

Understanding the role of genetics and genomics in health 2: implications for practice

With major advances in genetics and genomics, nurses need to develop their understanding of the topic and know how to integrate this into practice

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ABSTRACT Kirk M, Tonkin E (2009) *Understanding the role of genetics and genomics in health 2: implications for practice*. *Nursing Times*; 105: 46, 19-23. This second article in a two part series on genetics and genomics in healthcare focuses on the implications for nursing practice. Part 1 examined the progress of research in this area and discussed the implications for healthcare. This part outlines the nurse's role in supporting the patient pathway, and some of the ethical issues and challenges facing nurses as they integrate genomic healthcare into practice.

INTRODUCTION

The scientific advances that have continued in the wake of the Human Genome Project's completion in 2003 are bringing a phenomenal increase in understanding about the causes of genetically complex but common conditions such as asthma, diabetes, autism, bipolar disorder and schizophrenia (Department of Health, 2008).

This offers new opportunities to diagnose disease more accurately and to predict it ahead of any clinical manifestation. Concerning the latter, identifying population groups at increased risk could help to target health promotion resources more effectively (Box 1).

Single-gene disorders, such as sickle cell disease, and conditions caused by alterations to chromosome number or structure are relatively rare, but collectively numerous.

Applications to healthcare "have enormous further potential in improving and rationalising management of a broad range of diseases, and in advancing strategies for disease prevention and public health"

(House of Lords Science and Technology Committee, 2009). Nurses need to be aware of the implications of these advances for their practice, education and for the profession.

THE CHALLENGE OF GENOMIC HEALTHCARE

One implication for nurses is indicated in the review of the genetics white paper, where the DH (2008) said that "over time, the relevance of genetics across other medical specialties would become increasingly clear to clinicians and others outside the genetics specialty".

This is an important point because there is limited awareness about specialist genetics services among the nursing profession.

Many nurses have little understanding of the relevance of genetics to nursing care, do not see it as important and are not confident in applying it to their practice (Kirk et al, 2007). They are also unlikely to have had adequate education about genetics during their training (Kirk and Tonkin, 2006). Tony's story shows how this can affect patient care (Box 2).

If Tony's experience reflects the limited genetics knowledge and understanding many nurses have, integrating genomics into everyday practice presents a challenge.

Another factor is that, although the government is investing in specialist genetics services to build capacity in response to the already significant increase in demand, these will be unable to cope with the

anticipated scope and scale of the development of genomic healthcare.

New models of care are continually being developed involving nurses in primary, acute and tertiary care working in collaboration with, but independently of, nurses and other healthcare professionals in the genetics specialism. Pilot programmes in cancer genetics have already demonstrated some success (DH, 2008) and, more recently, a service development initiative that involves cardiac nurses in cardiovascular genetics has begun.

What this highlights is that nurses and other health professionals need to engage with genomic healthcare as new advances emerge with increasing frequency. In doing so, the challenge for nurses is to:

- Know about, and understand how, genomic science applies to their practice;
- Understand the implications for the profession and for society in general;
- Manage both the expectations and concerns of patients;
- Educate others (and themselves);
- Demonstrate leadership in communicating about and responding to developments in genetics/genomics and related technologies.

GENOMICS APPLIED TO PRACTICE Knowledge for practice

The distinction between diseases and ill health caused by "genetics" (that is, due to single-gene and chromosome alterations)

BOX 1. IDENTIFYING AT-RISK GROUPS FOR TARGETED TREATMENT

Cardiac specialist nurse Lowri talks about familial hypercholesterolaemia, a dominantly inherited condition associated with high morbidity and mortality from cardiovascular disease. It affects around one in 500 people.

"Obviously, it is a genetic condition but I think the fact that it is easily identifiable and is easily treatable to prevent premature cardiovascular disease is a big issue with this. You know that for many genetic conditions the outcome is not so easily treatable and there are huge issues then in terms of having children and other lifestyle issues. But, with this, I think it's generally positive because something can be done and treatment can be offered and you are not asking them to make huge choices."

