

The RCGP Curriculum: Clinical Modules

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➤ 3.02 Genetics in Primary Care

Summary

- 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

Knowledge and skills guide

Core Competence: Fitness to practise

This concerns the development of professional values, behaviours and personal resilience and preparation for career-long development and revalidation. It includes having insight into when your own performance, conduct or health might put patients at risk, as well as taking action to protect patients.

This means that as a GP you should:

- Demonstrate an awareness of your professional limits in regard to managing genetic conditions and know when and where to seek advice

- Demonstrate 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

Core Competence: Maintaining an ethical approach

This addresses the importance of practising ethically, with integrity and a respect for diversity.

This means that as a GP you should:

- 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)
- 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)

Core Competence: Communication and consultation

This is about communication with patients, the use of recognised consultation techniques, establishing patient partnership, managing challenging consultations, third-party consulting and the use of interpreters.

This means that as a GP you should:

- Demonstrate appropriate skills to communicate information and risk to patients about genetics in a comprehensible way with particular awareness of the need:
 - 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
 - to remain non-directive and non-judgemental
- 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
- Demonstrate 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
- Demonstrate an awareness that consultations involving the giving of genetic information and discussion may require more time

Core Competence: Data gathering and interpretation

This is about interpreting the patient's narrative, clinical record and biographical data. It also concerns the use of investigations and examination findings, plus the adoption of a proficient approach to clinical examination and procedural skills.

This means that as a GP you should:

- Be able to take and interpret a family history. This involves:
 - knowledge of relevant questions
 - knowledge of basic inheritance patterns (autosomal dominant and recessive, X-linked, mitochondrial, multifactorial)
 - 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)

Core Competence: Making decisions

This is about having a conscious, structured approach to decision-making; within the consultation and in wider areas of practice.

This means that as a GP you should:

- 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
- Demonstrate an awareness of the heterogeneity in genetic diseases and understand the principles of assessing genetic risk, e.g.
 - principles of risk estimates for family members of patients with Mendelian diseases
 - principles of recurrence risks for simple chromosome anomalies, e.g. trisomies
 - the use of information from susceptibility loci in common complex conditions
 - the ability to use online risk assessment tools as they become available
- 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)
- Demonstrate an awareness that it is not always possible to determine the:
 - cause of a condition (e.g. a learning disability) that may be genetic in origin
 - mutation responsible for a genetic condition
- 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

Core Competence: Clinical management

This concerns the recognition and management of common medical conditions encountered in generalist medical care. It includes safe prescribing and medicines management approaches.

This means that as a GP you should:

- 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
- 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
- Describe the reproductive options available to those with a known genetic condition (including: having no children; adoption; gamete donation; prenatal diagnosis)
- Describe local and national referral guidelines (for instance, for a family history of breast or colon cancer)
- Describe how to access guidelines for managing patients with genetic conditions (such as familial hypercholesterolaemia or sickle cell disease)

Core Competence: Managing medical complexity

This is about aspects of care beyond managing straightforward problems. It includes multi-professional management of co-morbidity and poly-pharmacy, as well as uncertainty and risk. It also covers appropriate referral, planning and organising complex care, promoting recovery and rehabilitation.

This means that as a GP you should:

- 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
- 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)

Core Competence: Working with colleagues and in teams

This is about working effectively with other professionals to ensure good patient care. It includes sharing information with colleagues, effective service navigation, use of team skill mix, applying leadership, management and team-working skills in real-life practice, and demonstrating flexibility with regard to career development.

This means that as a GP you should:

- Describe the organisation of genetics services and how to make appropriate referrals
- Describe the support services available for those with a genetic condition (e.g. Contact a Family)

Core Competence: Maintaining performance, learning and teaching

This area is about maintaining performance and effective CPD for oneself and others, self-directed adult learning, leading clinical care and service development, participating in commissioning, quality improvement and research activity.

This means that as a GP you should:

- Demonstrate 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

Core Competence: Organisational management and leadership

This is about the understanding of organisations and systems, the appropriate use of administration systems, effective record keeping and utilisation of IT for the benefit of patient care. It also includes structured care planning, using new technologies to access and deliver care and developing relevant business and financial management skills.

This means that as a GP you should:

- 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)

Core Competence: Practising holistically and promoting health

This is about the physical, psychological, socioeconomic and cultural dimensions of health. It includes considering feelings as well as thoughts, encouraging health improvement, preventative medicine, self-management and care planning with patients and carers.

This means that as a GP you should:

- 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

Core Competence: Community orientation

This is about involvement in the health of the local population. It includes understanding the need to build community engagement and resilience, family and community-based interventions, as well as the global and multi-cultural aspects of delivering evidence-based, sustainable healthcare.

This means that as a GP you should:

- Demonstrate an awareness that the makeup of the local population may affect the prevalence of genetic conditions and attitudes towards genetic disease

Case discussion

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 see the genetics service and that her results could be shared with her family. A 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.

