Report of November 2011 Meeting
Royal Society
Southern Highlands Branch

Speaker: **Professor John Shine**
Executive Director, Garvan Institute of Medical Research
Professor of Medicine and Professor of Molecular Biology
University of New South Wales

Topic: **An update on the impact of the human genome project**

Professor John Shine was welcomed by an audience of 45 at the last meeting of 2011 for the Southern Highlands branch. He became Executive Director of the Garvan Institute in 1990, when gene cloning was starting to have a big impact in medical research; now, 21 years later, he is about to move on to another position. His name is known to most undergraduate biology students for his role in defining the Shine-Dalgarno gene sequence, which is responsible for the initiation and termination of protein synthesis.

Shine has held numerous scientific advisory roles, including Chair of the National Health and Medical Research Council (NHMRC) from 2003-2006 and Vice President (Biological Sciences) Australian Academy of Science from 2002-2007. In 2010 he received the Prime Minister's Prize for Science, the nation's most highly respected award for scientific achievement.

Professor Shine focused his lecture on "big" science and health care impacts, covering the areas of genomics, epigenomics, proteomics, metabolomics and bioinformatics. In introducing his human genome project to the audience, he quoted form the United Nations' Universal Declaration on the Human Genome and Human Rights, "The human genome underlies the fundamental unity of all members of the human family, as well as the recognition of their inherent dignity and diversity. In a symbolic sense, it is the heritage of humanity."

The human genome project has made remarkable advances. It is known that the human genome sequence contains 3.2 billion bases. In 2000, the process of determining the sequence for an individual was estimated to take 15 years and cost approximately $3b. By 2011, the time frame had been reduced to 2 months and the cost to about $20k. It is now clear that by 2014, the process should take 1 week, and cost in the order of $500. It should not be long after that the determination of a person's genome sequence becomes a routine test.

This paradigm shift in medical research has resulted in a switch from "hypothesis" based approaches to "discovery" based science, with resulting tailoring of medical care to the individual. Traditionally, the medical practitioner would examine the family history,

behavior, environment and social circumstances of a patient to determine the most appropriate treatment. Tomorrow, molecular profiling, genetic testing, proteomic profiling and metabolomic analyses will inform the medical practitioner in a much more comprehensive manner, ensuring personalized medicine.

John Shine believes that beyond 2011, all human genes will be able to be cloned and sequenced, with the resulting production of extensive genome, proteome and metabolome data bases. Understanding of the matrix of genes involved in multifactorial disorders will follow, as will a wide range of new therapies developed from gene cloning and stem cell biology. The emphasis overall will be on preventative strategies and "individualization" of both prevention and treatment.

All of this will not come without complications in social issues however. There will be reproductive decisions to be made (embryo selection), and questions raised about conceptual and philosophical implications (e.g. human responsibility vs genetic determination). Other problems arising from the rapid generation of information on the genome of individuals will be the increasing gap between what we know how to diagnose and what we know how to treat.

Professor Shine added that there will also be an increasing gap between what we think we know and what we really know!

Anne Wood