee and are informed by recommendations by other bodies (e.g. the Screening Committee). Guidelines for target reporting times of molecular genetics tests by 2006 are published in the genetics White Paper, *Our Inheritance, Our Future*. These guidelines have been defined in further detail by the Clinical Molecular Genetics Society.

- *Our Inheritance, Our Future*. Department of Health, 2003
- Clinical Molecular Genetics Society: www.cmgs.org

9 Disposal of specimens and records

Protocols and policies for sharing genetic information, access to samples for family studies, and the retention and disposal of specimens and records should be available and should conform to the latest national guidelines from The Royal College of Pathologists, the Joint Committee on Medical Genetics, the Department of Health and the Human Tissues Act. 2004.

- *The Retention and Storage of Pathological Records and Archives (3rd edition)*. The Royal College of Pathologists, 2005
- *Consent and confidentiality in genetic practice. Guidance on genetic testing and sharing information*. A report of the Joint Committee on Medical Genetics. Royal college of Physicians, Royal College of pathologists, British Society for Human Genetics 2006.
- Human Tissue Authority: www.hta.gov.uk

10 Safety

Safety in the genetics laboratory is the responsibility of the head of department, in association with the appropriate senior staff and the designated safety officer. They should make periodic inspections and audits of the department to ensure that relevant safety procedures are being observed. Each department should comply with regulations of the Health and Safety Executive. Adequate facilities must be provided for the safe handling of samples from all patients, with the necessary additional facilities available for handling specimens from patients with infections of greater hazard.

- *Safe Working and the Prevention of Infection in Clinical Laboratories and Similar Facilities.* Health and Safety Executive, 2003
- *Revised Advice on Laboratory Containment Measures for Work with Tissue Samples in Clinical Cytogenetics laboratories.* Department of Health, 2001.
- *Prevention of Infection with Cytomegalovirus, Parovirus B19, New Variant CJD and Mycobacterium Tuberculosis in Cytogenetics Laboratories.* Association of Clinical Cytogeneticists, 2002.

11 Relationship with clinical colleagues and users of the service

The interpretative role and clinical liaison is essential. Clinical scientists in genetics laboratories advise colleagues from diverse specialties and should possess the skills necessary to communicate effectively with a wide range of healthcare staff and other associated professionals. However, they should not work outside their area of expertise. This guidance applies to any non-NHS work as well as to their daily practice within the NHS.

The needs and requirements of patients and users are central to determining the service repertoire and specifications of the laboratory. Clinical scientists must assess the needs of users by attending appropriate clinical meetings and by listening and giving advice to clinical colleagues. Clinical audit is an essential element in ensuring that the service is fit for purpose and being used optimally.

- *Substandard Performance in Pathology: Guidance for NHS Trusts and pathologists.* The Royal College of Pathologists, 2004.
- *Standards of Conduct, Performance and Ethics.* Health Professions Council, 2003

12 Quality assurance

Quality control is a key element of the CPA standards. The head of department should appoint a quality manager to monitor quality standards on a day-to-day basis. The quality manager may function in more than one discipline or department. Each department must have up-to-date documentation detailing all laboratory procedures and a mechanism for document control as part of their quality management system. The head of the laboratory is responsible for the maintenance of quality in all aspects of the work carried out in the department.

Internal quality control requires adherence to standards defined in the operating procedures of the laboratory. Written protocols should be clear and unambiguous. There should be a mechanism to ensure the clinical relevance of the investigations performed, the accuracy of results and the reliability of interpretative reports. There should be regular review. Internal quality control includes ensuring that the premises, equipment, reagents and consumables used for testing are 'fit for purpose' and that staff are suitably qualified and trained.

External quality assurance is validated by an appropriate quality assurance body (e.g. CPA) and is informed by satisfactory participation in a recognised external quality assessment scheme or schemes where they exist, results from which should be regularly evaluated and acted upon when necessary.

There are National External Quality Assessment Services (UKNEQAS) for clinical cytogenetics, clinical molecular genetics and Factor V Leiden genetic testing, and pilot schemes for the DNA diagnosis of haemophilia and haemoglobinopathies. There are also a number of schemes for molecular genetics offered by the EMQN and other providers, eg for DNA sequencing and DNA quantitation. The best-practice guidelines published by the professional bodies and EMQN form the basis for the marking criteria used in these schemes.

13 Clinical audit

Clinical scientists in genetic laboratories should participate in clinical audit activities to assess the quality and appropriateness of the services provided. Audit activity should include some multi-professional and multidisciplinary elements. Departments must also carry out horizontal, vertical and examination or witness audits of their procedures as required by CPA.

14 Research and development

Clinical scientists in genetic laboratories should actively encourage and participate in research. Departments with trainee clinical scientists must ensure that the trainees have time and the opportunity to be involved in research as required by their training programme.

15 Teaching and training

All clinical scientists in genetic laboratories should be willing and able to teach other health service staff and students both within and without the Trust hospital as required. Heads of department should ensure that their trainee staff follow an appropriate course of training and attain previously agreed standards before progressing to non-training posts. The department should ensure that adequate time is allowed for further education and professional training of all grades of staff.

All grades of clinical cytogeneticists and molecular geneticists must be aware of developments in their own discipline and must maintain their competence levels in order to meet the requirements of the Health Profession Council for re-registration. The department should be committed to the development of all staff through continuing professional development schemes and appraisal. All staff should have easy access to relevant literature and the opportunity to attend training courses, seminars and conferences.

Specialty Advisory Committee on Genetics

June 2006