3.02 THE CLINICAL EXAMPLE ON

Genetics in Primary Care

This statement is part of the curriculum produced by the Royal College of General Practitioners (RCGP) which defines the learning outcomes for the discipline of general practice and describes the skills you require to practise medicine as a general practitioner in the National Health Service (NHS) of the United Kingdom. Although primarily aimed at the start of independent work as a general practitioner, it must also prepare the doctor beyond the training period and provide support for a professional life of development and change.
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KEY MESSAGES

- It has been estimated that at least one in ten of the patients seen in primary care has a disorder with a genetic component.
- There are three main themes of genetics in primary care: identifying patients with, or at risk of, a genetic condition; clinical management of genetic conditions; communicating genetic information.
- Taking and considering a genetic family history is a key skill in identifying families with Mendelian disorders and clusters of common conditions such as cancer, cardiovascular disease and diabetes.
- General practitioners (GPs) have a key role in identifying patients and families who would benefit from being referred to appropriate specialist genetic services.
- General practice plays a key part in discussing results from the antenatal and newborn screening programmes which are identifying carriers and people affected with genetic conditions.
- Information about genetic susceptibility in common conditions (currently being gathered through research studies) is likely to offer additional information about risk factors to aid management.
- Genomic information is currently being utilised in the stratified use of certain medicines.
CASE ILLUSTRATION

Adrian, a healthy 44-year-old man, is concerned about developing cancer because his father was diagnosed with bowel cancer at the age of 45, and died at the age of 48. As this seemed a very early age for Adrian’s father to have cancer, the general practitioner asked if anyone else in the family had cancer.

Adrian said that his paternal grandmother also died from bowel cancer in her 60s, as did one of his father’s sisters in her 50s. Another sister (Diana) had a cancer in her early 50s, which was treated by hysterectomy. She is now well and in her mid-60s.

The general practitioner checked the guidelines on the local Regional Genetics Centre website and saw that such a cluster may suggest an underlying inherited predisposition. The GP referred Adrian to the clinical genetics service. A genetic counsellor telephoned him and took a more detailed family history. The genetics service then checked the histological diagnoses in the cancer registry and found that his aunt had had endometrial cancer.

Adrian saw a consultant clinical geneticist who explained that the family pattern could be consistent with one of the family cancer syndromes inherited in an autosomal dominant manner. The geneticist felt that it might be helpful to ask if Adrian’s aunt Diana would be seen in the genetic clinic to discuss the possibility of giving a blood sample to see if a mutation could be found in a particular gene. The consultant also recommended that Adrian be referred for colonoscopy.

Adrian’s aunt Diana was also one of the GP’s patients. She agreed to be seen by the genetics service and agreed that her results could be shared with her family. A molecular genetic test revealed a pathogenic mutation in the MSH2 gene, which predisposes to bowel and endometrial cancer. Diana went to see her GP to discuss the fact that the genetics service had recommended screening by colonoscopy. She was concerned because she thought the treatment for her ‘cancer of the womb’ had been the end of the matter.

Adrian decided to be tested to see whether he had inherited the mutation. However, while waiting for the result he became concerned about how best to explain the findings to the rest of his family, and particularly any implications for his two children. He discussed these concerns with his GP, whom he asked to review autosomal dominant inheritance with him, and the contents of the letter from the regional genetics service.

Adrian was delighted to find that he had not inherited the MSH2 mutation and therefore had the same probability as anyone else in the population of developing bowel cancer. During a subsequent consultation for another matter, Adrian mentioned that although he was pleased that his children would not be at risk, he did feel guilty that he had been ‘the lucky one’ in the family. He had also originally interpreted the fact that he had not inherited the mutation to mean he had no risk of bowel cancer, so his GP stressed that it would be wise to take part in the population screening for colorectal cancer at the appropriate age.
To help you understand how the GP curriculum can be applied to this case, ask yourself the following questions:

