Application of Ethical Theories to Human Genome Sequencing

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Abstract
We apply a number of ethical theories to ethical concerns an issues arising from the technical ability and practice of performing DNA and human genome sequencing. We identify the scientific facts of DNA and genome sequencing and the related ethical concerns. Subsequently, we attempt to apply relevant ethical theories to these concerns and issues. The ethically issues discussed include privacy, ownership of genome information, misuse of information, genome databases, and potential of using genome sequencing as a tool against selected groups.

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1. Introduction

*DNA (deoxyribonucleic acid)* is double helix molecule composed of building blocks called nucleotides, commonly referred to by the name of one of their components called base. There are four kinds of bases in DNA named adenine (A), guanine (G), cytosine (C), and thymine (T). Inside a DNA molecule, these bases are arranged into pairs, bases from one helix bonded to the corresponding base from the other helix. Each DNA molecule is stored in a structure called *chromosome*. We, as a human species, carry 23 different kinds of chromosomes pairs, one chromosome in each pair from one parent and the other from other parent. These 23 chromosome pairs collectively are called human *genome*, which reside in the nucleus of almost every type of cells in our body.

DNA *sequencing* is the process of letter-by-letter (base by base) readouts of the DNA, and *genome sequencing* refers to the sequencing all the DNA molecules in a genome. The first sequencing of a human genome, about 3 billion DNA base pairs (bp), was completed in 2003 by the world’s largest collaborative biological project ever [1]. That was both scientific and technological feat, which also brought “big science and big data” model to biology that was previously confined to physics [2]. The cost of sequencing the first human genome in 2003 was just about 3 billion U.S. dollars ($1 per bp). Since then, as shown in figure 1, the cost of sequencing of an average human genome has plummeted to only few thousand dollars largely due to the progress in sequencing technology [3]. It is already happening today that your doctor can order your genome sequencing for less than $10,000. In fact, we may be very close to realizing the goal of sequencing human genome for just $1,000.
Like almost any other breakthrough in science, DNA sequencing has come with a spectrum of ethical concerns and issues. As DNA sequencing cost is decreasing and getting closer to being affordable, related ethical concerns increasing.

2. **Scientific Facts of Human Genome Sequencing and Related Ethical Concerns**

Potential advantages of human genome sequencing are enormous ranging from finding causes and cures of genetics diseases to exploring the evolution life of Earth. The bigger are the advantages, the higher is the risks of wrong-happenings. To appreciate those risks or ethical issues, we need to understand the related scientific facts about DNA molecule and human genome sequencing. These facts are briefly discussed in the following.

**DNA Molecule, the Blueprint of Life.** DNA is often called the blueprint of life as we (and any other kind of life) are developed according to the instructions chemically coded in DNA
molecules of our genome. The single most important fact about genome is that it contains all the information needed to form a living organism. Genome sequencing reveals this biological information about an organism.

**Information at Individual Level.** The human genome sequencing reveals the most accurate and unique identity (ID) of an individual. This ID has or can be used to determine a lot of information about the individual including ethnic background, family tree, and the genetic diseases or disorders the individual is susceptible to. This very personal and valuable information raises the issues of privacy, and misusing (or abusing) the information and/or unfairly profiting from it. This also brings the consent issue to the forefront. By consent, we mean obtaining the consent of the information owner before using the information.

**Information at Fundamental Level.** Genome sequencing reveals the structure, i.e., the nucleotides and their sequential order in each DNA molecule. It offers us the opportunity to understand life and its processes most fundamental level, the molecular individual level. The knowledge obtained from this understanding will give us the ability to control and engineer some aspects or feature life at molecular and individual levels. This includes the ability to alter the existing life forms and create new ones. Overall, this research opens the door to many opportunities that include understanding genetic diseases and possibly developing cures. This of course is good news, but this also raises ethical issues such as unfairly profiting from genome information of others.

The ability to control (and manipulate) life at molecular level is a powerful thing, which runs the risk of dual research, that is, the research that can be used for both good and bad ends. This kind of concerns has been loudly expressed under labels such as Frankenstein science or *playing God.*
**Availability of Genome.** Genome can be found in almost all kinds of cells in our body. That means the genome can be collected from almost any part of our body such as blood, hair, skin, and saliva to sequence.

**Collection of Human Genome.** This is a scientific fact that real (or most) scientific and medical value comes from a collection of human genomes, and not from any individual genomes. This is because most information is extracted by directly or indirectly comparing lots of individual genomes. By indirect comparison, we mean that if a particular research project does not require such comparison, it might depend on the results of research where the comparisons were made.

