ISSUE 8

Pharmacogenomics

July 2025

Looking ahead, the NHS 10 year plan and beyond

Welcome,

As I was writing this, the NHS 10 year plan was published, with DNA making the front page, a clear statement that genomic medicine will play an important part in how we prevent disease, diagnose and treat patients. Within this issue, I have focused on three areas highlighted within the plan. The first relates to cancer, where the role of blood tests, also known as liquid biopsies, can help diagnose and identify which patients need targeted treatments. In many cases this approach is quicker than using a tissue sample. Nishat Damji, lung cancer pharmacist from The Royal Marsden Hospital and one of our genomic champions provides an informative overview and the impact it is having within non-small cell lung cancer.

Infectious disease is also having its own revolution and Daniel Murphy, another of our pharmacy champions, has highlighted the great work being conducted in the world of metagenomics, a method used to identify respiratory pathogens and improve diagnostic approaches within intensive care units. The work mentioned is on-going and not routinely available on the NHS, but indicates the progress being made to diagnose infectious diseases swiftly.

Thirdly, pharmacogenetics also made it into the plan, with emphasis on making sure more patients receive the right drug, at the right dose, at the right time. The aim over the next 10 years is to utilise pharmacogenetics for commonly prescribed drugs such as antidepressants, proton pump inhibitors, opioid analgesics, statins and clopidogrel. We are still a while away from a panel test approach, but a lot of work is happening to prepare for future implementation, and I have provided a few examples of some important work happening across the NHS and UK.

Finally, and most importantly, I want to say a massive thank you to Raliat Onatade, who stepped down this summer as our North Thames GMSA Chief Pharmacist and has been a huge support us. Raliat has been instrumental in raising the profile of pharmacy within genomics, which has included championing the role of pharmacy technicians.

This month, Rob Duncombe, has taken up the reins, welcome! 😊

Contents

Metagenomics and ICUs, impact on antimicrobial stewardship?

Liquid biopsies

The approach in NSCLC

Updates in PGx

Preparing for the future

NHS 10 year plan

Pharmacy Champions!!!

Dharmisha – Consultant Pharmacist, NTGMSA

New approach to may improve outcomes in intensive care for patients with respiratory infections – *Daniel Murphy*

A recent pilot study has demonstrated the potential of a novel gene-based diagnostic method that may lead to improved infection treatment and may help address the growing problem of antimicrobial resistance. Professor John Edgeworth and his team introduced a new test to three intensive care units (ICUs) in Guy's and St Thomas' NHS Foundation Trust in London, and between November 2021 and March 2022.

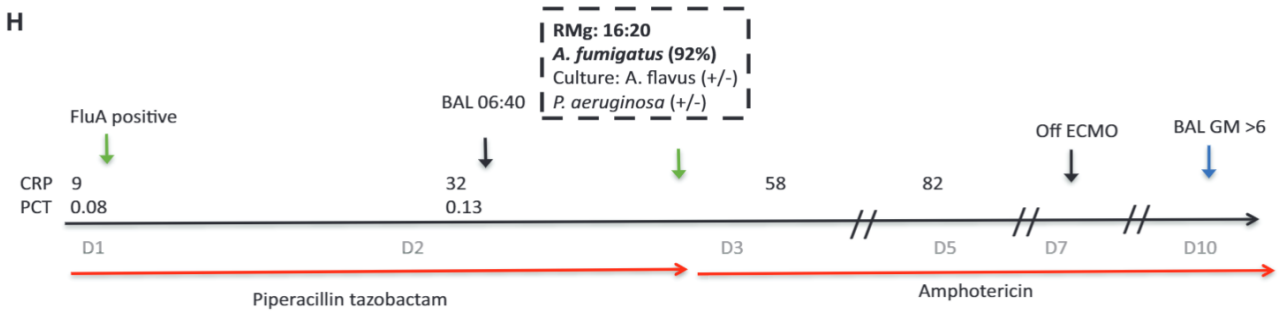
The method is known as **metagenomics**. This is a genetic testing technique that analyses the genetic material of all the organisms - typically microbes – present in a clinical sample. Unlike traditional culture-based methods, which take several days to provide results, metagenomics can deliver same-day findings. It is also an agnostic test, meaning it does not target specific pathogens but instead scans for all possible infectious agents. Importantly, it also detects antimicrobial resistance genes, supporting more accurate antibiotic selection.

This **pilot** focused on in ICU patients with respiratory infections, which are often complex and involve multiple pathogens – making them ideal candidates for this comprehensive testing approach. These cases also carry a high risk of drug-resistance, so early resistance detection is essential for guiding treatment decisions. Metagenomics was carried out in 128 samples from 87 patients. Same-day results were achieved in 87% of samples, with an average turnaround time of seven hours – a significant improvement over conventional diagnostics.