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<tr>
<th>Category</th>
<th>Question</th>
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<tr>
<td><strong>Primary care management</strong></td>
<td>How can I recognise individuals or families at the highest risk of genetic conditions?</td>
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<td><strong>Person-centred care</strong></td>
<td>How can I, as a GP, help with the impact of the genetic diagnosis in this family?</td>
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<td><strong>Specific problem-solving skills</strong></td>
<td>What are the best ways of taking, recording and interpreting a genetic family history?</td>
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<td><strong>A comprehensive approach</strong></td>
<td>What roles should the GP play in referral, and in the co-ordination of screening and management in this family?</td>
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<td><strong>Community orientation</strong></td>
<td>How would the views of the local community towards genetics and screening impact on the ways in which the family are likely to take up services?</td>
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<td><strong>A holistic approach</strong></td>
<td>Why might the person who was found not to have inherited a predisposition feel so guilty?</td>
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<td><strong>Contextual features</strong></td>
<td>How can I cover personal and family issues relating to genetics during the time available for a ‘regular’ consultation?</td>
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<td><strong>Attitudinal features</strong></td>
<td>How do my attitudes as a GP influence the way information about genetics and family results is given and shared?</td>
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<td><strong>Scientific features</strong></td>
<td>How can I ensure that information for my patients about the availability of genetic tests and targeted management is up to date?</td>
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LEARNING OUTCOMES

The following learning outcomes or objectives relate specifically to the management of genetics in primary care. These learning outcomes are in addition to those detailed in the core statement, *Being a General Practitioner*. The core statement and this statement should be used in conjunction with the other curriculum statements. In order to demonstrate the core competences in the area of genetics, you will require knowledge, skills and attitudes in the following areas:

**The RCGP areas of competence**

1 Primary care management

This area of competence is about how you manage your contacts with patients, dealing competently with any and all problems that are presented to you. (This area of competence is not limited to dealing with the management of the practice.)

This means that as a GP you should:

1.1 Be aware that variations in the human genome may have no effect, may lead to a predisposition to common diseases (such as coronary artery disease or cancer), or may result in serious conditions in a significant minority of your practice

1.2 Describe how to identify patients with, or at risk of, a genetic condition through considering the family history and applying knowledge of inheritance patterns, or patients with diagnoses known to have a genetic cause

1.3 Demonstrate an appreciation of the importance of identifying families with autosomal dominant conditions such as familial hypercholesterolaemia and polycystic kidney disease to ensure that affected family members receive appropriate treatment, and the importance of offering carrier testing for families with autosomal recessive conditions such as sickle cell, thalassaemia or cystic fibrosis

1.4 Demonstrate appropriate skills to communicate information to patients about genetics in a comprehensible way with particular awareness of the need:
   1.4.1 for confidentiality when information received from or about one individual can be used in a predictive manner for another family member in the same practice
   1.4.2 to remain non-directive and non-judgemental

1.5 Describe the reproductive options available to those with a known genetic condition (including: having no children; adoption; gamete donation; prenatal diagnosis)

1.6 Describe local and national referral guidelines (for instance, for a family history of breast or colon cancer)

1.7 Describe how to access guidelines for managing patients with genetic conditions (such as familial hypercholesterolaemia or sickle cell disease)

1.8 Describe the organisation of genetics services and how to make appropriate referrals
1.9 Demonstrate comprehensive management for those patients with, or at risk of, genetic conditions through co-ordination of care with other primary care professionals, geneticists and other appropriate specialists. This is particularly important because genetic conditions are often multisystem disorders.

1.10 Describe the support services available for those with a genetic condition (e.g. Contact a Family)

### 2 Person-centred care

This area of competence is about understanding and relating to the context of your patients as individuals, and developing the ability to work in partnership with them.

This means that as a GP you should:

2.1 Demonstrate an awareness of the potential emotional, psychological and social impacts of a genetic diagnosis on a patient and his or her family, particularly associated with guilt about ‘passing on’ a condition.

2.2 Demonstrate an awareness of the different implications for other family members depending on the genetic cause of a condition (autosomal dominant and recessive and X-linked single-gene inheritance; de novo and inherited chromosomal anomalies; mitochondrial inheritance and somatic mutation).

2.3 Demonstrate an awareness of the ethical issues that may arise, including confidentiality and non-disclosure of genetic information within families; genetic testing in children; the 'right not to know' and exercising care in the use of information (for instance in access to insurance or employment issues).

2.4 Demonstrate an awareness of the need to ensure that systems are in place to follow up patients who have, or are at risk of, a genetic condition and have chosen to undergo regular surveillance (for example: imaging for breast cancer and for adult polycystic kidney disease; or endoscopy for colon cancer).

### 3 Specific problem-solving skills

This area of competence is about the context-specific aspects of general practice, dealing with early and undifferentiated illness and the skills you need to tolerate uncertainty, and marginalise danger, without medicalising normality.