**Big Data.** As pointed out in section 1, a DNA molecule holds huge information. For example, barebones sequencing of a human genome, ie about 3 billion DNA bp, produce about 800 megabytes of data. But in practice considerably more data about each base is usually collected, and genes are often sequenced multiple times to ensure accuracy. So, it is common to arrange about 100 gigabytes per human genome [4]. This fact combined with the need of comparing different genomes to one another gives rise to the research requirement of setting up genome databases. These databases, in turn, give rise to a host of ethical issues including privacy, ownership, and profit.

How do we know what is the right way to handle ethical issues? For that, we turn to ethical theories. To make this document self-contained, we present a brief account of ethical theories [5] used in the paper, in Appendix A. However, before applying these theories to ethical issues at hand, we discuss their two most important characteristics.

### 3. Two Global Characteristics Ethical Theories

While applying these ethical theories to any issue, we should keep in mind the following two characteristics that apply to all ethical theories [6].
1. **Common Grounds.** A cardinal characteristic of an ethical theory is that it is clear, consistent, and universal. This characteristic is important for the theory in order to deliver a moral judgment that is based on logic, facts, and defensible moral principles, which consider the values at stake. All this provide common grounds for any viable discussion.

2. **Correlation.** All ethical theories attempt to answer the same question: What is morally right or wrong action in a given situation? However, because they may use similar or different criteria (premises), they may reach similar or different conclusions on what is right or wrong, given the same situation. Therefore, no ethical theory holds the absolute truth if there is such a thing called *absolute truth*.

   Keeping these two global characteristics in mind, we now apply ethical theories to ethical issues identified in section 2.

4. **Addressing the Ethical Issues**

   In this section, we address the ethical issues about human genome sequencing is light of ethical theories. The issues discussed here include playing God, misuse of genome information, privacy and ownership of genome information, genome databases, and use of genome sequencing as a tool to discriminate against women and minorities.

   **Playing God.** People with this concern believe that by performing genome (or DNA) sequencing and doing the related research, the researches are playing God. Therefore, this research is unethical and should be stopped. This argument has its roots in the divine command theory, and it has been presented in one or other forms after almost any major scientific breakthrough. In case of genome sequencing, in its strong form, it goes something like the following. God, the creator of the universe, is also the creator of human and any other life. Therefore, only God, the Creator, have the right to change life or create new life forms. The
DNA sequencing has the potential of enabling researchers to change the existing life or even create new life forms. Ultimately, it may lead to creating monsters or doing other deconstructive things, the Frankenstein science. Thus, by interfering with the God’s life creation process, the researchers are playing God, which is morally wrong.

Against this argument, we will make two counterarguments: one from within the divine command theory and other from outside of it. From the grounds of divine command theory, it can be argued that God have given us brains to use. This is a scientific fact that when you stop using your brain, you start losing it. By performing DNA sequencing, the researchers are using their brains to understand some natural laws and life processes. So, this is not wrong according to divine command theory; and this is ethically right thing according to the natural law theory.

Moreover, the playing God argument rests on rather vague premises (divine commands), which change in their meanings over different interpretations of religious text. In the history of scientific progress, from Galileo to Darwin to present times, this argument of playing God or violating God’s commands have been used many times in one or other form. In almost every case, eventually, an interpretation of religious text was found to reconcile with the discovered scientific facts or scientific theories supported by facts.

Moreover, halting DNA (or genome) sequencing research, we would be depriving humans of the potential benefits (and resulting happiness) to which the research might lead, such as cure of genetic diseases. That (i.e., to stop research) will be morally wrong according to utilitarian ethical theory. That would also be wrong according to Kantian ethical theory, because that would not a rational thing to do. Also, it is not rational to oppose a discovery or research due to the possibility of its misuse. In instant, appealing to Kant’s first formulation of categorical imperative, we can apply same criteria to atomic research. The atomic energy can be misused
through atomic bomb. Is this means that we would have been better off without the discovery of atom or atomic research, which lead to the electronic age, computers, and the Internet?

So, the right thing to do is rationally prevent possible misuse of genomic (or any) research instead of foster the culture of fear about the possible misuse.

**Genome Information Misuse.** Genome being the *blueprint* of life, the genome sequencing of a person reveals substantially more than just a globally unique ID. As discussed in section 2, genome sequencing essentially reveals the person's biology including ethnicity, susceptibility to disease, and psychological predispositions. This personal information could be of great value, and thus have a potential risk of abuse or misuse by many parties including insurance companies, employers, politicians, and governments. This genome information can potentially also be misused for profit.

One may construct an argument in favor of the abuse or misuse of genome information based on ethical egoism theory according to which each person ought to pursue his or her own self-interest exclusively. Theory of subjectivism and cultural relativism may also be used to argue in favor of this misuse genome information. In contrast, misuse of genome information would be morally wrong according to a number of ethical theories such as social contract theory (for lack of mutual benefit), utilitarian theory (because exploitation of many by the few would decrease and not increase the aggregated happiness), and Kantian theory (for using people a mere means to the end).