Source: NHS National Genetics Education and Development Centre (2009)

BOX 2. IMPACT OF LACK OF GENETICS KNOWLEDGE ON PATIENT CARE

Tony has sickle cell disease, a recessively inherited condition caused by a mutation in the beta-haemoglobin gene that affects about one in every 300 people of African-Caribbean origin. An estimated 125,000 people in the UK have the condition. Tony recounts his experience of transitional care as a young adult admitted to acute wards during episodes of excruciatingly painful sickle cell crisis.

“When I moved over to the adult wards, I found the staff had no knowledge of it at all really, hardly any of them.... I was in hospital and I just wanted to cry really. Because on the ward they put me on, the staff didn’t know anything about it. Not a bit.”

He describes one experience in relation to his inadequate pain management while on the ward and concludes: “That’s one of the worst experiences I have ever had and to me the most frightening. Like I said, now I’m scared of going back into hospital.”

Source: NHS National Genetics Education and Development Centre (2009)

Their understanding may be influenced by media reports or their own beliefs about how conditions run in families. While genetic counsellors are highly skilled at communicating information, patients spend relatively little time with them compared with, for example, community nurses. Sometimes, patients may not even be referred to genetics services. The nursing role here is to inform and support decision making about genetic testing, reproductive and lifestyle choices, and direct patients and families to further information;

● **Reflecting on practice** is particularly important where new knowledge and skills are being acquired and applied to practice. Reflection helps to maintain an awareness of the limitations of knowledge, and helps nurses to be aware of the wider implications of genetic information for patients, families and society in general.

The first step in this is for nurses to build on their “basic” knowledge to learn about how genetics/genomics applies to patients or clients in their particular area of practice. Diabetes is an example of this (Box 4).

and those from all other causes is no longer a valid model to apply to practice (see part 1 of this series). Rather, the combination of genetic make-up and the environment and their interaction leads to a continuum of diseases that affect patients across all healthcare settings.

While highly penetrant single-gene conditions lie at one end of the spectrum and those with a high degree of environmental causation – such as road traffic accidents – are at the other, what lies in between are the common complex conditions. These may involve interactions between several genes, the epigenome and the environment, with each gene playing its own part in the overall susceptibility to that disease, conferring an added or reduced risk. Knowledge of the genotype may play a part in clinical management and therapeutics.

What this means is that the core genetics knowledge that nurses need must now be broadened to encompass genomics together with the skills to apply this in practice along the patient pathway (Fig 1):

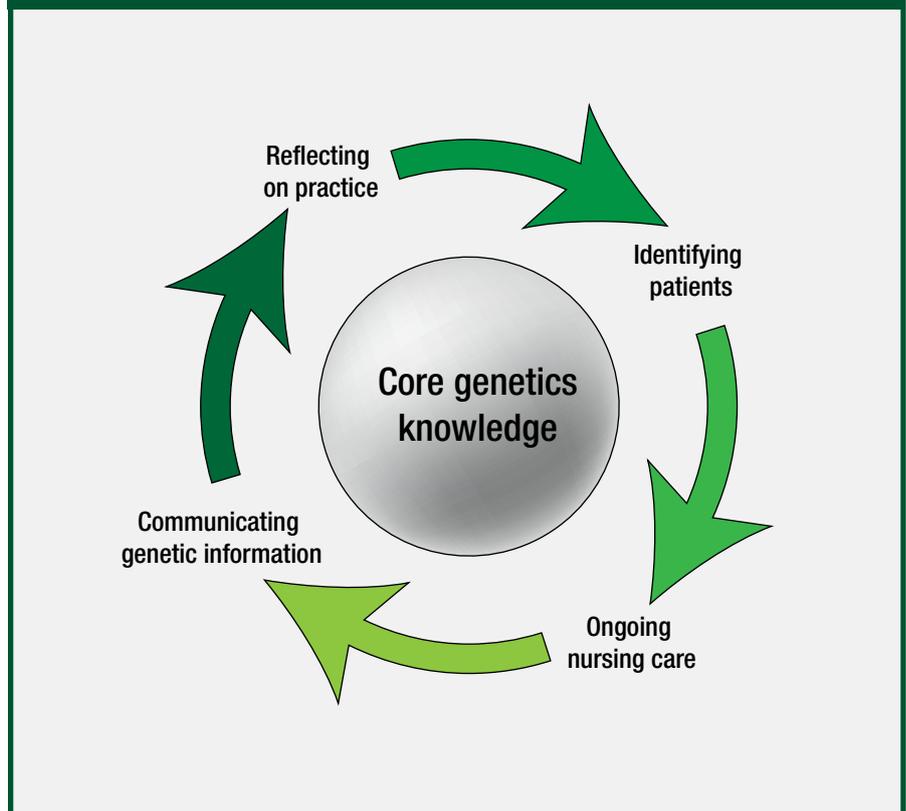
● **Identifying patients who might benefit from referral to genetics services or information:** this relies on nurses having an awareness of where genetics is likely to be most relevant to their patient group. Skills in family history taking help identify people potentially at risk. Knowing how and where to refer patients for specialist advice follows on from this (Box 3);

