Genomic medicine: the role of the nursing workforce

Genomics is the study of all the deoxyribonucleic acid (DNA) – the genome – of an organism. All living organisms, whether single-celled bacteria, multicellular plants, animals or humans, have DNA, which contains the information needed for an organism to grow, survive and reproduce. DNA is arranged into genes, a sequence of nucleotides which are code for proteins that are essential for building and repairing an organism. There are also non-coding regions, which influence how these genes interact. A genome includes both the genes and the non-coding regions.

Human DNA is made from four separate chemical bases, known as A (adenine), C (cytosine), G (guanine), and T (thymine), with the whole genome made up of more than three billion DNA bases. Every human’s genome is around 99.9% the same, but that 0.1% difference equates to around three million differences between one person’s DNA and the next person’s (Health Education England Genomics Education Programme, nd). Some of these differences in our DNA will have no impact on a person’s health, but other variations may cause a genetic condition, influence our predisposition to develop certain conditions, and can even affect how we respond to some drugs.

Over the past decade, transformative advances in DNA sequencing technologies have enabled a vast expansion in human genome analysis for the purpose of diagnosing and managing human diseases. The scope of genetic testing now ranges from analysing a single gene to using multigene panels – which can analyse from five to 100 genes known to be associated with the development of a condition or a collection of clinical symptoms – through to whole genome sequencing, where it is possible to determine the entire human genome (International Human Genome Sequencing Consortium, 2004).
Clinical Practice

Discussion

The use of this more advanced genomic testing has the potential to deliver tangible improvements for patients, including effective and quicker diagnosis of rare conditions, personalised treatment, better care for those with inherited conditions and cancers, and a clearer understanding of the underlying cause of diseases.

“The role of the nursing workforce will be to offer genomic testing with confidence”

Genomics in practice
In England, genomic testing, including whole genome sequencing, is now available for patients with certain rare diseases and cancers to enable better prediction, diagnosis and treatment.

In clinical practice, whole genome sequencing still requires the application of virtual panels for the analysis. As understanding of the genome and capacity in the NHS evolves, genomic testing will increasingly become part of routine care pathways for more conditions.

As a nursing community, we must be ready to adapt our current practice to reflect these innovations and ensure that our patients have access to the benefits that genomic testing can offer.

Nurses are often in an ideal position to offer support and advice to patients, whether they are living with cancer, long-term conditions or inherited genetic disorders, which need lifelong care. We work across a broad range of settings on the front line, are committed to our patients, and are typically strong and empathetic communicators, as described in the values set out in The NHS Constitution for England (Department of Health and Social Care, 2021). Nurses are vital to assisting the NHS in offering the latest advances, and it is important that we understand how genomics will affect our practice and the experiences of our patients.

Specialist nurses will work in partnership with the clinical team to offer tailored information and specific genetic testing relevant to the patient’s condition to inform treatment decisions. Senior nurses will also be able to support junior nurses to gradually acquire knowledge and understanding of genomic medicine to embed in their practice.

For nurses who have a lot of patient contact, gaining knowledge on the core principles of genomics may help them increase uptake of genetic screening, signpost service users to the right place to seek health advice, and could also help to address inequalities in access to genomic medicine services.

Genomic medicine is already embedded in some oncology nursing pathways. At present, genomic testing is available for inherited diseases or forms of cancer included in the National Genomic Test Directory (NHS England, 2022). Use of genomic testing could help the NHS reach its aim of diagnosing 75% of all cancers at stage 1 or 2 by 2028 (NHS England, 2019).

The North Thames Genomic Medicine Service (GMS) Alliance is working in partnership with key stakeholders to support the nursing workforce to acquire the skills and the level of knowledge to accomplish this mission effectively and efficiently. In some places, patients are driving forward conversations and we recognise the importance of patient involvement in establishing the best way to influence care pathways. Patient and public involvement is a key part of the North Thames GMS Alliance’s strategy, and collaborative work with patient groups is being developed.

Nurses’ role
With the right level of understanding of genomics, the support of their local trusts and the local systems, the role of the nursing workforce will be to offer genomic testing with confidence, helping to embed routine genetic testing in the NHS. This will mean:

- Working with our multidisciplinary teams to identify patients who could benefit from genomic testing;
- Discussing testing with patients;
- Requesting tests where indicated;
- Identifying patients who would benefit from a referral to clinical genetic services;
- Adjusting long-term care based on the results.

Evidence suggests that continuity of care positively affects health outcomes and patient experience (Lautamatti et al, 2020). Nurses are often well positioned to provide this and to build strong and trusting relationships with patients and their relatives.

Consequently, we can help our patients access better care, understand the impact of inherited disease on their lives, and give them the opportunity to plan for a better quality of life. Patients and their families will have access to more precise medicine that will give clinicians the ability to offer targeted and personalised treatment, based on their unique genome (pharmacogenomics).