Because of the great value of genome information, it is natural to ask if it should or should not be stored in databases.

**Permanent Storage of Information, Genome Databases.** The issue is permanent storage of genome information in both public databases such as those used for research and
private databases including those owned by hospitals and government agencies. It is important to realize that from these databases, it is possible to identify individuals even if they are kept ‘anonymous’ [7]. Such identification is can be made by combining the genome information with other publically available information. Therefore, the concern is that the individual genome information can be stolen from these databases and abused or misused.

In defense of these databases, one can say that the methods to extract useful information from the raw data files (stored in the databases) are complicated (specialized) enough that they are beyond the reach of most laypersons. However, most of the threat comes from powerful individuals and corporation who can hire experts. The threat is even more severe when the abusers are the owners of these databases such as governments and corporations. So, people (whose information these databases hold) may be used a means, which would be morally wrong, e.g., according to the Kantian theory. Even such databases set up by governments against terrorism and crime, which may includes the information about innocent peoples, would be morally wrong. Building on this argument, one may be tempted to suggest that no such databases should be setup.

However, some public databases are setup to facilitate scientific research. Multiple scientific and technological facts ant factors combined give rise to the need of public (shared) genome databases for use by researchers around the world. Without these databases, most of the research in this field would halt, and the remaining would slow down. This will diminish or reduce the common human benefits expected from the research, which in turn will rob humanity of the expected happiness or utility. Thus, this action of stopping genome databases would be morally wrong according to utilitarian theory. This will also be wrong according to Kantian theory.
because it would be irrational to shut down these databases. Similar argument could be made in defense of government databases that may be setup against criminals and terrorists.

We, however, would argue that in this case, the right thing to do is suggested by Aristotelian ethical theory. The right thing lies in the middle of two extremes (vices): misuse of the databases and eliminating the databases. This right thing may include wisely deciding (or regulating) whose and what information enters a given database, making information theft hard by law and by implementing proper security, and making sure that benefits from the information are distributed fairly.

Ethical concerns such as information theft, privacy, and profiting from formation are directly connected to the issue of who owns the information.

**Who Owns the Human Genome Information?** Who has the natural ownership right our individual (personal) genome information? The answer to this question should be simple, we. In capacity of a moral person, each of us has moral right to ownership on her (his) own genome information. Natural law theory support this claim of you being the natural owner of information extracted from your body. Ethical theories such as Kantian theory and social contract theory suggest the proper consent from the natural owner must be obtained before any use of genome information.

The real concern here is: Do we, in practice, have that ownership after our genomes are sequenced? This in an important question because as mentioned earlier, the ownership question is directly related to other ethical concerns. For example access to information extracted from an individual genome can be potentially misused. Also, bioinformation from a collection of genomes can be used to make profit.
It should be noticed that at least the data of publicly (government) funded databases is made public for research purpose (not used to make profit). Despite privacy concerns about these databases, which are already discussed in this paper, the research has the great potential of serving humanity. This makes these databases morally right, and ownership should not be a big moral issues if data is collected in a morally right way, e.g., owner consent is obtained.

But, when genome information enters private sector, ownership becomes a serious issue. Because private sector basically functions with the main goal of profit, the ownership issue turns into the question: Are the benefits or profit being distributed fairly? In this case, morality lies in implementing the things such as principle of mutual benefit (social contract theory), no party uses others as mere means (Kantian theory), and profit doesn’t go to only a few at the cost of many (utilitarian theory).

As we mentioned in section 2, the real (or most) scientific and medical value is extracted from a collection of (rather than single) human genomes. This fact should be considered in the fair distribution of benefits or profit. For example, corporations profiting from genome information may be taxed, and the tax money may go to common good such as education, environment, and scientific research. A set of ethical theory such as utilitarian, Kantian, and social contract support fair distribution of the benefit and profit from genome information among all parties.

**Genome Sequencing as Group Bias Tool.** The concern is that the genome sequencing can potentially be used as a tool against women and various minority groups based on specific genes, race, culture, disabilities, sexual orientation, and lifestyle preferences. In different countries possibly different groups of people may be discriminated based on right or wrong
interpretations of their genome information. This can be done in the name of science and humanity.

As a matter of fact, we had been there before in a very similar situation during the period of Eugenics movement.

