Editorial

The Sri Lankan personal genome

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Around six decades ago, in a landmark discovery and a giant leap forward for mankind, the elegant structure of deoxyribonucleic acid (DNA) double helix was delineated and the foundation of the genetic code was laid down by a collaborative research initiative[1]. Since then vast strides have taken place in our efforts towards unravelling the mysteries of Mother Nature's own genetic correlations in many different scenarios of human life.

Around half a century after the momentous discovery, an international collaborative effort, "The Human Genome Project", which started in October 1990, saw the final description of the human genome. The Human Genome Project was organized to map and to sequence the human genome. The project was formally completed in 2003[2]. That initiative succeeded in describing the three billion letters or the billion nucleotides that are found in our genetic code, the Human Genome[3,4]. The Human Genome Project was a gigantic effort between six leading nations of the world and over 1000 of the world's best scientists from a whole host of fields. It took 13 years and three billion dollars to complete the project. Since then, advances in technology has made it possible today to sequence a Human Genome in a very short period of time, at a fraction of the cost and with minimal manpower.

In modern molecular biology and genetics, the genome is the entirety of an organism's hereditary information. It is encoded either in DNA or, for many types of viruses, in ribonucleic acid (RNA). The genome includes both the genes and the non-coding sequences of the DNA/RNA[5]. The term "genome" was adapted in 1920 by Hans Winkler, Professor of Botany at the University of Hamburg, Germany. In Greek, the word *genome* (γίνομαι) means "I become, I am born, to come into being". The Oxford English Dictionary suggests the name to be a blend of the words *gene* and *chromosome*. A few related *-ome* words already existed, such as proteome and transcriptome, forming a vocabulary into which *genome* fitted in quite systematically[6].

Rapid progress in genome science and a glimpse into its potential applications have spurred observers to predict that biology will be the foremost science of the 21st century. Technology and resources generated by the Human Genome Project and other genomics research are already having a major impact on research across the life sciences. Some current and potential applications of genome research include molecular medicine, energy sources and environmental applications, risk assessment, bioarchaeology, anthropology, evolution, , human migration, DNA forensics such as identification and in areas as diverse as agriculture, livestock breeding and bioprocessing[7]. On the horizon is a new era of molecular medicine characterized less by treating symptoms and more by looking to the most fundamental causes of disease. Rapid and more specific diagnostic tests will make possible earlier treatment of countless maladies. Medical researchers also will be able to devise novel therapeutic regimens based on new classes of drugs, immunotherapy techniques, avoidance of environmental conditions that may trigger disease, and possible augmentation or even replacement of defective genes through gene therapy.

As genes are identified they can be used for genetic testing, gene therapy, tailor-made pharmaceutical products, predictive testing antenatally or later and to improve the understanding of disease mechanisms[8]. Genetic testing has potential benefits whether the results are positive or negative for a gene mutation[9]. Test results can provide a sense of relief from uncertainty and help people make informed decisions about managing their health care. A negative result can eliminate the need for unnecessary checkups and screening tests in some cases. A positive result can direct a person toward available prevention, monitoring and treatment options. Newborn screening can identify genetic disorders early in life so treatment can be started as early as possible.

Recently, in a classic demonstration of the usefulness of whole-genome sequencing in providing more optimized treatment and a better outcome, Bainbridge *et al*[10] showed that the addition of 5-hydroxytryptophan to l-dopa, produced maximal benefit in the treatment of dopa (3,4-dihydroxyphenylalanine)-responsive dystonia (DRD). The genetic analysis showed up an unusual mutation which suggested that the addition of 5-hydroxytryptophan to l-dopa is likely to be beneficial. This was proved to be right subsequently. This case showed the potential for

using genetic technology for fine-tuning treatment options for the ultimate benefit of patients.

The first Sri Lankan Personal Genome was successfully sequenced by scientists and bio-informaticians from the University of Colombo, Sri Lanka and the Institute of Genomics and Integrative Biology, New Delhi, India. This project was initiated by the Specialty Board in Biomedical Informatics of the Postgraduate Institute of Medicine, University of Colombo, Sri Lanka. The official launch of the Sri Lankan Personal Genome took place on the 10th of December 2010 at the Annual Scientific Sessions of the Sri Lanka Association for the Advancement of Science in Colombo, Sri Lanka. The hard disk containing the Sri Lankan Personal Genome was handed over to the Senior Minister for Scientific Affairs, Honourable Professor Tissa Vitharana by Professor Vajira H. W. Dissanayake, Chairperson, Specialty Board in Biomedical Informatics and Dr. Rajesh Gokhale, Director Institute of Genomics and Integrative Biology, India.

The first Sri Lankan Personal Genome sequence came from a Sinhalese man with both upcountry and low country heritage. It is hoped that the project would be extended to cover other ethnicities as well in the years to come. The biomedical informatics group in the University of Colombo will continue to analyse the data generated to make sense of the vast amount of information available in the Sri Lankan Personal Genome. In addition the data has been made available freely to all scientific groups in the country to analyse, explore and make their discoveries freely available to the scientific and the medical communities.

The Sri Lankan Personal Genome is Sri Lanka's contribution to the growing pool of scientific knowledge on genomes. With the launch of the Sri Lankan Human Genome, genetic research in Sri Lanka has truly entered the post genomic era. Let us join hands to ensure that the scientific discoveries that we make when we analyse our genome will benefit the Sri Lankan society at large.

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