In 50% of cases, test results led to changes in prescribing, including switching to more appropriate antimicrobials or de-escalating treatment when antibiotics were no longer required. In 30% of cases, results provided reassurance in continuation of existing therapy, or helped exclude infection altogether, prompting other diagnoses such as inflammatory conditions.

This study highlights the value of metagenomics in **guiding antimicrobial prescribing and enhancing antimicrobial stewardship**. It also shows the potential for enhancing infection control and informing public health decisions. However, larger multi-centre studies will be required to validate these findings and further define the role of metagenomics in clinical care.

Although still in the **evaluation stage**, metagenomics has the potential to transform infection management in ICUs. However, it is not yet widely adopted in general wards, where conventional tests are often sufficient for less severe infections. Moreover, metagenomic testing remains costly and resource-intensive. As healthcare professionals, it is increasingly important to understand emerging diagnostic tools that may significantly enhance patient outcomes and promote responsible antimicrobial use. ***Reference:*** *Charalampous T, Alcolea-Medina A, Snell LB, et al. Routine Metagenomics Service for ICU Patients with Respiratory Infection. Am J Respir Crit Care Med. 2024;209(2):164-174.*



Example of patient with influenza with secondary invasive aspergillosis prompting urgent treatment with metagenomics, image H directly taken from Charalampous et al. (2024).

Blood biopsies in lung cancer, improving faster to treatment – *Nishat Damji*

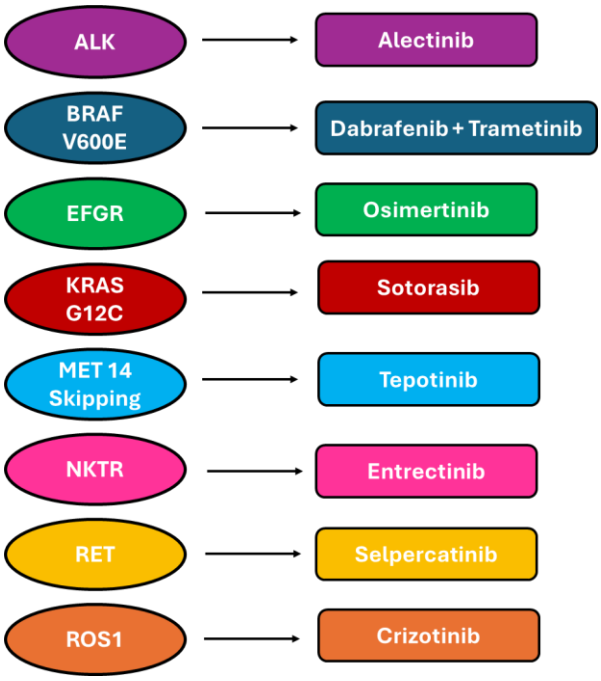
In April 2025, the NHS officially approved and began rolling out circulating tumour DNA (ctDNA) testing—a form of "liquid biopsy"—across England for patients with suspected non–small cell lung cancer (NSCLC). With this approval, NHS England became the first national health service in the world to adopt a "blood test-first" diagnostic pathway before pursuing conventional tissue biopsies.

A ctDNA test identifies tumour-derived DNA fragments shed into the bloodstream, enabling rapid detection of key mutations which drive cancer cell growth (e.g., *EGFR, ALK*, *BRAF*, *KRAS*, *MET* exon 14 skipping, *ROS1*, *RET*, and *NTRK*) via a multi-target panel. Pilot data from approximately 10,000 patients across 176 hospital trusts showed results could be delivered 16 days earlier than tissue biopsy routes. This significantly expedites onboarding to precision therapies and sparing some patients from unnecessary additional biopsies and chemotherapy. Clinically, the impact is profound: patients can access targeted treatments faster; tumour profiles are captured even when tissue samples are inadequate; quality of life improves; and health inequalities diminish thanks to the test's non-invasive nature. Financially, the service is expected to save ~£11 million annually in lung cancer care alone.

Technology and infrastructure have been embedded into the NHS Genomic Medicine Service and the National Genomic Test Directory, with ~1,600 patients already tested since April, and ~15,000 eligible patients per year now benefiting. Importantly, the blood test has shown comparable accuracy to tissue-based molecular diagnostics, while reducing the risk of biopsy-related complications.

In the future it will also enables serial monitoring, potentially helping clinicians assess treatment response and detect emerging resistance mutations earlier.

Within the NHS 10-Year Plan, ctDNA testing stands out as a flagship innovation: a cornerstone of its vision for personalised, genomics-driven care—delivering faster diagnoses, smarter treatment decisions, fewer invasive procedures, better patient outcomes, and cost-effective operations. Its national adoption marks a paradigm shift in how lung cancer is now diagnosed and treated within the UK.



