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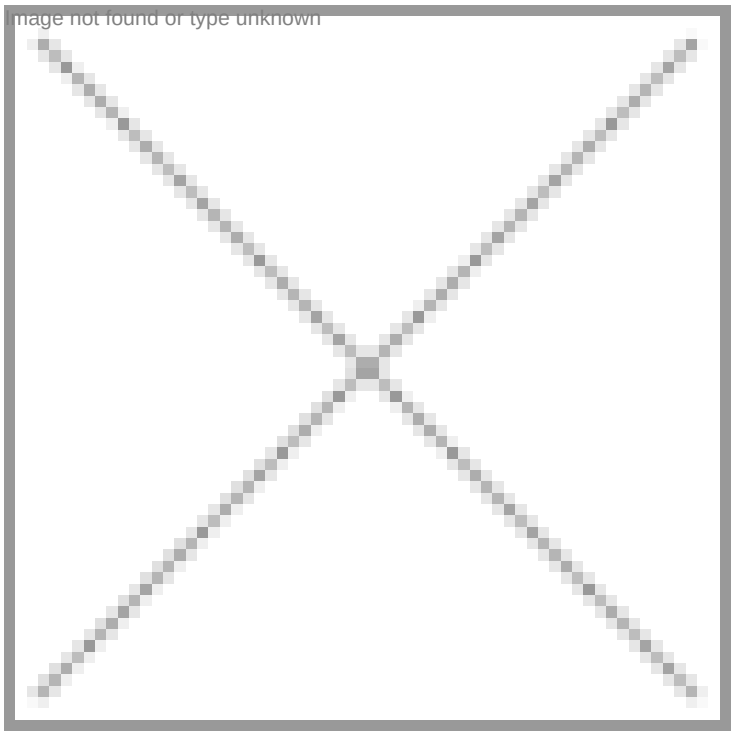

Unraveling of complete human genome sequence: hope or hype?

The deciphering of genomes will be invaluable in revealing the detailed mechanism of the immune response and would provide new targets for drugs, as well as the basis for designing new vaccines. Gene discoveries and understanding the human genome have resulted in improved understanding of the genetic basis of disease and created new opportunities for diagnosis, treatment, and prevention of disease.

Since the completion of the Human Genome Project in 2003, research efforts have been aimed at analyzing the functions of various sequences in the genome, using both experimental and computational strategies.

Biologist J Craig Venter is back with the decoding of his own genome. He and his team recently announced that they had finished the first mapping of a full, or diploid, genome, made up of DNA inherited from both parents. This new genome sequence is considered by many scientists to be of much higher quality and contains much more detail than the genome completed by the Human Genome Project consortium.

Published in the online open access journal PloS Biology, the 2.8 billion contiguous bits of genetic code will also hasten advances in preventive medicine. The findings overturn what had in a few short years become genetic gospel: that all human beings are, genetically 99.9 identical.

Dr Craig Venter

This set of 23 paired chromosomes, made up of six billion chemical units in total, is the first full human genome ever decoded

for an individual.

Dr Venter has spent the last five years and an extra $10 million of his institute's money in improving the draft genome he prepared at Celera. That genome was based mostly on his own DNA, and the new diploid version is entirely so.

Like James Watson, the co-discoverer of DNA, whose genome is also being decoded, Dr Venter has made individual DNA sequences public to advance knowledge and hasten the era of personalized genomic medicine.

Dr Venter has also been in the news recently for seeking to patent the world's first man-made species referred to as Mycoplasma laboratorium, a synthetic microbe that could be used to produce fuel. It is part of an effort to create designer bugs to manufacture hydrogen and biofuels, as well as absorb carbon dioxide and other harmful greenhouse gases.

In 1998, Dr Venter founded Celera Genomics to sequence the human genome using the whole genome shotgun technique, new mathematical algorithms, and new automated DNA sequencing machines. The successful completion of this research culminated with the publication of the human genome in February 2001 in Science. In addition to the human genome, Dr Venter and his team at Celera sequenced the fruit fly, mouse, and rat genomes.

A less complete version has been reported in 2001 by the Human Genome Project, a consortium of academic centers supported largely by the National Institutes of Health and the Wellcome Trust, a medical philanthropy in London.

The first two maps of the human genome were based on a patchwork of DNA from several donors. Both versions were also half maps, decoding only one set of the 23 chromosomes on the assumption that the two sets would hardly differ.

