

Learning outcomes in genetics: a continuum of education for medical practitioners

Although the frequency with which health professionals in the NHS will meet patients with genetic disorders will depend on their role and specialty, it is important that at qualification professionals are familiar with core genetic concepts to be able to respond to patients' questions and refer appropriately. Not only do we need to ensure that knowledge available now enhances current clinical management, but that we are prepared for the advances that will come from an increased understanding of the genetic contribution to common disorders and responses to medication.

Education in genetics therefore should not be a static event, but may best be seen as beginning with undergraduate education and continuing in professional development and workplace learning. We have called this the "continuum of genetics education" (see Figure 1).

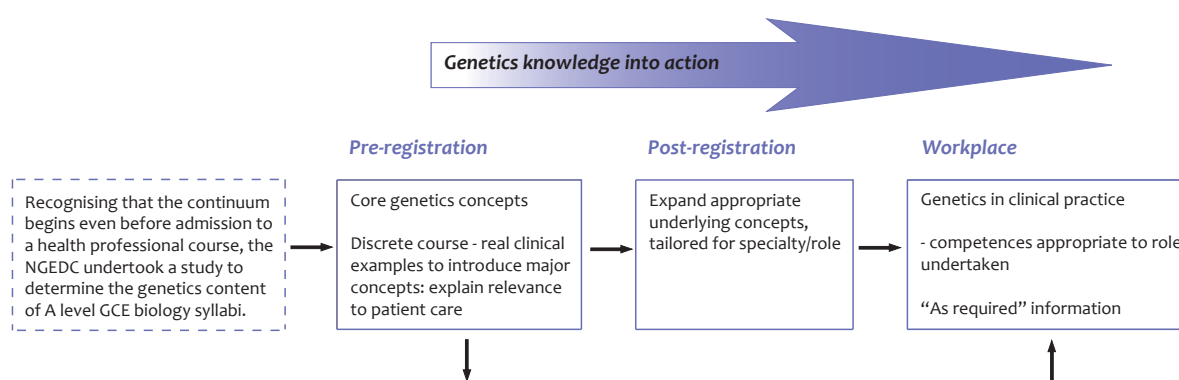


Fig. 1 The continuum of genetics education

Establishing core concepts, learning outcomes and competences

Learning outcomes in genetics for **medical students** and **non-genetic speciality registrars (SpRs)** have been developed by the NHS National Genetics Education and Development Centre (the Centre), building on the results of previous work for medical students and a national Delphi survey of practitioners to identify key genetics knowledge, skills and attitudes appropriate for non-genetics speciality trainees. The Joint Committee on Medical Genetics and medical student teaching leads have endorsed the learning outcomes for medical students; following consultation with specialty education boards and geneticists, there is widespread agreement that the learning outcomes for non-genetics specialist registrars are appropriate for a wide range of specialties.

The development of the GP curriculum statement '**Genetics in Primary Care**' by the Centre, in partnership with the Royal College of General Practitioners (RCGP), was based on a study of educational needs, incorporating the views of practitioners (GP trainers, programme directors and registrars) and specialists (geneticists). This inclusive approach has enabled the identification of learning outcomes which directly reflect clinical practice. Core genetics topics for GPs were identified, prioritised and developed into competency statements in the style of the curriculum structure of the RCGP. The full curriculum statement is available from the RCGP website http://www.rcgp-curriculum.org.uk/rcgp_-_gp_curriculum_documents.aspx

Genetic topics have been included in the **Foundation Programme Curriculum**, stating trainees should: "know risk factors for disease including genetics"; "know genetic susceptibility to adverse drug reactions"; "take a focused family history", and "construct and interpret a family tree when relevant".

The centre has developed, with Skills for Health, a set of **workforce competences** in genetics which follow the patient pathway. They describe in detail how and what activities should be carried out in clinical practice and the underpinning knowledge, skills and attitudes required. They build on the core concepts from pre-registration training and apply them in clinical practice. These competences are complemented by "just in time" information about genetic conditions and their management that health professionals can access through the National Library for Health Specialist Library for Genetic Conditions, for which the Centre is responsible.



