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ETHICAL, LEGAL AND SOCIAL ISSUES IN BIOINFORMATICS APPLICATIONS

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Abstract

Bioinformatics is the use of computing for acquisition, management, organization, storage and analysis of biological data. The introduction of various bioinformatics applications plays a leading role in the health sector in recent years. Though, the bioinformatics applications possess many advantages, each application makes some ethical, legal and social issues in the community. This study analyses applications of Bioinformatics in forensic databases with DNA fingerprinting, Biological weapons with genetic engineering, Pharmacogenomics, Personalized medicine and Detection of mutation with NGS through critically assessing available literature. The ethical, legal and social issues are arising due to the computational analysis of genetic data which reveal some personal hidden information about individuals. Because the result may reveal some ugly and bitter truth which cannot be accepted in the society sometimes. The privacy is the most concern as most of the Bioinformatics applications are dealing with genes which is specific and private to each individual. Each bioinformatics application should be proven as safe and secure as those input data and the results are considered as more sensitive and private. This study proposes some healthy practices and policies to reduce the above mentioned non-technical impacts.

Keywords: Bioinformatics, gene profiling, ethical issues

Introduction

Advancement in computational technologies are improved massively in this decade. Several scientific dreams became true due to these massive technological changes in computing as well as Biology. Further, considering Biology, there were several hidden and unknown facts are revealed by applying computational algorithms. The growth of data science such as Bigdata analysis, Data Mining and Pattern Recognition are contributed a lot in the Biological domain. This advancement leads to a new discipline called Bioinformatics where modification to both computing and Biology has been done. Bioinformatics includes variety of applications and each contributes a lot to health sector in many ways such as treatment techniques, monitoring, predicting etc. However, there are many issues may arise due to the social, cultural, ethical bindings of mankind. It is easy to know about the physical and mental behavior of individuals if we have some computational pipeline and his/her genetic data. This may expose of confidential medical information or other details that could potentially harm the particular individual. This study has been undertaken in order to figure out the ethical, legal and social issues as a result of bioinformatics applications in health industries and to suggest some solutions to reduce the impacts up to some extent.

Methodology

This study has been carried out through analyzing five bioinformatics applications through critically reviewing the existing literature. Also, YouTube videos and some online documents submitted by the scientists and biologists regarding the influence of Information Technology and issues possessed on the functioning of these Bioinformatics applications were analyzed in terms of ethical and cultural issues and also possible solutions. The most widely used ten bioinformatics applications are DNA and protein sequencing, Protein Modelling, Evolutionary Studies, Pharmacogenomics, Genetic Engineering and Biological weapons, Personalized medicine with IBM Watson, Detection of mutation using NGS, use of genomics with NGS, Forensic databases with DNA fingerprinting and Proteomic technology according to survey done by Deniz and Canduri [1]. A survey was done in order to figure out the commonly used applications among the ten applications mentioned above. There were 120 questionnaires were circulated and 100 valid questionnaires were got responded. The results are shown in Figure 1.
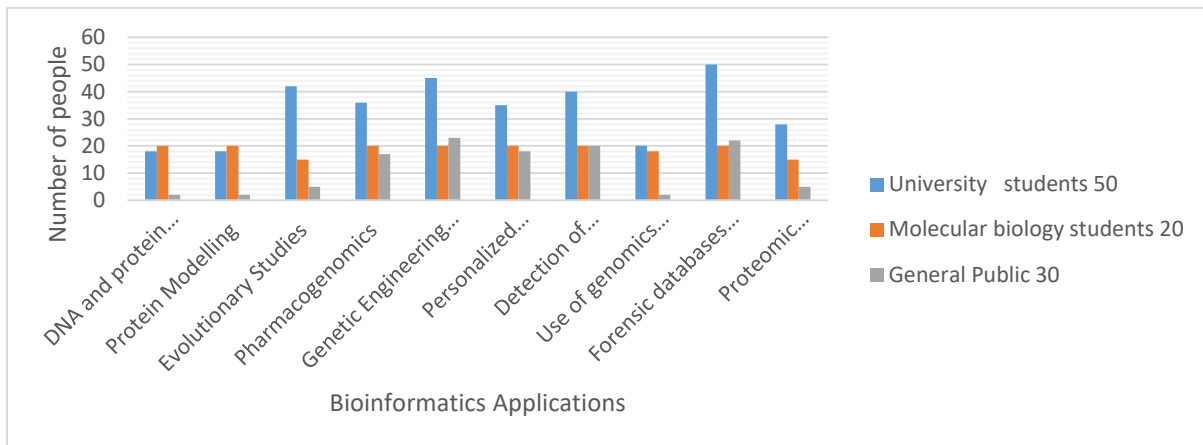