Reflective questions

To help you understand how the GP curriculum can be applied to this case, ask yourself the following questions:

Core Competence	Reflective Questions
<p>Fitness to practise This concerns the development of professional values, behaviours and personal resilience and preparation for career-long development and revalidation. It includes having insight into when your own performance, conduct or health might put patients at risk, as well as taking action to protect patients.</p>	<p>How do my own views and experience influence the way I share information about genetic tests and results, in particular those that may impact on the wider family?</p>
<p>Maintaining an ethical approach This addresses the importance of practising ethically, with integrity and a respect for diversity.</p>	<p>What potential ethical dilemmas could such a case present, and how would I address them?</p>
<p>Communication and consultation This is about communication with patients, the use of recognised consultation techniques, establishing patient partnerships, managing challenging consultations, third-party consulting and the use of interpreters.</p>	<p>How can I, as a GP, communicate the risk of various common patterns of genetic inheritance in simple language to the patients and their families?</p> <p>What do I need to be mindful of when giving any information relating to a genetic disorder?</p>
<p>Data gathering and interpretation This is about interpreting the patient's narrative, clinical record and biographical data. It also concerns the use of investigations and examination</p>	<p>What tools are available to GPs to recognise and stratify patients with potential genetic cancers?</p> <p>How can I recognise individuals or families at the highest</p>

findings, plus the adoption of a proficient approach to clinical examination and procedural skills.	risk of genetic conditions?
<p>Making decisions</p> <p>This is about having a conscious, structured approach to decision-making; within the consultation and in wider areas of practice.</p>	<p>What are the best ways of taking, recording and interpreting a genetic family history?</p> <p>When am I likely to refer patients to secondary care?</p>
<p>Clinical management</p> <p>This concerns the recognition and management of common medical conditions encountered in generalist medical care. It includes safe prescribing and medicines management approaches.</p>	What guidelines exist to guide my management of people with genetic conditions?
<p>Managing medical complexity</p> <p>This is about aspects of care beyond managing straightforward problems. It includes multi-professional management of co-morbidity and poly-pharmacy, as well as uncertainty and risk. It also covers appropriate referral, planning and organising complex care, promoting recovery and rehabilitation.</p>	<p>What roles should the GP play in referral, and in the co-ordination of screening and management in this family?</p> <p>What other sources of advice and support are available to GPs?</p>
<p>Working with colleagues and in teams</p> <p>This is about working effectively with other professionals to ensure good patient care. It includes sharing information with colleagues, effective service navigation, use of team skill mix, applying leadership, management and team-working skills in real-life practice, and demonstrating flexibility with regard to career development.</p>	<p>How can GPs work with the local genetics department to facilitate a seamless transfer of information in both directions?</p> <p>How can the practice work as a team to ensure that patients with identified cancer predisposition are not lost to follow ups?</p>
<p>Maintaining performance, learning and teaching</p> <p>This is about maintaining performance and effective CPD for oneself and others. This includes self-directed adult learning, leading clinical care and service development, participating in commissioning*, quality improvement and research activity.</p>	<p>How can I ensure that information for my patients about the availability of genetic tests and targeted management is up to date?</p> <p>How do I keep myself updated about new developments in genetics?</p> <p>Where can I access quick and reliable information about any query regarding a genetic disorder?</p>
<p>Organisational management and leadership</p> <p>This is about the understanding of organisations and systems, the appropriate use of administration systems, effective record keeping and utilisation of IT for the benefit of patient care. It also includes structured care planning, using new technologies</p>	<p>What are the Read codes for recording a family history of cancers or any other genetic disorder?</p> <p>What 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?</p>

to access and deliver care and developing relevant business and financial management skills.	
<p>Practising holistically and promoting health</p> <p>This is about the physical, psychological, socioeconomic and cultural dimensions of health. It includes considering feelings as well as thoughts, encouraging health improvement, preventative medicine, self-management and care planning with patients and carers.</p>	<p>Why might the person who was found not to have inherited a predisposition feel so guilty?</p>
<p>Community orientation</p> <p>This is about involvement in the health of the local population. It includes understanding the need to build community engagement and resilience, family and community-based interventions, as well as the global and multi-cultural aspects of delivering evidence-based, sustainable healthcare.</p>	<p>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?</p> <p>Where are the local genetic departments and are there any agreed local protocols for referrals?</p>

How to learn this area of practice

Work-based learning

In primary care

As a GP 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.

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.

Useful learning resources

Books and publications

- Bradley-Smith G, Hope S, Firth H, Hurst J. *Oxford Handbook of Genetics* Oxford University Press, 2009
- Harper PS. *Practical Genetic Counselling* London: Hodder Arnold, 2010

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

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

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

Screening:

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