Genetic medicine – just over the horizon?

Genetic medicine promises a revolution in health care. But major obstacles remain, not least the complexity of extrapolating clinical practice from organic molecules. But the INFOBIOMED team hopes to link biological and medical informatics in a bid to make genetic medicine a reality.

The day you are born, your birth certificate carries a detailed genetic profile showing your predisposition to allergies and certain diseases, your health risks and information useful to minimise your chances of falling sick. With all this information so early on, you and your health professional can take steps to minimise the effects of disease, before age-related illnesses begin to develop 60 years on.

In the meantime, if you do fall sick, doctors can better diagnose and tailor treatment and drugs to your exact genetic profile, with a greater chance of success. The era of genetic medicine is upon us.

From research lab to patient benefit

That scenario illustrates the promise of genetic medicine. Yet problems remain before the genetic revolution can move from research labs to patient benefit. The human genome is immensely complicated, and extrapolating clinical practice from genetic data is fraught with problems. Medical doctors and molecular scientists talk a different language; their expertise has a different focus and their computers can even use incompatible databases.

Which explains why one project team is working on this very problem. The partners in INFOBIOMED are attempting to link biological informatics with medical informatics, to match observed laboratory phenomena with clinical outcomes.

"Bioinformatics works mainly with data on a molecular and DNA level, while medical practice is more centred on 'higher' data levels for human organs or tissues. There's a big gulf between the two," notes project manager Carlos Diaz of Fundació IMIM in Barcelona. "We want these two information systems to develop a common ground between what happens in your DNA and what happens with disease."

INFOBIOMED has already had a huge impact since its beginnings in 2004. The doctors, scientists and IT experts involved in the project are now talking a common language and working on shared problems, something which is a major step forward.

New wave meets punk rockers

"When we first held meetings with all these specialists, it was like new wavers talking to punk rockers. They couldn't relate to each other. But they do now. That's the value of a multidisciplinary Network of Excellence," Diaz says.

The project also put students from different disciplines into a house together during one week, split them into teams and gave them problems to work on jointly. "It was like a scientific 'Big Brother', as all the prejudice and barriers posed by different languages and traditions were exposed, and it taught us a lot about getting different specialists to work effectively together," he remarks.

But that's not the most difficult problem. Some diseases can be associated in a relatively simple way with a specific genetic anomaly. But most diseases depend upon a huge variety of genes, plus many other factors – lifestyle, environment, even chance. The problem of understanding the mechanisms of disease is extremely complex.

"It is difficult to extrapolate clinical practice from genetic information, but we hope to provide a feedback loop, through the combined informatics systems – so-called 'biomedical informatics' – so that doctors and geneticists can find links between genomics and medical outcomes," notes Diaz.

Over time this will develop a picture of the relationship between the two. It could also mean that

drugs are tailored specifically to individual patients, or groups of patients, with greater odds on a successful outcome.

Major strides towards genetic medicine
But that is for the future. Yet INFOBIOMED has already taken some major steps toward that horizon. It has produced reports on the state-of-the-art in biomedical informatics, and developed databases and software that link both bioinformatics and medical informatics in a meaningful way.

The project partners have also developed pilot applications in four medical areas: pharmaco-informatics, genomics and infection, genomics and chronic inflammation, and genomics and hereditary colon cancer. And these pilots aim to deliver limited clinical results as proof of concept of the benefits of discipline integration.

"Using advanced biomedical informatics approaches, the pharmaco-informatics study found a previously unsuspected bio-molecule that could play a key role in the development of the Complex Regional Pain Syndrome, a painful disorder affecting one or more extremities of the body, that ultimately can even lead to amputation. The results obtained could serve as basis for new drug treatments, and could also have a relationship with other inflammatory diseases," says Diaz. "The method followed in the research could be applied to other complex diseases as well."

Looking for partners
INFOBIOMED ends in July 2007, but Diaz hopes they will find a way to continue the work. "We're currently looking at options – we could for example start a professional society to run a journal, with regular meetings to move biomedical informatics forward," he says.

"We would also be interested to hear from companies, investors or partners who have an interest in the area." Diaz suggests that many organisations could find the project results useful – health, pharma or informatics companies, or institutes with specialist departments in this field. The project piloted four medical fields, but others could benefit from the same approach.

A role for ethics
There are ethical issues too. "Ethics has a very important role to play," warns Diaz. "Who can access your genetic information, what impact will it have on your rights, how you keep it secure – these are all problems that only add to classical security concerns in clinical settings, and that will evolve over time."

But he believes that the promise of the field will triumph. "We could eventually see a revolutionary era in healthcare, where people are provided with optimised healthcare when they fall sick, through treatments that are specifically tailored to solve the problem."

As an added bonus, in the very long term genetic medicine could mean cheap healthcare, as prevention begins to trump cure. The era of molecular genetic medicine could be just over the horizon.

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