Examples of drugs which can be matched to specific NSCLC mutations.

Pharmacogenetics – preparation for implementation

– Dharmisha Chauhan

Implementing pharmacogenetic testing especially through the primary care route requires consideration to guidelines, sharing of data and education and training. Without these key elements, variations in practice will emerge and would certainly delay implementation across England. Given that pharmacogenetics has been highlighted as a preventative genomic medicine measure within the new NHS 10 year plan, what is being done to tackle these areas?

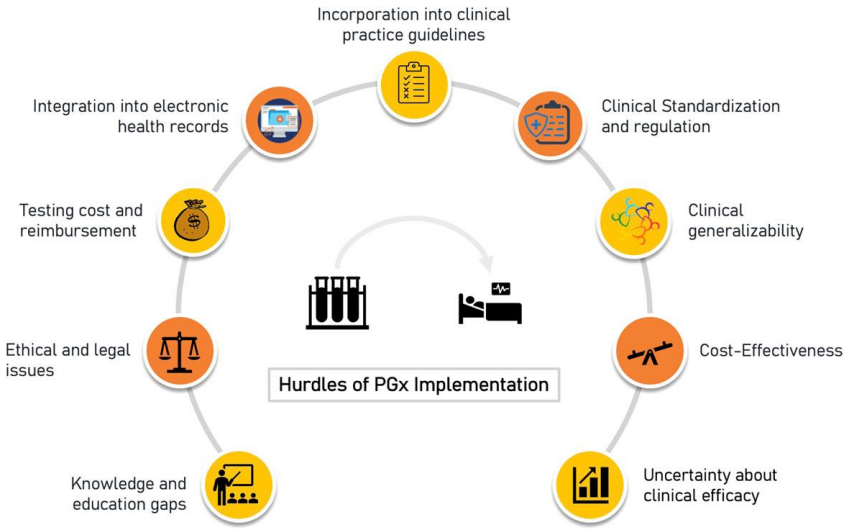
Supporting prescribers to prescribe safely

Internationally, pharmacogenetics guidelines are available from Clinical Pharmacogenetics Implementation Consortium and the Dutch Pharmacogenetics Working Group. These are incredibly useful resources but do not always reflect UK clinical practice. To address this gap, two key projects have emerged. The first was launched in January 2025 via a group led by Liverpool University via the leadership of Professor Sir Munir Pirmohamed to explore the potential of pharmacogenomics via a programme called Centres of Excellence for Regulatory Science and Innovation, also known as CERSIs.

CERSIs aim to promote innovation in regulatory science and work in partnership with Medicines and Healthcare products Regulatory Agency (MHRA), Office for Life Sciences, and the Medical Research Council. There are seven CERSIs within the UK. Within the Pharmacogenomics CERSI group, one key project is the development of UK based guidelines, which will reflect UK clinical practice and aims to provide guidance on drug dosing, which genes and variants should be tested, health economic evaluations and research recommendations.

The second project is being led by UK Clinical Pharmacy Association (UKCPA) by the Genomics Community. Inspired by the trusted Renal Drug Handbook this work led by Lucy Galloway, consultant pharmacist from South East GMSA, are producing easy to access pharmacogenetic drug monographs to help guide clinical decision making within a fast-paced clinical environment.

It should be noted that these guidelines can include genes which may not be routinely commissioned on the NHS but are a great starting point to plug the gap on guidelines more reflective of UK practice.



Hurdles within pharmacogenetics, focus within the UK includes all barriers shown but strides are being made to UK specific guidelines, digital solutions and education and training needs. Image directly taken from Maruf et al. Psychiatry and Clinical Neurosciences Reports. 2022;1(2).

Data sharing

Once a pharmacogenetic test has been conducted, it only needs to be done once. So how can we share the results across different healthcare sectors, especially when the variant identified impacts drugs more commonly prescribed e.g., antidepressants? An approach taken by NHS England, is to develop a Unified Genomics Record (UGR). The aim is to develop a single point of access to a patient's genomic record to help guide treatment plans. This will prevent data transfer across different NHS systems enabling a streamlined way of accessing genomic data.