● **Ongoing nursing care:** providing ongoing care and support is a fundamental part of the nurse’s role. In this context, this includes knowing or finding out about the condition, listening to patients and families, helping them to understand the concept of risk, explaining prognosis, finding out about support groups and acting as a patient advocate. The psychosocial impact of

genetic information or diagnosis on patients and families may require longer term support from nurses who are involved in their ongoing care. Feelings of guilt and blame, loss and bereavement are not uncommon and require sensitive, empathic nursing care;

● **Communicating genetic information** in a way patients/families can understand is crucial, given the complexity of the field.

FIG 1. APPLYING GENETICS/GENOMICS KNOWLEDGE AND SKILLS IN NURSING PRACTICE



Catherine Hollick

BOX 3. REGIONAL GENETICS CENTRES

There is a network of over 20 regional genetics centres across the UK (including five in London) that offer a range of diagnostic and genetic counselling services for people with, at risk of, or susceptible to, genetic conditions. Some offer specific expertise in a particular genetics speciality. Each serves a population of 3-5 million. Genetic counsellors (including genetic specialist nurses) play a vital role in service delivery.

Contact information and details of services offered can be found online at tinyurl.com/genetics-directory.

ADVICE FROM GENETIC NURSE SPECIALISTS

The Association of Genetic Nurses and Counsellors (AGNC) has a wealth of expertise in caring for and supporting patients and families with genetic health issues. We asked members to identify the key factors that would promote effective integration of genetics/genomics into nursing practice. Their responses highlighted the importance of:

- Effective communication with patients and families;
- Knowledge of specialist genetics services, and using them for support;
- Developing knowledge and skills around taking and using family histories;
- Self-awareness, especially of the limitations of genetics knowledge and skills;
- Professional awareness, in relation to where genetics fits into the practice area, and in appreciating how integrating genetics might improve patient experience;
- Professional approach to practice, in working accurately and ethically;
- The working environment, in providing good support.

Box 5 presents some advice on incorporating genetics into practice.

REAL LIFE EXAMPLE

Consider this example from *Telling Stories: Understanding Real Life Genetics* (NHS National Genetics Education and Development Centre, 2009). Achondroplasia is a dominantly inherited condition associated with restricted growth. Most cases arise from a new mutation so “come out of the blue”. Maria recounts her feelings of shock when her son was diagnosed, and talks about the support that she received.

“I don’t really remember fully being told what achondroplasia was. We left the hospital feeling very confused and unsure of

what my son’s future held. I feel the situation could have been handled much better. When you are given such a life changing piece of information, there is a lot to take in.

“It was very fortunate for us that we had a brilliant health visitor who gave us all the support she could offer. She helped us find out all that she could, even though she knew as little as we did. She spent much of her own time doing research on the internet about the condition and where to get some special growth charts for my son to record his progress.