This means that as a GP you should:

3.1 Be able to take and interpret a family history. This involves:

3.1.1 knowledge of relevant questions

3.1.2 knowledge of basic inheritance patterns (autosomal dominant and recessive, X-linked, mitochondrial, multifactorial)

3.1.3 understanding that while some genetic conditions always present with the same signs and symptoms, others can show variability between family members, particularly some autosomal dominant conditions (such as neurofibromatosis type 1)
3.2 Demonstrate an awareness of the heterogeneity in genetic diseases and understand the principles of assessing genetic risk, e.g.
   3.2.1 principles of risk estimates for family members of patients with Mendelian diseases
   3.2.2 principles of recurrence risks for simple chromosome anomalies, e.g. trisomies
   3.2.3 the use of information from susceptibility loci in common complex conditions
   3.2.4 the ability to use online risk assessment tools as they become available

3.3 Demonstrate an awareness of the different uses of genetic tests (diagnostic, predictive, carrier testing), their limitations and ethical considerations (for instance associated with testing in children and with presymptomatic testing)

3.4 Demonstrate an awareness that it is not always possible to determine the:
   3.4.1 cause of a condition (e.g. a learning disability) that may be genetic in origin
   3.4.2 mutation responsible for a genetic condition

3.5 Demonstrate an awareness of the genetic aspects of antenatal and newborn screening programmes (e.g. Down’s syndrome, cystic fibrosis, sickle cell and thalassaemia) and know their indications, uses and limitations, and from where to obtain information

4 A comprehensive approach

This area of competence is about how you as a general practitioner must be able to manage co-morbidity, co-ordinating care of acute illness, chronic illness, health promotion and disease prevention in the general practice setting.

This means that as a GP you should:

4.1 Demonstrate an awareness that preventative measures or targeted treatments exist for some genetic conditions (for example: lifestyle interventions; mastectomy and/or oophorectomy for BRCA1/2 mutation carriers; colectomy for adenomatous polyposis coli (APC) mutation carriers; statin use for familial hypercholesterolaemia; venesection for haemochromatosis; losartan for patients with Marfan syndrome)

5 Community orientation

This area of competence is about the physical environment of your practice population, and the need to understand the interrelationship between health and social care, and the tensions that may exist between individual wants and needs and the needs of the wider community.

This means that as a GP you should:

5.1 Demonstrate an awareness that the makeup of the local population may affect the prevalence of genetic conditions and attitudes towards genetic disease
6 A holistic approach

This area of competence is about your ability to understand and respect the values, culture, family structure and beliefs of your patients, and understand the ways in which these will affect the experience and management of illness and health.

This means that as a GP you should:

6.1 Demonstrate an awareness that a patient’s cultural and religious background and beliefs concerning inheritance and genetics are important to consider in providing care for people and families with, or at risk of, genetic conditions. An example of a belief concerning inheritance is that a particular genetic disease in a family is linked with a particular physical appearance.

The essential features of you as a doctor

The three essential features (EFs) below are concerned with the features of you as a doctor which may influence your ability to apply the core competences to real life in the work setting.

EF1 Contextual features

This essential feature is about understanding your own context as a doctor and how it may influence the quality of your care. Important factors are the environment in which you work, including your working conditions, community, culture, financial and regulatory frameworks.

Examples of this are:

   EF1.1 Demonstrating an awareness that consultations involving the giving of genetic information and discussion may require more time
   EF1.2 Demonstrating an awareness that a genetic diagnosis in an individual may have implications for the management of other family members who may ask for a consultation
**EF2 Attitudinal features**

This essential feature is about your professional capabilities, values, feelings and ethics and the impact these may have on your patient care.

Examples of this are:

- **EF2.1** Demonstrating an awareness of your professional limits in regard to managing genetic conditions and knowing when and where to seek advice
- **EF2.2** Demonstrating an ability to discuss information about genetic conditions in a non-directive, non-judgemental manner, ensuring that your own beliefs do not influence the content of the consultation and the management options offered to a patient

**EF3 Scientific features**

This essential feature is about the need to adopt a critical and evidence-based approach to your work, maintaining this through lifelong learning and a commitment to quality improvement.

Examples of this are:

- **EF3.1** Demonstrating an awareness that genetics is a rapidly evolving area. It is therefore important that you keep up to date with clinical advances and their implications on ethical debate and service planning, particularly how genomic information can contribute to risk factors in common conditions and the personalisation of management through stratified use of medicines
LEARNING STRATEGIES

Work-based learning – in primary care

As a GP specialty trainee, primary care is the ideal setting for you to learn about genetics because of the family-based focus and opportunities for staged counselling.