5. Learning From History: Don’t Forget Eugenics

As we are entering the “gene age” through DNA sequencing and other research techniques, the ethics, in general, reminds us of a tragedy in the field of genetics that unfolded in early decades of the 20th century. The tragedy, historically remembered by the name of eugenics (or eugenics movement), started with Francis Galton (a cousin of Charles Darwin), who coined the term *eugenics*, and ended with Adolf Hitler. The field of eugenics started off as a research effort to apply Mendel's genetics to the inheritance of human traits. Nevertheless, from the very beginning, its main goal was “to breed better human being by encouraging the reproduction of people with ‘good’ gene and discouraging the reproduction of those with ‘bad’ genes” [8]. Based on a common ‘diagnosis’, people with disabilities and members of racial and ethnic minorities were often declared to be with ‘bad’ genes and hence inferior. Eugenics quickly went haywire as different sections of society ranging from prestigious universities, to governments across the world, to racist organizations joined the movement with different intentions. For example, in U.S. laws were made (or old laws were re-enforced) to bar interracial marriages, to allow forced sterilization of certain groups, and to restrict the immigration of certain groups based on race, ethnicity, and region. In Germany, the Nazi Government implemented its genocidal agenda in the name of eugenic principles by referring to American leadership in implementing those principles. In nutshell, the field of eugenics that was started as an effort to apply Mendel's
genetics to human species, quickly went haywire, and after years of harm and unjust ended with Adolf Hitler.

In light of ethical theories, we should continue genome sequencing and the related research, but as a society must remain on guard to prevent similar tragedy.

6. Conclusions

In this section, we present a summary of conclusions obtained by applying ethical theories to ethical issues in human genome sequencing. As far as the dual research issue goes, while steps should be taken to prevent its possible harmful use, genomics research must continue. Regarding ownership, ethically, an Individual have the ownership right to his/her own genome information. Recognizing and respecting this right, proper consent of the owner must be obtained before any use of genome information. Furthermore, genome databases are necessary for research. But steps must be taken to ensure privacy and only proper use of information.

Profit from genome information should be fairly distributed among all parties. Part of profit should go to common good. Last but not the least, learning from history of the eugenics movement, we should be on guard to make sure that genome sequencing is not used as a tool to commit injustice against any individual or group.
References


Appendix A: Some Ethical Theories in Nutshell

In the following, we present a very brief account of ethical theories referred to in this paper [5].

**Divine Command Theory.** In this theory, God decides what is right or wrong. Therefore, if the actions are in accordance with the God’s commands, they are morally right, otherwise they are morally wrong.

**Aristotelian Theory Virtue Ethics.** According to the theory of virtue, goal of morality is to achieve *eudaimonia*, meaning human flourishing, success, or happiness. Success can be achieved by the proper functioning of things, for which virtue is necessary. Virtue is a mean between two extremes (vices): one of excess and one of deficiency. Virtue is a trait of character that is manifested in natural (habitual) in actions. Because virtue leads to success, it is good for anyone to have.

**Natural Law Theory.** In this theory, actions which are consisting with natural law are right, and those which are not, are wrong. The world is a rational system with its each component having a special purpose. The very concept of *natural law* and *rational system* links this theory to science. Actually the word *science* (and hence *scientist*) was not in use before 19th century. Until then, scientists were called natural philosophers. So, it should not come as a surprise that Aristotle who often is cited as the first to describe the concept of a natural law in detail, is also known to be the first physicist.

**Kantian Ethical Theory.** The core elements of Kantian ethical theory such as universal nature of principles, rationality, and superiority of human over any other kind of creation are summed up in Kant’s two formulation of *categorical imperative*:

“Act as if the maxim of thy action were to become a universal law of nature.”
“Act that you use humanity, whether in your own person or in the person of any other, always at the same time as an end, never merely as a means.”

**Social Contract Theory.** According to this theory the purpose of morality is to make social living possible. This morality lies in the mutually beneficial social contract to which all parties agree. In other words, morality consists of a set of rules accepted by rational people that others accept too.

**Utilitarian Ethical Theory.** According to the utilitarian theory, actions are right in proportional to the happiness they tend to promote, and wrong in proportional to the reverse of happiness they tend to produce. Universal nature of this theory is two-pronged: happiness (or sorrow) integrated over all affected individuals is considered, and rules applied to a class of actions to implement the principle of utility should always be followed in that class of actions.

**Theory of Ethical Egoism.** This states that each person ought to pursue his or her own interest exclusively. Actions that serve your self-interest are right for you.

**Theory of Cultural Relativism.** According to this theory, the folkways in a given culture are the morally right way. In other words, the rights or wrongs are determined by the standard set up in one’s society. Because standards vary from culture to culture, there are no universal moral truths.

**Ethical Theory of Subjectivism.** In this theory, there is no right or wrong objects. Moral judgments are mere statements of facts about our feelings or attitudes. Therefore, moral disagreements are disagreements in attitude and not disagreements in beliefs.