Figure 1: The result of survey

The rest analysis has been carried out with the selected five applications based on the results of the survey to analyze the ethical, legal and social issues caused by applying advanced technology. The selected five applications among the ten discussed by Diniz and Canduri [1] are Forensic databases with DNA fingerprinting, Genetic Engineering and Biological weapons, Pharmacogenomics, Personalized medicine and Detection of mutation using NGS. The information collected regarding the role of IT in each of the above-mentioned bioinformatics application based on literature are described below. The information collected regarding the role of IT in each of the above-mentioned bioinformatics application based on literatures are mentioned below.

Forensic databases with DNA fingerprinting

DNAforensics is a branch of forensic science that focuses on the use of genetic material in criminal investigation. It is first made its way into the courts in 1986, when police in England asked molecular biologist *Alec Jeffreys*, who had begun investigating the use of DNA for forensics, to use DNA to verify the confession of a 17-year-old boy in two rape-murders in the English Midlands[2]. The software tools used under DNA fingerprinting are, GelJ, and

automatic DNA Diagnosis for 1D Gel Electrophoresis images using Bio-image processing technique (GELect).

Genetic engineering and biological weapons

The basic principle of genetic engineering is gene transfer. Many foods consumed today are either genetically modified whole foods, or contain ingredients derived from gene modification technology. Billions of dollars in U.S. food exports are realized from sales of GM seeds and crops. [3]The principal concern seemed to be the fear of a public health disaster because there is a risk of some genetically superpowered microorganism may be accidentally released into general public [4]. Biological weapons are designed to spread disease among people, plants, and animals through the introduction of toxins and microorganisms such as viruses and bacteria. [5] In the bioweapon industry, genetic engineering can be used to manipulate genes to create new pathogenic characteristics aimed at enhancing the efficacy of the weapon through increased survivability, infectivity, virulence, and drug resistance. While the positive societal implications of improved biotechnology are apparent, the "black biology" of bioweapon development may be one of the gravest threats we will face in near future.

Pharmacogenomics

Pharmacogenomics is a discipline that aims to explain the inherited basis for differences in drug response between individuals. It is defined as the application of whole-genome technology for the prediction of the sensitivity or resistance of an individual's disease to chemotherapy. Evaluation of the safety profile of medicines that are already on the market is also an important area in which Pharmacogenomics profiling can be used. Microarray or Biochip is the DNA microarrays which have been proven to be a state-of-the-art technique for high throughput comprehensive analysis of thousands of genes in parallel.

Personalized Medicine with IBM Watson

The ability to offer the right drug, the right patient, for the right disease, at the right time, with the right dose. Personalized medicine involves identifying the genetic information which paves way for the predictions to be made about a person's susceptibility. IBM Watson is cognitive computing applied to clinical decision support in personalized medicine. Personalized medicine has tremendous potential benefits for patients and healthcare providers, as well as for regulatory agencies and pharmaceutical and diagnostic companies, but the advancement of this innovative therapeutic strategy depends on identifying biomarkers functioning as companion diagnostics for the targeted drug.

Detection of Mutation with NGS

Detection of unknown mutations can involve sequencing of DNA, often in many patients. This has led to the development of methods to screen DNA for mutations as well as methods to detect mutations. Next Generation Sequencing is a high-speed sequencing technique. It generates a comprehensive molecular profile of a patient. It is a high throughput sequencing discovery of mutated genes that drive oncogenic phenotypes in tumors. Targeted

next-generation sequencing (NGS) provides a promising method for diagnostic purposes by enabling the simultaneous detection of multiple mutations in various genes in a single test.