Learning outcomes in genetics for medical students

By the end of undergraduate training, the medical student will:

Understand and describe the mechanisms that underpin human inheritance

- Be able to describe the structure, function and replication of DNA as the genetic material
- Be able to describe gene structure, expression and regulation
- Be able to describe the chromosomal basis of inheritance and how alterations in chromosome number or structure may arise during mitosis and meiosis
- Be able to describe the main modes of Mendelian and non-Mendelian inheritance

Have an understanding of the role of genetic factors in health and disease

- Understand how changes to DNA and chromosomes can affect gene function or dosage
- Understand the use of polymorphisms as genetic markers
- Be aware of the role of both genetic and environmental factors in multifactorial conditions such as congenital anomalies, cancer, diabetes and psychiatric illness
- Understand why population ancestry may affect the frequency of Mendelian diseases and of susceptibility alleles

Be able to identify patients with, or at risk of, a genetic condition

- Be able to take a family history and construct and interpret a pedigree
- Understand the clinical implications of the following genetic phenomena: incomplete penetrance, variation in expression, anticipation and new mutations
- Be aware of the possibility of heterogeneity in a genetic disease and the potential impact on diagnosis
- Understand the principles of risk estimation in Mendelian disease
- Be aware of examples of clinical indicators that suggest an inherited predisposition to cancer
- Be able to describe clinical features of common Mendelian diseases
- Be able to describe clinical features of common chromosomal disorders
- Be aware of the types of clinical features which suggest a dysmorphic or malformation syndrome
- Be aware of the roles of genes and teratogens in human congenital anomalies

Be able to communicate genetic information in an understandable, non-directive manner, being aware of the impact genetic information may have on an individual, family and society

- Be familiar with the aims, methods and practice of genetic counselling
- Be aware of the impact of genetic diagnosis on the extended family
- Be able to communicate the concept of risk in a manner that can be understood by a patient
- Be aware of major ethical issues in genetics
- Be aware of the potential uses and misuses of genetic information

Be familiar with the uses and limitations of genetic testing and the differences between testing and screening

- Understand the distinction between genetic screening and genetic testing
- Be aware of the differences and similarities between diagnostic, presymptomatic, carrier and susceptibility genetic testing
- Be aware that 'genetic tests' can include clinical examination, metabolite assays and imaging as well as analysis of nucleic acid
- Be aware of the main laboratory techniques to investigate genetic material and their advantages and limitations
- Be aware of the main methods for sampling genetic material for prenatal diagnosis
- Be able to interpret a standard genetics laboratory report (cytogenetic and molecular genetic)
- Be aware of parameters governing population genetic screening, current population genetic screening programmes and guidelines for the introduction of such programmes

Know how to obtain current information about scientific and clinical applications of genetics, particularly from specialised genetics services

- Recognise situations where it is important to obtain genetic information and advice and be able to describe ways of obtaining these
- Know when and how to make relevant referrals to the specialised genetics services

Learning outcomes in genetics for specialist registrars in non-genetics specialities

By the end of specialist training, the trainee will:

Be able to identify patients with, or at risk of, a genetic condition

- Be able to draw and interpret a family tree
- Be able to recognise basic patterns of inheritance
- Be able to identify single gene disorders in the specialty, incorporating an understanding of the effects of penetrance and variation in expression in autosomal dominant disorders
- Be aware that patients may present with a genetic condition of which there is no family history

Describe the mechanisms that underpin human inheritance and the role of genetic factors in disease

- Be able to describe DNA as genetic material and how mutations and variants contribute to human disease
- Be able to describe the chromosomal basis of inheritance
- Be able to describe the inheritance patterns of single gene disorders in the specialty
- Understand the contribution of genetic and environmental factors in multifactorial inheritance
- Understand the genetic mechanisms that lead to cancer

Appreciate the heterogeneity in genetic diseases and understand the principles of assessing genetic risk

- Be aware of the principles of risk estimates for family members of patients with Mendelian diseases
- Be aware of the principles of recurrence risks for simple chromosome anomalies, e.g. trisomies

