Our Nature (2003) 1

MISCELLANEOUS NOTES

1. A Glimpse on Human Genome

Cell is a basic unit of life. That is the smallest structures, capable of basic life processes, such as taking in nutrients, expelling waste, and reproducing. All the instructions needed to direct their activities are contained within the chemical DNA. Any organism's complete set of DNA (chromosomes) including its gene is genome. The set of chromosomes contain certain number of genes, which are different in every genus and species. Chromosome is microscopic structure found within cells that carries the DNA, the hereditary material that influences the development and characteristics of each organism. DNA carries hereditary information in a form that can be copied and passed intact from generation to generation. A gene is a segment of DNA. There are numerous genes in an organism and each of these genes are responsible for expressing the different characters, e.g. hair color, eye color, hair texture etc.

Since the earliest days of plant and animal domestication, around 10,000 years ago, humans have understood that the characteristic traits of parents could be transmitted to their offsprings. The first to speculate about how this process worked were Greek scholars around the 4th century B.C., who promoted theories based on conjecture or superstition. Some of these theories remained in favor for several centuries. The scientific study of genetics did not begin until the late 19th century. Until the 1980s, genetic researchers focused their work on the fundamental genetic processes in simpler organisms, such as bacteria, plants, and fruit flies. Today an expanded array of tools available for the direct study of human genetics attracts scientists from around the world to collaborate to identify and study every human gene.

The Human Genome Project began in earnest in the United States in 1990 with the expansion of funding from the National Institutes of Health (NIH) and the Department of Energy (DOE). The first director of the U.S. program was American Biochemist **James Watson**. Many nations have official human genome research programs as part of this collaboration, including the United Kingdom, France, Germany, and Japan. In a separate project intended to speed up the sequencing process and commercialize the results, **Celera Genomic**, a privately funded Biotechnology Company, used a different method to assemble the sequence of the human genome. Both the public consortium and Celera Genomic completed the first phase of the project, and they each published a draft of the human genome simultaneously, although in separate journals, in February 2001 (Fasman 2002).

The project goals are to identify all the approximate 30,000 genes in human DNA, determine the sequences of the 3 billion chemical base pairs that make up human DNA, store this information in databases, improve tools for data analysis, transfer related technologies to the private sector, and address the Ethical, Legal, and Social Issues (ELSI) that may arise from the project. Knowledge about the effects of

DNA variations among individuals can lead to revolutionary new ways to diagnose, treat, and someday prevent the thousands of disorders that affect us. Besides providing clues to understanding human biology, learning about nonhuman organisms' DNA sequences can lead to an understanding of their natural capabilities that can be applied toward solving challenges in health care, energy sources, agriculture, and environmental cleanup. To help achieve these goals, researchers also are studying the genetic makeup of several nonhuman organisms. These include the common human gut bacterium *Escherichia coli,* the fruit fly, and the laboratory mouse. Discovering approximately 25,000 to 35,000 human genes (the human genome) make them accessible for further biological study, and determine the complete sequence of the 3 billion DNA subunits (bases) (Wolfsberg *et al.* 2001). In February 2001 a “rough draft” of the DNA sequence of the human genome was published. The draft provided a basic outline of 90 percent of the human genome. Researchers expect that a finalized version of the complete sequence of the human genome will be finished by 2003, two years earlier than originally projected. With the simultaneous description of a second draft of the human genome by Celera Genomics, scientists now have a more detailed blueprint of the human genetic code. Scientists were surprised to learn that the actual number of human genes is far lower than expected—only about 31,000 genes compared to the predicted 100,000 genes. This number is a little more than twice the number of genes found in the fruit fly (Fasman 2002). It is the achievement of a coordinated effort involving 20 laboratories and hundreds of people around the world. It reflects the scientific community at its best: working collaboratively, pooling its resources and skills, keeping its focus on the goal, and making its results available to all as they were acquired. The Human Genome Project has achieved the following progress until February 12, 2001.The human genome contains 3164.7 million chemical nucleotide bases (A, C, T, and G).The average gene consists of 3000 bases, but sizes vary greatly, with the largest known human gene being dystrophin at 2.4 million bases. The total number of genes is estimated at 30,000 to 35,000, much lower than previous estimates of 80,000 to 140,000 that had been based on extrapolations from gene-rich areas as opposed to a composite of gene-rich and gene-poor areas. The order of almost all (99.9%) nucleotide bases is exactly the same in all people. The functions are unknown for over 50% of discovered genes. The human genome's gene-dense "urban centers" are predominantly composed of the DNA building blocks G and C. In contrast, the gene-poor "deserts" are rich in the DNA building blocks A and T. GC- and AT-rich regions usually can be seen through a microscope as light and dark bands on chromosomes. Chromosome 1 has the most genes (2968), and the Y chromosome has the fewest (231). The ratio of germline (sperm or egg cell) mutations is 2:1 in males vs. females. Researchers point to several reasons for the higher mutation rate in the male germline, including the greater number of cell divisions required for sperm formation than for eggs.

