

Using Science to Improve the World

Report of the ALDES autumn conference fringe meeting

“The human genome, scaled up 100,000 times, can be likened to this thread of cotton” said Sir John Sulston, 2002 Nobel prize winner for medicine, as he drew red thread from a cotton reel. “At this scale it would be 60 miles long but still extremely thin and in fact coiled back and back on itself into the famous double helix configuration.”

The human genome contains a 3000 million ‘letter’ code. It is that code which has now been read. The data can be held on a single CD, a copy of which Sir John held up. The code is likely to provide the pathway for discovery of the reasons for differing strengths and weaknesses in human beings.

The 3000 million ‘letters’ comprise about 30,000 separate genes. It is not yet known precisely how many genes there are, nor where most of them start and finish on the genome, nor which provide the ‘patterns’ for the different proteins from which skin, hair and so on are made. However though companies are not certain what is there, they are worried someone else will discover parts with commercial value. The situation is the medical equivalent of the Klondike gold rush. Private investors are desperate to stake out claims both to ‘mine’ and ‘fence off’ promising chunks of the genome using patents. One US company wanted to patent the whole genome. It was only the determination and expertise of Sir John’s team at the Sanger Institute, aided by the considerable backing of the Wellcome Trust (who contributed one third the cost) that kept the code in the public domain.

The code is as basic, but as essential, as multiplication tables. Knowing the code does not itself take medicine very far but variations on the human genome occur at a frequency of about one per thousand so, once the code is known, it becomes possible to seek links between variations of the code and specific illnesses. The prevalence of ill health is still thought to be due to a genetic susceptibility which is triggered by an environmental factor. If the key gene (or genes) can be determined, those with defective versions may be able to adopt precautionary lifestyles and drug research will be aided because the chemistry of the defective gene will be known.

It is foreseeable, said Sir John, that within 20 years we will receive a print out of our individual genome at birth. From this we will discover our personal risk of different illnesses, aptitudes (eg in sport), adult physique, and so on. More precision with drug dosing should be possible, for some need less than others. Anaesthesia could become safer because a genetic risk is present here. Genetic therapy should advance. A breakthrough should have occurred in cancer treatment. It may also be possible to understand better how the brain works and why we think as we do! This is intriguing but somewhat scary, almost to the point of fatalism. However genetically predisposed traits such as aggressiveness will still be affected by environmental exposure. Options of channelling it and other traits for good or ill, remain. New knowledge usually creates new ethical problems. Sir John

was not bothered by discrimination (eg obtaining life insurance), because discrimination in other areas, such as gender, had been addressed. A more difficult issue is that of privacy. For example genetic defects frequently affect blood relatives. Is it best to warn and risk anxiety? Is it right to request testing of relatives in the hope of pinning down the defect or environmental trigger more closely?

Sir John’s greater concern however was how scientific knowledge would be used to improve health. Per capita expenditure by the rich is thousands of times greater than by the poor. One of the costs of poverty is ill health. Drug companies focus on illnesses with the greatest markets: those affecting the most people for the most years.

Thus minorities as well as the poor get a bad deal. Priorities for health R+D are skewed. We should not be surprised that, though drug R+D is expensive, 2-3 times as much is actually spent marketing them. Sir John thanked heaven for the NHS. The NHS was prepared to pay for costly drugs for minority groups - not the case in many other countries. He thanked heaven too for NICE, the National Institute for Clinical Excellence. It is widely believed that a company promoting an anti-flu drug (up to 60 million doses a year, every year) threatened to re-locate outside the UK if its use was not supported. NICE resisted. It considered the drug’s efficacy marginal and limited promotion to vulnerable groups.

Sir John compared the free market to fire: of great value but utterly destructive if allowed to get out of hand. He proposed 3 changes. First, if health in developing countries was to be improved, there would have to be a global fund to support the necessary R+D (a levy on drug sales?). Second, there must be joined up government thinking. It was absurd, for example, to increase R+D for obesity and simultaneously support schools who encouraged crisp purchase to get computers. Third, government must recognise the value of scientific independence. Scientists will not be fully trusted on the crucial issues of health, safety, or efficacy when their university departments and research institutes are critically dependent on private company funding. Either government R+D must be increased and the return obtained from genuinely merited patents, or independent advisory groups established. “In essence” said Sir John, “we accept a system of ‘licensed greed’. We have to bring more ethical considerations into our daily lives, our trade and aid”.

One could ‘hear a pin drop’ throughout Sir John’s talk. The audience was enthralled. Baroness Lindsay Northover, who chaired the meeting, concluded with a very perceptive comment. “Many scientists are good at ‘looking down’”, she said. “Sir John has shown how a scientist could look up as well as down”.

The BBC televised the talk and showed it in its entirety on the Parliamentary channel on Saturday 27th September.

Sir John Sulston has written a book with Georgina Ferry ‘The Common Thread’ , now available in paperback from Black Swan



Sir John Sulston