Learning opportunities which present during consultations include: how to recognise conditions with a genetic component; the appropriate management of the genetic implications for the individual and the family, particularly where there are ethical, social and legal issues; and referring patients appropriately to specialist services. As many common conditions seen in primary care are multifactorial, with a genetic component – including cancer, diabetes and heart disease – these can contribute to a developing awareness of how genetic components can affect disease.

Many of the skills required for management of families with conditions with a genetic component are part of the core skills of a general practitioner, and will be consolidated through reflective practice and discussion with your GP trainer. You should also supplement your counselling and management skills with the theoretical knowledge outlined in the ‘knowledge base’ in this statement.

Work-based learning – in secondary care

GP trainees with a particular interest in genetics may also wish to take up the opportunity to learn from consultant geneticists and genetic counsellors working in regional specialist genetics services. This should include developing your understanding of the genetic counselling process, diagnosis and management of genetic conditions, and reproductive options including prenatal diagnosis for at-risk couples.
LEARNING RESOURCES

Examples of relevant texts and resources

- Harper PS. Practical Genetic Counselling London: Hodder Arnold, 2010
- RCGP. InnovAiT (the journal for Associates in Training (AiTs)), August 2008 edn contains articles covering aspects of 'Genetics and Primary Care', http://ino.sagepub.com

Web resources

*Genetics in Practice*

**Taking a genetic family history**
The NHS National Genetics and Genomics Education Centre website has a number of resources designed around taking and drawing a family history, including a series of factsheets, pedigree drawing exercises and videos. In addition, the website contains information about core concepts in genetics, information about genetic conditions and an extensive knowledge base of genetic terms.

www.geneticseducation.nhs.uk

**Accessing specialist genetics services**
The British Society for Human Genetics (BSHG) provides a directory of the UK genetics centres, including links to each regional genetic centre’s website as well as the contact details of clinicians to whom referrals can be made. Under the section ‘For patients’, there is detailed information about what patients can expect at their medical genetics appointment.

www.bsgm.org.uk

**Availability of genetic tests**
UK Genetic Testing Network.

www.ukgtn.nhs.uk

**Information for families**
Contact a Family
Contact a Family is a UK-wide charity that provides advice, information and support to parents who have a child with a disability.

www.cafamily.org.uk
Unique
Unique is a UK-based charity which provides information and support to both families and individuals affected by rare chromosomal conditions, as well as the health professionals involved in providing ongoing medical management and care.
www.rarechromo.org

Databases of genetic conditions
GeneTests
This site, funded by the US National Institute of Health (NIH), has a GeneReviews section that gives information on specific diseases and an illustrated glossary in its Educational Resources section. GeneTests also has an international directory of genetic testing laboratories and clinics.
www.genetests.org/

Orphanet
Orphanet is a database of rare diseases (90% caused by genetic factors) and orphan drugs. It includes peer-reviewed information on these diseases and a directory of current services.
www.orpha.net

Genetics education for primary care physicians
e-GP
The e-GP course on Genetics in Primary Care includes topics such as taking, drawing and interpreting genetic family histories, communicating genetic information and managing and referring patients.
www.e-GP.org

The NHS National Genetics and Genomics Education Centre
This website contains information about core concepts in genetics, information about genetic conditions and an extensive knowledge base of genetic terms, as well as practical information on taking and recording a genetic family history.
www.geneticseducation.nhs.uk

Cancer genetics
National Cancer Institute
This US website provides information for healthcare professionals on prevention, genetics and causes of a number of different types of cancer.
www.cancer.gov
Screening

PEGASUS
Professional Education for Genetic Assessment and Screening (PEGASUS) was commissioned by the NHS Sickle Cell and Thalassaemia Screening Programme and aims to facilitate training in basic genetics for health professionals involved in antenatal and newborn screening in England.

www.pegasus.nhs.uk

UK National Screening Committee
This webpage give access to all the UK screening programmes, including the antenatal and newborn and cancer programmes.

www.screening.nhs.uk
ACKNOWLEDGEMENTS

This curriculum statement is based on the original statement *Genetics in Primary Care* in the 2007 version of the RCGP Curriculum. It has been developed by the RCGP in partnership with the NHS National Genetics Education and Development Centre.

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**Date of this version:** May 2014

The 2007 version of the statement and subsequent updates can be found on the RCGP website. The Royal College of General Practitioners would like to express its thanks to all the individuals and organisations who have contributed so generously to past and present versions of this statement.