“There were numerous amounts of calls made by her to our consultant in an effort to get appointments or just to chase things up. She also... made sure that we got referrals to all the right people. We were just made to feel very looked after, in spite of the fact that the rest of the system seemed to be failing us.

“It felt like somebody was on our side and looking after our best interests.”

Think about what Maria needed:

- To be helped to understand her son’s diagnosis and the implications for her and her family;
- To be given this information at her own pace;
- To feel supported during a critical period in her family life.

To meet Maria’s needs, the health professional involved (in this case, the health visitor) needed to:

- Know when and how to make appropriate referrals;
- Act as advocate for the patient and family;
- Give support;
- Know how to obtain and communicate current, accurate genetics information;
- Recognise the limitations of her expertise.

It is clear from Maria’s account that the health visitor was able to meet her needs and delivered good quality care.

IMPLICATIONS FOR NURSING AND SOCIETY

Nurses are well placed to integrate new genetics/genomics knowledge and technologies into practice. They already have experience and expertise in caring for patients within their specialist area and will have established clinical networks. They will have an appreciation of patient experiences in their area of care so may have good insight into how genetic information may impact on this. However, there are challenges this added dimension could bring:

- **Time:** to acquire new knowledge and skills and to apply them in practice;
- **Nurses’ attitudes and awareness:** particularly with regard to confidence and a willingness to embrace change;
- **Maintaining competence:** in a rapidly advancing research field;
- **Organisational issues:** overcoming indifference among colleagues, fuelled by a lack of awareness of the relevance of genetics/genomics to the healthcare setting;

BOX 4. GENOMICS IN PRACTICE: DIABETES

At least seven different genetic subtypes of diabetes have been identified. Murphy et al (2008) described four single-gene subtypes that account for 1-2% of cases. These differ in terms of age of onset, pattern of hyperglycaemia, response to treatment and association with other non-pancreatic effects. They recommended a new disease classification based on the molecular genetic basis, as it gives a clearer indication of appropriate treatment:

- In the case of diabetes diagnosed before six months of age. For example, infants with genetic mutations affecting the ATP-sensitive potassium channel respond well to high doses of sulphonylureas such as glibenclamide, rather than insulin;
- In familial, young onset diabetes, people with the HNF1a or HNF4a mutations need low doses of sulphonylureas.

Murphy et al (2008) said knowledge of the genetic aetiology of diabetes enabled more appropriate treatment, better prediction of disease progression, screening of family members and genetic counselling.

NURSING IMPLICATIONS

Diabetes specialist nurses have a role in:

- Applying their knowledge and understanding of genetics to identify patients who might have a genetic subtype of the condition;
- Helping patients to understand the nature of their diabetes and any implications there might be for other family members;
- Supporting patients and carers who may be transferring from insulin to sulphonylureas;
- Liaising with specialist diabetes geneticists as appropriate.

BOX 5. ADVICE FROM THE ASSOCIATION OF GENETIC NURSES AND COUNSELLORS

AGNC members have the following advice for nurses on how to integrate genetics/genomics into practice:

- Familiarise yourself with how genetics fits into your area of practice and concentrate on this;
- Learn key warning signs to look for in the patient group you are most in contact with;
- Do not assume that all patients consider having a genetic condition in the family to be a problem;
- Be aware of ethical issues and family dynamics and do not assume everyone wants to be tested;
- Be honest and do not say to a patient that you understand their result when you do not. Speak to genetic counsellors who are always happy to advise;
- Know who your contacts are in the regional genetics service and make use of them;
- Never be afraid to pick up the phone and ask for advice.

● **Clarity:** being clear about how the professional role relates to genetics;

● **Genetics itself:** a complex subject.

Added to these challenges are the ethical issues raised by genetics/genomics and related technologies. Many of these relate to ownership and use of genetic information or technologies:

● **Confidentiality:** this can become complicated where a family member does not wish to share information that may be of relevance to others in the family. The nurse's role in upholding confidentiality could become compromised, especially if there is a bearing on reproductive decision making. Likewise, a genetic test for one person may reveal the genetic status of another close relative who may not wish to know this.

