Six broad learning outcomes in genetics, in the UK which it is proposed that medical students should attain by the end of their training, are shown below. They are in active development by the NHS National Genetics Education and Development Centre, in conjunction with UK medical school genetics leads. They reflect current clinical practice whilst drawing on curriculum statements developed in 1990 and 1996 and published in ‘Teaching Medical Genetics to Undergraduate Medical Students’ (British Society for Human Genetics, 2003). The learning outcomes shown below were in draft form in July 2006 and may have been amended subsequently. Please visit www.geneticseducation.nhs.uk for updates.

These pages have been annotated to indicate the main places in the book where the relevant material can be found. Note that many of the more general clinical and ethical issues emerge from the Case studies, rather than being the subject of a specific single section of the text and so we have just listed the chapters containing the most specific instances.

**Proposed Learning Outcomes**

*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 *(Chapter 3)*
- Be able to describe gene structure, expression and regulation *(Chapter 3)*
- Be able to describe the chromosomal basis of inheritance and how alterations in chromosome number or structure may arise during mitosis and meiosis *(Chapter 2)*
- Be able to describe Mendelian and non-Mendelian modes of inheritance *(Chapters 1, 7 and 13)*.

**Have an understanding of the role of genetic factors in health and disease**
- Understand how mutations can affect gene dosage and function *(Chapter 6)*
- Understand the use of polymorphisms as genetic markers *(Chapter 9)*
- Be aware of the role of genetic and environmental factors in multifactorial conditions such as congenital anomalies, cancer, diabetes and psychiatric illness *(Chapters 12 and 13)*
- Be aware that population ancestry may affect the frequency of susceptibility alleles and of Mendelian diseases *(Chapter 10)*

**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 *(Chapter 1)*
- Understand the clinical implications of phenomena such as incomplete penetrance, variation in expression, anticipation and new mutations *(Chapters 1, 4 and 10)*
• Be aware of the possibility of heterogeneity in a genetic disease and the potential impact on diagnosis (Chapters 1 and 9)
• Understand the principles of risk estimates for Mendelian diseases (Chapters 1 and 14)
• Be aware of clinical indicators that suggest an inherited predisposition to cancer (Chapter 12)
• Be able to describe clinical features of common Mendelian diseases (all Case studies and Disease boxes)
• Be able to describe clinical features of common chromosomal disorders (Chapter 2)
• Be aware of the types of clinical features which suggest a dysmorphic or malformation syndrome (Chapters 1, 4 and 6)
• Be aware of the roles of genes and teratogens in human congenital anomalies (Chapters 1, 4 and 6)

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 (Chapter 14)
• Be aware of the impact of genetic diagnosis on the extended family (Chapters 4, 9 and 14)
• Be able to communicate the concept of risk in a manner that can be understood by the patient (Chapters 1 and 14)
• Be aware of major ethical issues in genetics (all Case studies)
• Be aware of the potential uses and misuses of genetic information (Chapter 11)

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 (Chapter 11)
• Be aware of the differences and similarities between diagnostic, predictive and carrier genetic testing (Chapter 14)
• Be aware that ‘genetic tests’ can include clinical examination, metabolite assays and imaging as well as analysis of nucleic acid (Chapters 8 and 14)
• Be aware of the different laboratory techniques to investigate genetic material and their advantages and limitations (Chapters 2, 4 and 5)
• Be able to interpret a standard genetics laboratory report (cytogenetic and molecular genetic) (Chapters 2, 4, 5 and 6)
• Be aware of parameters governing population genetic screening, current population genetic screening programs and guidelines for the introduction of such programs (Chapter 11)

Know how to obtain current information about scientific and clinical applications of genetics, particularly from specialised genetics services
• Know when and where to get genetic advice and information
• Know when and how to make relevant referrals to the specialised genetics services

Potential topics for advanced study (eg in Special Study Modules)
• Epigenetics, including imprinting (Chapter 7)
• The impact of selective advantage and natural selection on human genetic disorders (Chapter 10)
• Developmental genetics: selective transcription; differentiation; stem cells (Chapters 3 and 14)
• Gene therapy (Chapter 14)
• Pharmacogenetics (Chapter 8)