The Human Genome Project will be ended in the third- and final plan “**September 30, 2003”** with providing numerous achievements to the mankind. After completing the Human Genome sequence we will be able to know about all genes function, position, composition, identification and regulation. The

availability of a reference human DNA sequence is a milestone toward understanding how humans have evolved, because it opens the door to large-scale comparative studies. The major impact of such studies will be to reveal just how similar humans are to each other and to other species (Pääbo 2001). The sequencing of the human genome heralds a new age in medicine, with enormous benefits for the general public. For example, it will allow scientists to identify all of the genes contributing to a given disease state, leading to a more accurate diagnosis and precise classification of disease severity. In addition, healthy patients can know the diseases for which they are at risk, giving them the opportunity to make beneficial lifestyle changes or to take preventive medications to protect their health. Understanding the genetic bases of heritable diseases also will allow researchers to develop therapeutics at the molecular level, resulting in better treatments with fewer side effects (Jeffords and Daschle 2001). The identification of all human genes, for example, will help improve diagnosis of most inherited diseases. The research also will help uncover the mechanisms that make some people more susceptible to certain diseases, such as heart disease, stroke, and several kinds of cancer, and pave the way to more effective therapies and preventive measures, scientists believe. A virtually complete list of human gene products will give us a vast repertoire of potential new drugs. Our medical record will include our complete genome as well as a catalogue of single base-pair variations that can be used to accurately predict our responses to certain drugs and environmental substances. This will permit us to be treated as a biochemical and genetic individual, thus making medical interventions more specific, precise, and successful. In the next 15 to 20 years, more effective drugs will be developed, and doctors will test individual genetic profiles against panels of drugs available for a specific condition and choose the treatment with the greatest potential benefit.

Today, some 100,000 people die each year from adverse reactions to drugs, and millions of others must bear uncomfortable or even dangerous side effects. As genes and other DNA sequences that influence drug response are identified, we can expect the number of toxic responses to drop dramatically and most side effects to be eliminated. One of the most difficult issues is determining the proper balance between privacy concerns and fair use of genetic information (Anon 2002). The growing number and use of genetic tests has many worried about discrimination due to inappropriate access to, and use of, private genetic information. Most protections, whether in terms of employment or health insurance discrimination, are at the state level. If there is not strict legislation for fair uses of genetic information, we may face some unpleasant consequences unless society makes some hard choices. Based on genetic information, employers may try to avoid hiring workers they believe are likely to take sick leave, resign, or retire early for health reasons (creating extra costs in recruiting and training new staff), file for workers' compensation, or use healthcare benefits excessively. Some employers may seek to use genetic tests to discriminate against workers--even those who do not and may never show signs of disease--because the employers fear the cost consequences.

Genetic predisposition or conditions can lead to workplace discrimination, even in cases where workers are healthy and unlikely to develop disease or where the genetic condition has no effect on the ability

to perform work. Insurers can still use genetic information in the individual market in decisions about coverage, enrollment, and premiums. Insurers can still require individuals to take genetic tests. Individuals are not protected from the disclosure of genetic information to insurers; plan sponsors (employers), and medical information bureaus, without their consent. Rapid progress in genome science and its potential applications make 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.

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2. Avifauna of Tinjure Forest, Eastern Nepal

Tinjure Forest, one of the ecologically important regions, lies on the eastern mid-hills of the country at a distance of about 9 km. from Basantapur bazaar of Tehrathum district. Its altitude ranges between 2400-3010 m and covers 22 sq. km. lying in between Basantapur and Chauki (Rai 1998). Basantapur is a small market located on the boarder of Sankhuvasabha and Terhathum district. Infrastructure of development, such as road and communication has turned this area into the trading centre for Terhathum, Sankhuvasabha and some parts of Bhojpur district. Large amount of *Lokta' (Daphne* spp.*)* and medicinal plants are collected there. The area is mostly steep with forested slopes and ridges. The broad-leaved deciduous moist forest mostly covers the area and the lower canopy is composed of *Rhododendron* spp., *Quercus* spp., and *Daphne* spp. It is rich in biodiversity and provided the home to various indigenous and rare birds like Kalij Pheasants (*Lophophora leucomelana*) and Satyr Tragopan (*Tragopan satyra*). There is no specific geographical barrier in between the adjoining areas so that the movement of wildlife and birds within these areas is possible. The area is covered with magnificently beautiful and splendid *Rhododendron* jungle intermixed mainly with oak and other tree species such as *Magnolia* spp., *Castanopsis* spp., *Abies spectabilis*, *Betula utilis*, *Taxus baccata* with different kinds of shrubs, herbs, climbers, epiphytes and grasses. In these regions, survey of bird was conducted in 1997-1998. A total of 73 bird species belonging to 23 families were recorded. Birds were observed in dense forest, degraded forest, bamboo grooves, scrub and open habitats.

Birds were identified (Table 1) with the help of available literature (Fleming *et al*. 1979, Inskipp and Inskipp 1985, Ali and Ripley 1995).