Be able to manage genetic aspects of a condition including referring patients to genetic services where appropriate

- Be familiar with national guidelines that influence healthcare provision for those with genetic conditions
- Be aware of one's own professional limits in regard to managing genetic conditions and know when and where to seek advice
- Be aware that, because genetic conditions are often multi-system disorders, comprehensive patient management is likely to involve liaison with other healthcare professionals
- Be aware of support services for those with a genetic condition (e.g. Contact a Family)
- Recognise the need to offer appropriate referral for comprehensive genetic counselling
- Be able to make appropriate referrals to clinical genetics services
- Be familiar with the organisation of genetics services

Be able to obtain and communicate up-to-date information about genetics in an understandable, comprehensible, non-directive way

- Know where to access credible genetic information on-line and off-line for self and patient
- Be aware that consultations involving the giving and discussion of genetics information may require more time
- Appreciate that genetic information impacts not only on the patient but also on their family
- Be able to discuss genetic conditions in a non-directive, non-judgemental manner, being aware that people have different attitudes and beliefs about inheritance
- Be able to discuss treatment/management and reproductive options available to patients/families with, or at risk of, a genetic condition

Be able to use genetic testing appropriately, recognising its uses and limitations

- Understand the distinctions between genetic screening and genetic testing, and the differences and similarities between diagnostic, predictive and carrier genetic testing
- Appreciate that 'genetic tests' can include clinical examination, metabolite assays and imaging as well as analysis of nucleic acid
- Know the clinical indications for ordering genetic tests
- Know how to organise genetic testing, including how to access help via the local clinical genetics service
- Understand the ethical issues involved in genetic testing, such as confidentiality, testing children, and pre-symptomatic testing
- Incorporate the concepts of informed choice and consent into practice

Genetics in Primary Care - Learning Outcomes

The learning outcomes in 'Genetics in Primary Care' (curriculum statement 6 of the RCGP curriculum) reflect the three main themes of genetics in practice: identifying patients with or at risk of a genetic condition; clinical management of genetic conditions, and communicating genetic information.

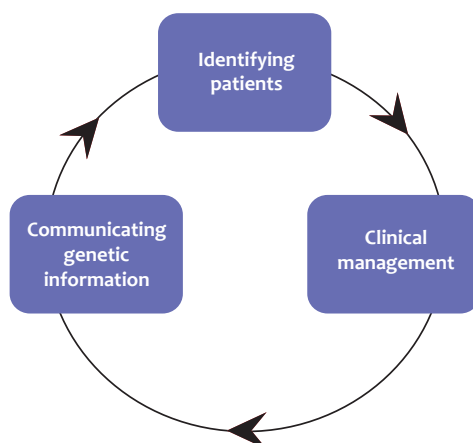


Fig. 2 The three main themes of genetics in practice

By the end of their GP training, the trainee will:

Identifying patients with or at risk of a genetic condition

- Knowledge of common and/or important genetic conditions
- Be able to take and interpret family history information
- Drawing a family tree (pedigree)
- Knowledge of patterns of inheritance
- Understand principles for assessing genetic risk
- Understand DNA as genetic material and how genetic changes contribute to human disease
- Awareness of antenatal and newborn screening programmes

Clinical management of genetic conditions

- Be able to use referral and management guidelines
- Be able to access specialist help and advice from, and refer appropriately to, genetic services
- Be aware of management options (reassurance, managing uncertainty, reproductive options, preventative measures and surveillance)
- Be able to co-ordinate care for people with genetic conditions
- Be able to provide patient centred care, including being aware of patient support services
- Be aware of the different uses of genetic testing, including emotional, ethical, legal and social issues

Communicating genetic information

- Be able to communicate genetic information in an understandable way, helping patients make informed decisions including an appreciation of the non-directive nature of genetic counselling
- Appreciate the emotional, ethical, legal and social impact of genetic information on a patient and their family

Supporting the Learning Outcomes

Resources supporting these learning outcomes and workforce competences, as well as references relating to their development can be found at www.geneticseducation.nhs.uk