What are biomarkers and why are they important?

Tim Barrett
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One of the global health priorities of the International Rare Diseases Research Consortium (IRDiRC, www.irdirc.com) is on to find new treatments for the 80% of rare diseases that currently have no cure or means to treat. We know of over 8,000 rare diseases, so this is a really big challenge. As everyone knows, it is a huge task to come up with a treatment, then get it approved so that patients can have it in the clinic. Once you have come up with a treatment, people ask: 'how do you know it will work?'.

The best example I can think of is anaemia. You may be feeling tired and pale with a lack of energy, and go to your doctor – she takes a blood test, tells you that you are anaemic, and prescribes you some iron medicine. You take the medicine, and after a few weeks you feel better so your doctor invites you back for another blood test, says your blood count has come up, and you can stop the medicine. That blood count is what we call a 'biomarker' – something that we can measure to see if a treatment has worked. The doctor needs it to decide when to tell you to stop taking your medicine. Without it, she doesn't know whether you feel better because the medicine worked, or because you are eating better, or just because the sun has come out!

A biomarker is anything that we can measure, to see if a treatment is working. If the treatment is to help you lose weight, the biomarker is your weight. If you have a metabolic problem where a toxin builds up in your blood, then the biomarker is the level of toxin in your blood. It can even be a change seen on X-ray or brain imaging scan.

Biomarkers become especially important when you are treating a disease that takes months or years to progress- if you start taking a treatment, you want to know it is helping you, without waiting for years to find out if you are going to develop any complications of the disease.

When we study a medicine to see if it will treat a disease like Wolfram, it may take at least two years, or probably longer, to tell if the medicine has stopped the disease getting worse. If after 2 years, the person's disease is still getting worse, then the medicine has not worked, and he/she has to start the process from the beginning with another treatment.

You might ask – why not try several different treatments at once? That's a good question but there are two problems: firstly, the treatments may not work; or only one of them may work and the others are unnecessary; and then every treatment has side effects, which may get worse when you mix them with other medicines.

So why might biomarkers help? Well, to use the example of anaemia again: if you have something that you can measure, like a blood count in anaemia, you can quickly find out if the treatment is working, even before the person who was anaemic feels any better.

In Wolfram, if we ask people to take a treatment for the disease, we want something we can measure that will tell us quickly whether the treatment is working or not, without having to wait 2 or more years to tell if the disease has stabilised.

Biomarkers become really useful in clinical trials of a treatment: we really need a biomarker that will tell us whether the treatment is working within 6 months; so that if it is not, we can stop the trial, and switch to another treatment.

This is why we will be asking people to donate blood samples during our upcoming clinical trial. We want to make the samples available for the wider research community, and support the international effort to find biomarkers to measure the effectiveness of treatments in Wolfram.

The consortium

Left : Prof. Timothy Barrett,

Right, 1st line: Prof. Melanie Calvert, Dr Kristian Brock, Dr Zsuzsanna Nagy

Right, 2nd line : Prof. Richard Sinnott, Dr Anita Slade, Dr Ben Wright