

Learning Outcomes in Genetics for Medical Students

Six broad learning outcomes in genetics that medical students should attain by the end of their training are shown below. These have been developed by the NHS National Genetics Education and Development Centre in collaboration with UK medical school genetics leads. They are based on curriculum statements developed in 1990 and 1996 and published in 'Teaching Medical Genetics to Undergraduate Medical Students' (British Society for Human Genetics / Joint Committee on Medical Genetics, 2003).

These broad learning outcomes in genetics for medical students lay the foundations for learning outcomes in genetics in clinical practice for specialist registrars and general practice registrars.

These learning outcomes were endorsed by the Joint Committee on Medical Genetics in October 2006.

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