Image from DNA Data Storage | Kilobaser

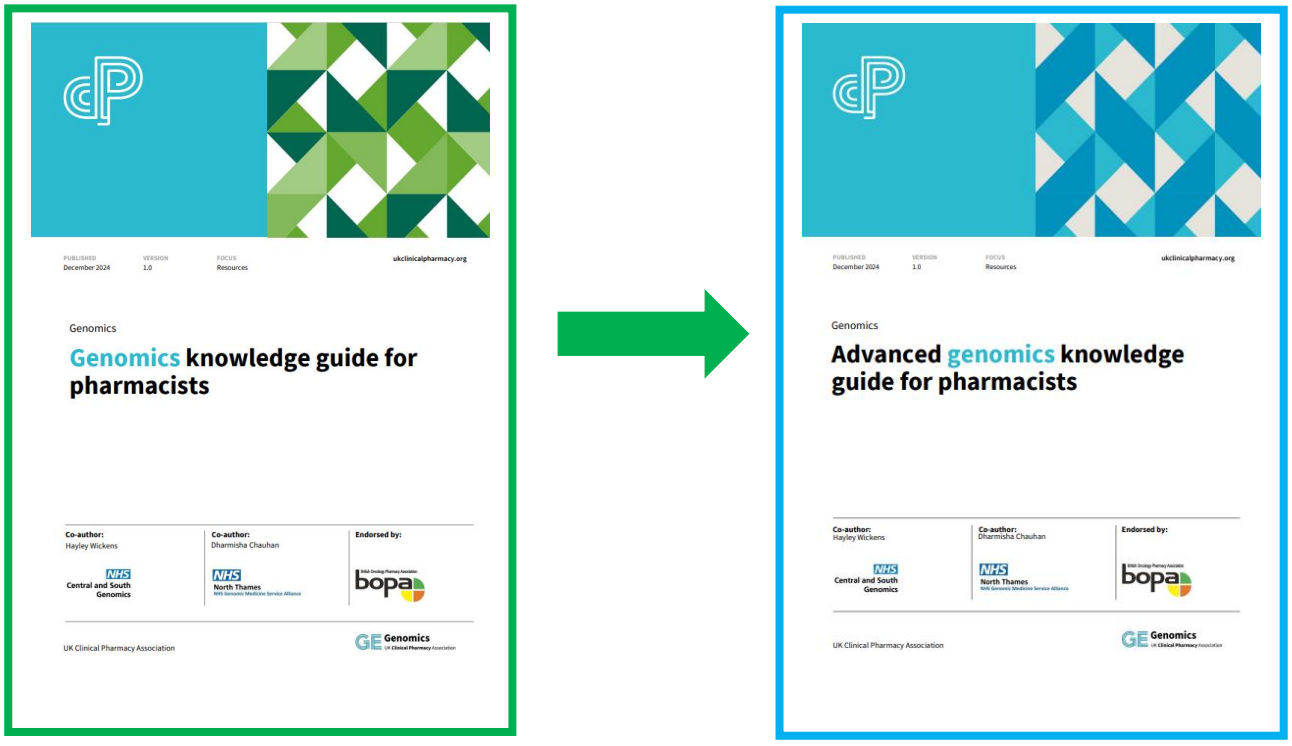
Education and training – development tools

Tools and resources continue to be developed for all healthcare professionals from foundational knowledge through to a taught MSc Genomic Medicine programme. But we now have tools to help pharmacists identify gaps within their genomics knowledge, via "Genomic Knowledge Guides".

These guided have been created in collaboration with UKCPA, Central and South GMSA and North Thames GMSA. Two have been published, one for all pharmacists and the second for pharmacists who work within specialist fields and want to develop their knowledge beyond the basics. There are plans to develop similar tools for pharmacy technicians.

The tools are split into 3 areas and can be found here.

1. Fundamentals of genomics
2. Application of genomic medicine
3. Genomic skills and behaviours for pharmacists



The NHS 10 year Plan

The 10 Year Health Plan for England sets out 3 shifts:

1. Hospitals to community
2. Analogue to digital
3. Treatment to prevention

Genomic medicine has been weaved into all three but one element which jumped out was the need to continue to train genomic champions to develop the knowledge to support local and equitable implementation of genetic testing across the GMSAs.

We have a strong Pharmacy Genomic Champion Network within North Thames GMSA and if you are interested in joining, please do contact me via dharmisha.chauhan1@nhs.net. The network welcomes everyone from novices to experts, and I share details of upcoming educational events and genomic opportunities from new roles to research projects.

Thank you and welcome!

This summer we have said goodbye to Raliat Onatade, who has stepped down from her role as our North Thames GMSA Chief Pharmacist, but Raliat will still be involved in the world of genomics within other national leaderships teams, so it's not a complete farewell 😊 Taking up the reins is Rob Duncombe, who also works as the Chief Pharmacist for The Royal Marsden Hospital.

Next issue…

I hope you all enjoy this issue, and if you would like to contribute or share your work within genomic medicine through this newsletter, please do get in touch!

For the next issue, we take a dive into *HLA* testing and the role it plays to prevent serious adverse drug reactions.

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