Analyzed issues behind the above-mentioned bioinformatics applications based on literature reviews

Several countries nowadays maintain DNA banks of people who committed crimes. Unfortunately, there have been some cases when the DNA of people who were arrested but not convicted were accidentally entered into the database. In this case, DNA fingerprinting can be seen as a tool that violates the privacy of people and makes their personal information easily available to others. New organisms created by genetic engineering could present an ecological problem. The eagerness to increase crop products has resulted in the genetic manipulation of plants, which has raised much polemics ranging from political, ethical and social problems. One cannot predict the changes that genetically engineered species would make on the environment. In addition, many genetically engineered foods use microorganisms as donors whose allergenic potential are either unknown or untested. As well, genes from non-food sources and new gene combinations could trigger allergic reactions in some people, or exacerbate existing ones. [3] In the context of applications of genetic engineering in human life, misuse of this technology in the production of biological warfare or weapons is a biggest disadvantage. The creation of transgenic animals and plants promises many benefits, although it also raises critical questions about how far we should go in applying genetic engineering techniques. This raises political concerns and have been the subject of legislative hearings, regulatory actions and court deliberations. [6]. Recently, Jian-kui HE, a Chinese scientist, have created the first gene-edited babies, called Lulu and Nana, who are naturally immune to the human immunodeficiency virus (HIV). HE has used the CRISPR-Cas9 technique to modify the babies' germline gene. China's guidelines and regulations have banned germline genome editing on human embryos for clinical use because of scientific and ethical concerns. Jian-kui HE's human experimentation has not only violated these Chinese regulations, but also breached other ethical and regulatory norms. 122 Chinese scientists stated that while CRISPR-Cas involves serious off-target risks and associated ethical considerations, and so should not be used to produce genetically modified babies. So, this gene modification may bring little substantial benefit to the babies, while exposing them and their future generations to unknown and uncontrollable risks. [7]

Advancement in the use of individual pharmacogenomics profile makes privacy at risk. The U.S. Senate and the U.S. House of Representatives are attempting to pass the Genetic Information Nondiscrimination Act of 2007 in hopes of protecting individuals from genetic discrimination in terms of health insurance and employment. [8] If patients consult Watson directly then they may receive wrong diagnosis without ever being seen by a doctor. In this case, who is to be blamed? Malicious human interaction with Watson results in wrong output and individuals get to know wrong diagnosis about them and become depressed.

Results and Discussion

The figured-out bioinformatics applications are working under different technologies. Due to the Information Technology, each application performs fast and produce results within minutes. Up to some extent forensic science can violate people's privacy as it focuses on the use of genetic material in criminal investigation. This results in making the personal information of general public easily available to others. Just a small amount of human error

(such as exposing the sample to other substances or incorrectly identifying two samples as identical) can ruin the process or alter the results. The result will hardly affect the whole family of the particular individual. An accident in engineering the genetics of a virus or bacteria (for example could result in a stronger type), which could cause a serious epidemic when it released to outer environment. This could create problems ranging from minor medical problems, to death in human genetic engineering. An individual's privacy and confidentiality are at risk through the development of individual pharmacogenomics profiles which would become easy to access and sharable. Even a simple change in a gene will affect a whole generation as the behavior or the changes are inherited. Unlike western countries, we the Asian countries are tightly bound to cultural and social facts. Genetic and genomic information are sensitive by nature, if it leaked it will result in big social and cultural issue.

Recommendation and Conclusion

However, even though there are many advantages due to the advancement in Biology, there are possibilities to make some social issues which may collapse civilization structure and cultural aspects of a community as human are social animal. The analysis of the ethical, legal and social issues of the bioinformatics applications create an awareness among the general public who are using these applications for their easy and accurate results. Some proposed solutions are given below in order to reduce the ethical, social and legal issues.

1. Need to have stringent quality control checks in DNA fingerprinting labs worldwide.
2. Policies and practices related to patient data & privacy, must ensure appropriate consent.
3. New laws to prevent genetic discrimination by insurers and employers.
4. Researchers should evaluate the legitimacy of their work while handling with genetic engineering.
5. Existing technical and ethical guidelines should be refined and more rigorously enforced to guide and standardize relevant research and applications.
6. Conducting Awareness programs to general public.

Finally, whatever the advance technology there some impacts and issues, it can be managed if there are proper awareness, quality measures and regulation policies introduced by relevant authorities.

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