

Genomic medicine in blood cancer

APPG on blood cancer

1. What are the major benefits and challenges of genomic medicine for individuals with blood cancer?

Benefits:

- More accurate diagnosis: Genomic medicine may give us a better understanding of the causes of blood cancer and can provide information about a patient's condition to inform their prognosis and treatment.
- More personalised medicine: Treatment can be more easily personalised using information from genomic testing which may increase treatment effectiveness and, possibly, reduce toxicity. Genomic medicine can be used to monitor how a patient responds to treatment and accurate diagnoses will enable more effective selection of stem cell transplant donors.
- Identifying risk: Genomic testing can identify predisposition to blood cancer which may be relevant for family members.
- Prevention: Genomic screening could allow blood cancer to be detected and treated at an earlier stage compared to existing care pathways.

Challenges:

- Fast-changing nature of genomics: Genomic data is complex to interpret and is changing very fast. For most applications of genomic medicine, the clinical benefit remains unproven. It is often hard to determine the nature of different genetic variants, and what impact they might have on disease. For many variants there is a lack of targeted therapies, so that even when a genetic cause has been identified, the patient may not benefit from the more accurate diagnosis.
- Workforce capacity: Considerable clinical expertise is needed to use this new technology wisely and safely in clinical practice. It also requires additional resourcing as genomic medicine will add an extra time burden to existing care pathways. A [recent survey from the Royal College of Pathologists](#) found that haematologists are already finding it increasingly difficult to undertake vital diagnostic work.

- Over-reliance on genomic testing: Genomic testing should not replace existing clinical and laboratory methods but should be used appropriately alongside existing diagnostic methodology.
- Effectiveness of screening: When genomic testing reveals a predisposition to blood cancer in family members, the likelihood and timescale of developing the disease are often unknown. In this situation it is uncertain what screening or surveillance of healthy people is appropriate without more research into genetic epidemiology.
- Informed decision making: Patients need help making informed decisions about genomics. There is no consensus on how to manage the output from genomic tests, especially in healthy people. Unlike other blood tests, the result is essentially the same throughout the life course, but may have no relevance for several decades, or only for future generations. For example, knowing that a child is at an elevated risk of blood cancer as an adult may not be a helpful result for parents to receive if there is nothing that can be done about this by treatment or interventions until adulthood.

2. The NHS Long Term Plan aims to embed genomic medicine across the NHS by 2029. What would such a service look like (eg a genomically literate workforce / routine use of whole genome sequencing across rare disease and cancer)?

Given the complexity of genomic medicine and the rapid rate of change in this sector, it's difficult to predict what the NHS with genomic medicine embedded would look like in 2029. The [Academy of Medical Royal Colleges produced a statement on genomic medicine](#) that outlines ten key principles to maximise the benefit and minimise the harm of genomic medicine in the NHS.

What is certain is that to effectively embed genomic medicine across the NHS will require a genomic-literate workforce. This not only includes clinicians but also scientific and nursing staff. Staff need to be well equipped to interpret and explain genomic findings to patients and support them in accurate decision making.

While genomic medicine may move into use by a wide range of health professionals, tests relating to the diagnosis and treatment of major diseases (like blood cancer) should remain within the realm of professionals with established expertise in managing and treating those diseases.

Laboratories should be well equipped and resourced to provide genomic services with streamlined consolidated testing across each genomic laboratory hub. Experienced laboratory clinicians and scientific staff are essential to deliver this service.

Basic information about genomics, its limitations and potential should be common knowledge, and patients should be supported to make informed decisions about their care based on accurate genomic information.

3. What needs to happen for us to transition between where we are at present (question 1) and where you expect us to be by 2028/29 (question 2)?

At present genomic testing means additional time and resources in an already-stretched system. To effectively embed genomic medicine in the NHS there must be considerable investment in workforce education and capacity in both laboratory and clinical services.

Key to creating a genomic-literate workforce is compulsory genomics education for all medical and nursing students, and ongoing education for already qualified staff. This will ensure that all staff will be well equipped and educated to feel comfortable interpreting genomic findings and supporting patients in accurate decision making.

Strong investment in genetic epidemiology is urgently needed to establish the likelihood of genetic variants causing blood cancer and other illnesses. One priority would be to establish, in the UK Biobank and other large datasets, the likelihood of genetic variants causing blood cancer to better inform decisions for patients and their families. Without this it will not be possible to take advantage of genomic medicine to accurately personalise treatment for patient benefit.

There also needs to be research into the clinical benefit of using genomic data compared with standard care pathways to determine the clinical utility of genomic medicine.

Patients and the general public should be educated on the benefits and limitations of genetic testing, which will require engagement with patient groups, production of educational literature and educational events.

4. Please share any other thoughts or reflections you have on the subject of the inquiry. Example: the availability of patient information, the role of stakeholders in maximising the benefits afforded by genomic medicine, etc.

We fully support the topic of this inquiry and would urge similar inquiry into applications of genomic analysis in other branches of medicine.

About the Royal College of Pathologists

The Royal College of Pathologists is a professional membership organisation with more than 11,000 fellows, affiliates and trainees, of which 23% are based outside of the UK. We are committed to setting and maintaining professional standards and promoting excellence in the teaching and practice of pathology, for the benefit of patients.

Our members include medically and veterinary qualified pathologists and clinical scientists in 17 different specialties, including cellular pathology, haematology, clinical biochemistry, medical microbiology and veterinary pathology.

The College works with pathologists at every stage of their career. We set curricula, organise training and run exams, publish clinical guidelines and best practice recommendations and provide continuing professional development. We engage a wide range of stakeholders to improve awareness and understanding of pathology and the vital role it plays in everybody's healthcare. Working with members, we run programmes to inspire the next generation to study science and join the profession.