Discovery of the genetic code, with its unique signature for each living organism, has until recently had limited application in the routine clinical laboratory and little impact on the man in the street. However, this is set to change as advances in genomics technology make it more accessible and as public awareness of its benefits grows.

Looking at DNA is highly specialised but technological advances including automation, miniaturisation and the cost reductions that occur with scalability have moved the utility of genetic information out of research and into mainstream clinical laboratories. Affordable high-throughput genomic technologies enable rapid provision of results and expanded commercial platforms. This process is likely to accelerate, challenging the capacity of the clinical laboratories with new work processes and a requirement to store massive amounts of genomics data and maintain their integrity.

When the Human Genome Project was completed in 2003, the next obvious question was “what can we do with this information?” Finding the causes of some common hereditary diseases (e.g., cystic fibrosis) was a logical starting point, but this has since been broadened to include many other common conditions. Research into various cancers and the push to find suitable treatments for other previously difficult-to-treat diseases has also been targeted for investigation. Preventative strategies such as the surgical removal of non-essential, cancer-prone tissue (e.g., breast tissue) in familial risk groups has now become popular. So, if it is written in our genes, we can now know and do something about it.

**Genomics and personalised medicine**
The interest in using genomics for personalised medicine has expanded rapidly and even health funders in some countries are joining the discussion, as they believe there is a cost-benefit to testing for preventative health before expensive curative medicine is required. The caveat is the ongoing ethical debate around who should have access to what information, the potential for discriminatory use, and the unintended consequences of knowing for the patient. Not surprisingly, there is also an increased sensitivity to, and an awareness of, security in the population in relation to genetic testing.

However, many of the everyday genomic tests performed increasingly in clinical laboratories will analyse the genetic make-up of tumours, not patients. Laboratories will help to deliver personalised medicine by identifying a cancer down to its genetic code.

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so clinicians can target the most appropriate treatment. Using genomic analysis, many tumours can now be subtyped and matched to treatments that yield the best clinical outcomes, and research is ongoing for an ever-expanding range of cancers.

**Genetic testing for pathogens**

There is also the emergence of genetic testing of bacteria and other infectious agents, with dramatic improvements in turnaround times. With genetic testing, traditional microbiology tests that take 48–72 hours can be reduced to hours so that prompt, targeted treatment can be assured. In the case of slow-growing mycobacterial organisms, there has been a massive reduction in turnaround time as tests that used to take up to seven weeks are now performed through genomics in two hours.

As testing costs come down, there will be a fundamental change in how pathology deals with tumours, microbiology tests, and even blood grouping. DNA screening tests for an individual that used to cost hundreds of thousands of dollars is now approaching US$1000. And while genomic tests for cancers, for example, are still more expensive than this, the higher success rates of treatment can result in a compelling cost-benefit analysis.

Advances in technology have made DNA sequencing platforms, once solely the preserve of research departments, accessible to routine clinical laboratories. Hospitals that used to send colonic tumours away to specialised laboratories are now asking “why can’t we do that here?” As medical care decisions become more precise and personal, laboratories increasingly will be required to provide genomic testing services, and this will also influence their choice of information systems.

**Laboratory systems challenges**

The need to incorporate genetic information about an individual into their electronic medical record (EMR) will no longer simply be nice to have, but necessary to deliver a clinical service. Dealing with the data in a way that meets the exponential increase in demand for secure storage as well as ready accessibility for analysis is also challenging clinical laboratory information management systems (LIMS).

Some of the key issues are already accommodated in current laboratory processes, including security of patient demographics, accurate identification of tissue and specimens, long-term storage requirements of specimens, and some analytics. However, traditional LIMS were not designed to cope with the new bioinformatics demands for storage or analysis, as a typical genomics test can generate a file 30–70 megabytes in size.

Risk mitigation also requires traceability of all aspects of the process. Ensuring full audit ability and adherence to standard operating procedures (SOPs) are crucial criteria for a laboratory management system. When laboratories perform a genetics test, it must retain evidence of who handles the sample at every point of processing, how it has been stored, and the complex workflows. Many current LIMS will not handle this type of complex requirement for chain of custody.

**New breed of system required**

Providing security of information and allowing access by authorised healthcare workers via the EMR provides more complexity. All these considerations need to be part of any evaluation of new systems as legacy applications have not been designed to cope with this disruptive demand. This necessitates a new breed of system, termed a laboratory business management system (LBMS; InterSystems). Features of an LBMS required to support genomics testing in clinical laboratories include:

- support for connected care models, with a contiguous pathology patient record integrated within the EMR, including genomics data
- configuration and enforcement of SOPs, including sample preparation and chain of custody for genetics testing, with full audit ability within the system
- access to virtually unlimited amounts of low-cost data storage, while maintaining high levels of system performance, fully controlled and secured within the system
- ability to perform complex analytics on genomics data without the need to purchase or integrate with third-party solutions

The nature of laboratory business is changing dramatically and genetics testing is one of the major drivers. Genomics testing is becoming more prevalent, and its growth will accelerate as diagnostic and therapeutic use becomes ever more evident.

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*Gene Elliott is Physician Executive at InterSystems.*