● **Discrimination:** a person's genotype has the potential to indicate their future health. The potential for discrimination based on genotype, with consequent effects on employment or insurance, raises concern, although in both the UK and US there is some protection against this happening.

● **Storage and access to stored DNA:** as DNA can reveal information about an individual's future health, there are some concerns about its storage and who might access it, particularly where it might be held on national and/or police databases.

● **Genetic engineering:** the use of genetic technologies to manipulate the genome, or to select for or against particular attributes, continues to be controversial, especially when used for sex selection or for non-medical enhancements.

● **Informed consent:** once a test result is known, it cannot be unknown, and such genetic information can be life changing. Informed consent for any genetic intervention is therefore essential. To support this, nurses need some understanding of the benefits and limitations of genetic testing and screening. Genetic

testing and screening of children raises particular concerns and geneticists adhere to a code of practice on this.

MANAGING EXPECTATIONS

The real life stories of Tony and Maria give an indication of what patients and families look for from healthcare professionals.

Burke et al (2007) interviewed 27 people with or at risk of genetic conditions as well as parents of children affected by such conditions. They asked about their experiences of receiving genetic information

from healthcare professionals and their views on this. The interviewees felt there needed to be greater awareness of genetic aspects of conditions among healthcare professionals and that their concerns should be taken seriously.

Communication skills were clearly valued, with information given without judgement, sensitively and at an appropriate pace, with an acknowledgement that some genetic information can be hard to understand. The use of terminology was highlighted. Interviewees wanted healthcare professionals to recognise that some terms can be confusing or even hurtful. Therefore, although "gene mutation" is the proper scientific term, when talking to patients and families, terms such as "gene alteration" or "gene change" are preferred.

The emotional impact of genetic information can be devastating and can have implications for wider family members. Feelings of guilt and blame may affect parents and grandparents and interviewees wanted healthcare professionals to be aware of this. In addition, it can be difficult to take in what may be complex genetic

BOX 6. THE UK NURSING COMPETENCES IN GENETICS

All nurses, midwives and health visitors, at the point of registration, should be able to:

1. Identify clients who might benefit from genetic services and information:

- Through an understanding of the importance of family history in assessing predisposition to disease;
- Seeking assistance from, and referring to, appropriate genetics experts and peer support resources;
- Based on an understanding of the components of the current genetic counselling process.

2. Appreciate the importance of sensitivity in tailoring genetic information and services to clients' culture, knowledge and language level:

- Recognising that ethnicity, culture, religion and ethical perspectives may influence clients' ability to use these.

3. Uphold the rights of all clients to informed decision making and voluntary action:

- Based on an awareness of the history of misuse of human genetic information;
- Based on an understanding of the importance of delivering genetic education and counselling fairly, accurately and without coercion or personal bias;
- Recognising that personal values and beliefs may influence the care and support provided to clients during decision making.

4. Demonstrate knowledge and understanding of the role of genetic and other factors in maintaining health and in the manifestation, modification and prevention of disease expression, to underpin effective practice.

5. Demonstrate knowledge and understanding of the utility and limitations of genetic testing and information:

- Including the ethical, legal and social issues related to testing and recording genetic information;
- And the potential physical and/or psychosocial consequences of genetic information for individuals, family members and communities.

6. Recognise the limitations of one's own genetics expertise:

- Based on an understanding of one's professional role in the referral, provision or follow up to genetics services.

7. Obtain and communicate credible, current information about genetics, for self, clients and colleagues:

- Using information technologies effectively to do so.

Source: Kirk et al (2003)

information at a stressful time, so being informed about where and how to access more information was seen as important.

Nowadays, genetics is rarely out of the media for long. Reports of gene discoveries and new treatments can be reported with more enthusiasm than accuracy, raising expectations about the availability and scope of genetic tests and cures.