Both sides have achieved remarkable success with their chosen approaches. Celera's whole genome shotgun approach has proved faster, but both with its fruit fly and the human genome, Celera has made use of data obtained by the consortium's clone-by-clone approach. If Celera's version of the human genome proves as good as its fruit fly genome, scientists may judge it to have chosen the better path. Nonetheless, both sides can fairly claim credit for the final result.

The two groups even differ on the size of the gene-coding part of the genome. Celera says it is 3.12 billion letters of DNA; the public consortium claims that it is 3.15 billion units, a letter difference of 30 million. Neither side can yet describe the genome's full size or determine the number of human genes. Both versions of the human genome meet the important goal of allowing scientists to search them for desired genes, the genetic instructions encoded in the DNA.

Such speedy gene sequencing would represent a technology breakthrough for medical research. With this type of knowledge now in hand and a better understanding, the human genome is expected to revolutionize the practice of medicine where genome sequence information becomes a critical reference to assist with health-related decisions. Biologists expect to develop an array of diagnostics and treatments based on it and tailor it to individual patients, some of which will exploit the body's own mechanisms of self-repair.

Genes can give useful information about increased risk for disease, but in most cases they do not determine the actual cause of the disease, or the actual incidence of somebody getting it. Most biology will come from the complex interactions from all the proteins and cells working with environmental factors, not driven directly by the genetic code. But by knowing before a disease develops what one is susceptible to, one can try to prevent it, thus lowering the healthcare costs. Preventive medicine is expected to be the future. For example, if a disease is detected early, because a person knew he had a genetic risk and was being frequently examined, the surgery would be relatively inexpensive and average survival is far greater than 10 years. The most extreme suggested use for the human genome data is editing the DNA inheritance bequeathed from one generation to the next. This involves identifying an abnormal gene and then correcting it in the cells, which are used to pass genetic information to offspring - eggs and sperm. Therefore no subsequent generation would then be afflicted by their ancestors' gene defect.

The era of personal genomics is yet to begin. Next generation genomics technologies are breathing new life into the market, and are expected to contribute to the robust growth of the US genomics market and other continents between 2005 and 2012, according to several industry experts. Top US industry participants are successfully developing specific applications for each evolutionary stage of the genomics research process, and are likely to maintain revenue streams, while strategically positioning themselves to penetrate the future markets for clinical applications of genomic technologies.

New analysis from Frost & Sullivan, strategic analysis of the US genomics markets, reveals that revenues in this market totaled $1.85 billion in 2006, and is likely to reach $3.69 billion in 2012. That doubling in revenue will occur along with a huge increase in the amount of DNA sequence produced per dollar spent. It is also expected that sequencing costs will drop by three times perhaps in the next five years from now. The cost of sequencing DNA has been dropping by half every two years.

The market for DNA-sequencing machines and chemicals is expected to increase between 4 percent and 5 percent per year.

Scientists are doing most of the DNA sequencing for their own research purposes today. But at some point in the next 5-10 years, the desire to learn one's own personal genome sequences will become the biggest source of demand for DNA sequencing services. Also demand will grow for surreptitious DNA sequencing services so that people can learn the DNA sequences of prospective employees, celebrities, and business competitors. Science is expected to turn up all sorts of practical uses of DNA sequence information and the genetic privacy will become very hard to protect.

While DNA patents create a wide variety of possible benefits and harms for science and technology, the evidence at this point in time supports the conclusion that it will probably promote rather than hamper scientific discovery and innovation. However, since DNA patenting is a relatively recent phenomena and the biotechnology industry is in its infancy, efforts would be to gather evidence about the effects of DNA patenting on scientific innovation and discovery as well the economic, social, and legal conditions relating to intellectual property in biotechnology. The markets, the courts, researchers, and patent offices should be given a chance to settle issues related to innovation and discovery, before we seek legislative remedies are sought, since new laws proposed at this point would lack adequate foresight and could do more harm than good. However, one should be open to new laws or regulations on DNA patents if they are required to in order to deal with some of the biases and limitations of the free market.

It will be a while before one will be able to garner a full understanding of how genes influence our lives. Within five years, faster and cheaper sequencing techniques could produce complete genomes for 10,000 people, laying the foundation for an era of individualized genomics.

Quick and affordable decoding of many people's DNA will accelerate the effort to understand the links between genes and diseases and people may one day plug their genetic information into Internet search engines to download the latest information on their genetic strengths and susceptibilities. It is going to be the best way to get information about lifestyle choices. Biology has progressed from near total ignorance of the hereditary material to possession of the entire human genome within 50 years and is testament to a hectic pace of discovery. There is little doubt that the revelation of the human genome will benefit healthcare in the short and long term.

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