As genomic medicine continues to evolve and additional tests become available that can benefit patient care, nurses can also help define how these tests can be brought into practice across their specialities. We are right at the beginning of the use of genomic medicine in the NHS, and there is a real opportunity for nurses to be part of service design. We have the chance to help improve guidelines across many areas of clinical practice and be international leaders (Tonkin et al, 2018).

By embedding genomic medicine in mainstream care and providing nurses (and other practitioners such as midwives) with appropriate training on ordering genomic testing to patients, practitioners will also be able to embrace their autonomy and clinical judgement to provide genomic services to patients more effectively and efficiently.

There are, of course, ethical issues to consider when discussing the widespread use of genomic medicine in the NHS. For example, we know that some patients will not want to know their risk for future conditions or their carrier status. There is also an ongoing national discussion around whether sequencing newborn babies for rare conditions beyond the current heel-prick test should become part of practice, and how this data and consent would be managed. As nurses, it is important that we bring our knowledge and perspectives to these conversations as the genomic medicine system is being designed and embedded in the NHS (Box 1). For now, where genetic tests provide actionable results that can help us improve patient care and outcomes, nurses are vital to

Box 1. Embedding genomics services in the NHS

- NHS England launched the NHS Genomic Medicine Service (GMS) in 2018 to harness the power of genomic technology and science to improve the health of the population and support the NHS to become the first health service in the world to systematically embed genomics into routine care
- The GMS is working to ensure that all patients have access to the genomic tests they need, regardless of where they live
- Genomic tests are commissioned at a national level
- The National Genomic Test Directory (NHS England, 2022) lists all available genomic tests as well as who can order them and the patient criteria for each one
making these accessible for patients in an equitable way and supporting them to make informed decisions.

GMS Alliances
We believe that nurses are a key element in the implementation of routine genetic testing in the NHS, but for nurses to assist with better use of genomics in mainstream healthcare they will need financial aid for training and support to acquire the appropriate skills and knowledge. Existing workforce pressures, a historical lack of secure funding on education and training, as well as inequalities in health, are all barriers that need to be acknowledged and addressed.

There is some support already being put in place for the nursing community through the establishment of seven regional GMS Alliances established in England in January 2021. One of the main purposes of the GMS Alliances is to provide training and education opportunities, which will help health professionals, including nurses, to increase their knowledge of genomics and keep up to date with how this may affect their clinical practice. There are also several training and education resources provided by Health Education England at national and regional levels, including continuing professional development programmes supported by the educational teams within each GMS Alliance (Health Education England Genomics Education Programme, nd).

At a service development level, nurses are also being consulted on how genomic medicine can work within their organisations (see Box 2 for an example of how this is used in practice). The GMS Alliances are working with healthcare organisations and nurses to improve current frameworks and pathways of care, tackling inequalities in accessing genomic testing, and promoting patient-centred care via the increased presence of patient and public involvement groups.

There is a need for further training opportunities to help prepare the nursing workforce for the future, and for ongoing consultation to ensure genomics is implemented in the right way. Powerful and influential nursing leadership is required in the UK to embed the genomics framework into nursing education pathways at all levels of practice and make sure the impact of genomics on the nursing workforce is understood and considered in decision making. This is to ensure that nurses can develop the core competencies and confidence in their understanding of genomic medicine required to meet the service users’ needs.

Conclusion
GMS Alliances are working with nurses, patients and the public to build trust in genomics and to support the multiprofessional workforce to use genomics safely and effectively. By embedding genomics into the mainstream health service, our aim is to deliver improvements for patients, including better and quicker diagnosis of rare conditions, personalised treatment, and care for those with inherited conditions and cancer, as well as building a better understanding of the underlying cause of many diseases. To achieve this, nurses need support to develop genomics literacy and begin to use genetic testing within their care pathways. NT

Box 2. Lynch syndrome: an example of genomic medicine in current practice

- Lynch syndrome is an inherited condition, which results in an increased risk of colorectal cancer (Hegde et al, 2014) and, in women, an increased risk of endometrial cancer (Li et al, 2021). It is estimated that more than 200,000 people are likely to have this condition in the UK but only 5% of people have been diagnosed (Bowel Cancer UK, 2018). The remaining 95%, unaware that they have the condition, do not seek health advice or enrol on screening programmes.
- St Mark’s Hospital in London is a reference centre for genomic mainstream healthcare for colorectal cancer, polyposis and Lynch syndrome, providing long-life support and advice. Patients referred to this centre receive assessment and personalised advice from a multidisciplinary team (St Mark’s Hospital, 2019).
- Patients are seen by nurse specialists who have a good knowledge and understanding of genomics and who can take their family history and order tests where needed. They liaise with gastroenterologists to offer targeted therapies and personalised treatments. The team ensures that patients are followed up long-term and receive the advice and care they need to remain healthy.
- The North Thames GMS Alliance is supporting partners to implement this model across the region.

- If you are a nurse or student nurse and have an interest in genomics, please do not hesitate to contact us for more information. If you would like to further your career by completing a masters in genomics or are interested in increasing your genomics literacy, please contact us for guidance. You can reach the North Thames GMS Alliance at nt-gmsa@uclpartners.com.

References