Consumer genetic testing is available via the internet. Private companies will provide a genetic profile for a large range of conditions and characteristics. Nurses need to be able to evaluate such reports and manage patient expectations accordingly. For this, their own education in genetics/genomics is essential.

EDUCATION

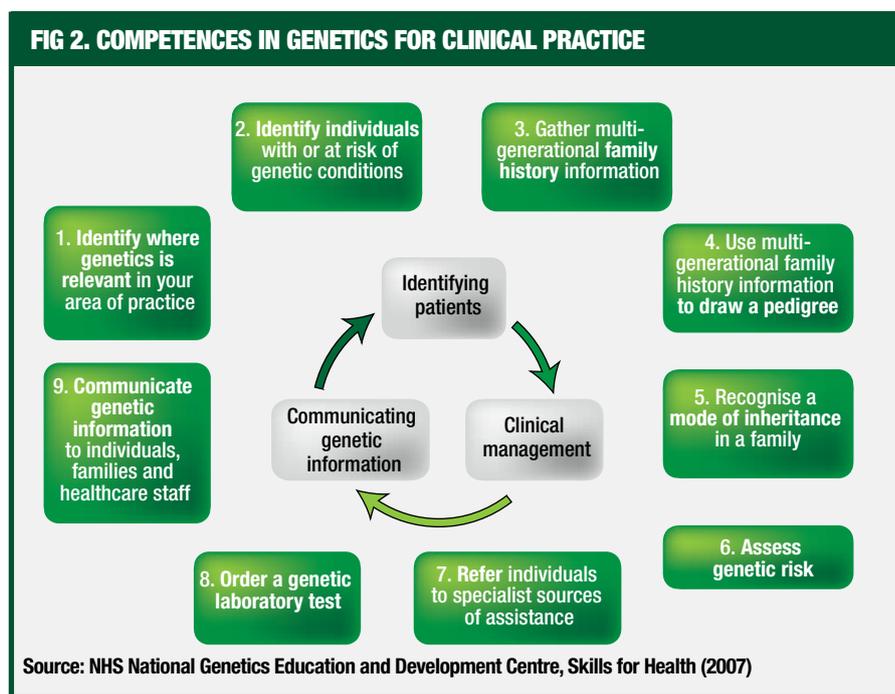
The knowledge, skills and attitudes in genetics that UK nurses need to support patients and address their concerns are set out in an education framework based on seven competences (Kirk et al, 2003; Box 6). Competences for US nurses have also been developed (Jenkins and Calzone, 2007).

The UK education framework is intended to guide pre-registration curricula in particular and forms a platform on which to develop knowledge and skills that qualified nurses need in their field of practice. In relation to this, and to inform the Knowledge and Skills Framework, the NHS National Genetics Education and Development Centre and Skills for Health (2007) have described nine genetics activities for healthcare staff outside the specialty of genetics, to support the patient pathway. These were accepted as national occupational standards in 2007. Not all the competences apply to all healthcare professional roles (Fig 2).

The NHS National Genetics Education and Development Centre was established in 2004 to support educators and practitioners

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as they look to integrate genomic healthcare into practice. It works with a network of genetic education facilitators to advise on education and training approaches, and to develop education resources.

NURSING LEADERSHIP IN GENOMIC HEALTHCARE

Nursing professionals have been notably active in leading change in genetics education and practice in healthcare. The International Society of Nurses in Genetics represents nurses from countries all over the world and has been a strong voice in influencing policy. National organisations such as the AGNC have sought to raise awareness of the contribution that nurses can make in genetic healthcare.

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CONCLUSION

The scope and potential of genomic healthcare has major implications for practice and brings its own set of challenges. Nurses are well placed to integrate these advances into practice to benefit patients, and must have adequate preparation to ensure they have the necessary knowledge, skills and awareness of the possible ethical issues. ●

An online question and answer resource on the role of genetics and genomics in health and disease is now available on nursingtimes.net. The resource has been set out to reflect the patient pathway with examples of questions that nurses might be asked. The resource is available at tinyurl.com/genetics-resource

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