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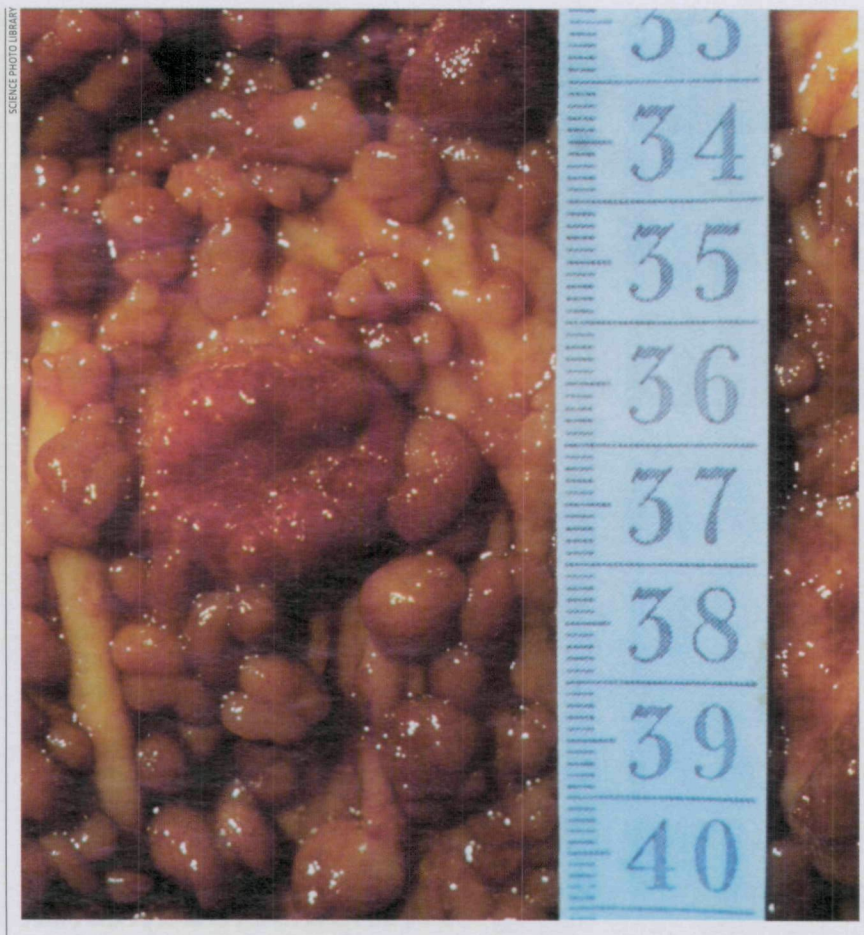

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Polyps in the colon of a patient with familial adenomatous polyposis

GENETICSINPRIMARYCARE

Genetic testing, families and confidentiality

Clinical geneticist **Professor Anneke Lucassen** and GP **Dr Imran Rafi** offer advice on communicating genetic risk with relatives, recording information and protecting privacy

There are now almost 2,000 tests for genetic diseases available, compared with around 500 just 10 years ago.

Advances in genetic and genomic research mean that identification of a genetic condition – or susceptibility to disease – is now possible in routine healthcare as well as being offered on a commercial basis, as highlighted in other articles in this series.

Impact on family members

Genetic tests have huge potential to guide the management of disease in an individual, but at the same time these tests may reveal that others in the family are at risk (see table below for examples).

For instance, identifying that a woman has a BRCA1 gene mutation means that her sisters and daughters have a 50% chance that they have also inherited this condition. That figure is the same for her brothers and sons, although their risks of cancer will be lower. Affected relatives might benefit from the interventions available. The others can be reassured that they have not inherited the familial genetic risk.

Genetic services and testing

Genetic diagnoses are not new to medical practice and some indirect genetic tests – like renal ultrasound for polycystic kidney disease – have been around for many years.

Until recently, the routine delivery of predictive DNA tests has largely been the preserve of regional genetic services.

Unlike general practice, they often hold family rather than individual case notes and an initial consultation usually involves a detailed description of the family – people who might have an interest in subsequent diagnoses.

Most patients seek genetic advice with at least a partial aim of also helping their relatives. But there may be situations where family members are not in close contact, or because of concerns to protect people from distressing information, relatives may remain in ignorance.

Health professionals may then know about individuals to whom they could provide more accurate information, but concern about breaching the confidentiality of another prevents them from doing so.

This is not an unfamiliar scenario to GPs with many members of the same family on their lists. It's not unusual to know that one relative does not want another to know aspects of their medical history.

Usually keeping such confidences is standard practice, but where this impedes optimal management of a relative, there may be reasons to share what might be regarded as familial information.

Implications for general practice

The best way to ensure family members can avail themselves of appropriate genetic medicine is to document at the stage when an individual first provides consent for a genetic test the possible need for communication with other family members.This approach is in line with the notions of 'familial altruism' and 'genetic solidarity' endorsed by the Human Genetics Commission.

Genetic services already do this routinely, but as genetic medicine expands, they will increasingly seek the help of GPs to manage such familial communication.

As genetics enters mainstream medicine, GPs will increasingly be faced with questions and dilemmas such as those in the case study on the right. GPs tackle consent issues daily, but it's unusual to routinely seek consent for the use of the result in the management of others.

Now GPs will need to consider when it is appropriate to do so, and what can be done if such consent has not been obtained. It is sometimes possible to broach with a family member the fact they have a family history of a disease, and to suggest a genetic test, without revealing the identity of the person originally tested.

Other challenges will include how genetic information – family histories, risk assessments and genetic test results – is recorded on IT systems.

There will need to be good communication between geneticists and primary care as well as new models of service such as community-based genetic counselling.

Policy and guidance

The GMC guidance on confidentiality has helpfully expanded the list of possible breaches of confidence to include genetic medicine.

It emphasises that a breach of confidence may be legitimate if you are concerned that relatives do not know about potential genetic risks for which there may be a possible intervention.

This is useful in rare cases where consent has been actively withheld, but further guidance will also be useful for the more routine situation where a GP is unsure whether consent to share has been gained.

The Joint Committee on Medical Genetics is currently revising its guidance on consent and confidentiality in genetic practice.[2] The new guidance will be issued later this year in 2011 and will give guidance suggesting what acceptable practice is.

The Department of Health is currently consulting on its information strategy, *Liberating the NHS: An Information Revolution*,[3] which will have possible implications on how information on patient records is recorded and shared.

CASE HISTORY

Hereditary nonpolyposis colon cancer

A female patient of yours has an hMSH2 mutation – putting her at increased risk of hereditary nonpolyposis colon cancer. She has has been unable to tell her children that they have a 50% chance of having inherited this mutation and if they have they will require two-yearly colonoscopy surveillance. The patient has told the geneticist the family are under your care and they write to you to ask you to discuss the genetic risks with the children. Can you do this without breaching the patient's confidence?

You need to consider whether it might be legitimate to use the genetic information in question even without consent, particularly if the use does not disclose the identity of the individual. For example: 'We know you have a strong family history of condition X and a genetic test Y will tell you your risk.' This won't be a solution in all cases, because this may reveal the identity of the person whose confidence you are trying to protect.

GENES, DISEASE PREDISPOSITION AND POSSIBLE INTERVENTIONS

Genetic status	Predisposition	Example Intervention
BRCA1 or BRCA2 gene mutations	Breast cancer	MRI of breast and/or mammography screening
Mutation in APC gene	Familial adenomatous polyposis	Prophylactic colectomy
LDL-receptor mutation	Familial hypercholesteroaemia	Statins
LQT and cardiomyopathy	Cardiac arrthymias, sudden death gene mutations	Pharmacological therapy, implantable defibrillators
MLH1, MSH2, MSH6 mutations	Colon cancer	Surveillance by colonoscopy and possible chemoprevention

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The aim of the PCGS is to support and facilitate the educational needs of primary care health professionals, helping them translate the continuing advances in clinical genetics into practice. For further information visit www.pcgs.org.uk.

Further information

The Public Health Genetics Foundation in Cambridge (www.phgfoundation.org/) has considered the issue of introducing genetic practice into mainstream practice and the National Genetics Education and Development Centre (www.geneticseducation.nhs.uk) has useful case histories and teaching tools.The British Society for Human Genetics (www.bshg.org) also has useful guideline documents.

References

1 General Medical Council. Confidentiality – the duties of a doctor registered with the General Medical Council. October 2009. www.gmc-uk.org

2 Joint Committee on Medical Genetics. Consent and confidentiality in genetic practice – guidance on genetic testing and sharing genetic information. April 2006. www.bshg.org.uk.

3 Department of Health. Liberating the NHS: an information revolution. Consultation published October 2010. www.dh.gov.uk

Further reading

Lucassen A and Parker M. Confidentiality and sharing genetic information with relatives. *Lancet* 2010;375:1507-9

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