THE HUMAN GENOME PROJECT
AND HUMAN DIGNITY

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What is it that ultimately justifies scientific projects which cost millions of dollars? What ought to be done to ensure that the knowledge and power gained through scientific research be used for man’s ultimate good? What precautions ought to be taken to make sure that this knowledge and the power which stems from it, is not abused to man’s detriment? In this article I will attempt to examine these basic questions with regards to an area of research which will certainly revolutionize the whole of medicine: the Human Genome Project.

The aim of the Human Genome Project is to decipher the entire human genetic code. This is an immense task: Human DNA is calculated to be about two meters long and 1 nm\(^1\) thick. It is made up of about three thousand million nucleotide bases.\(^2\) To isolate a genetic sequence within the human DNA is a very difficult enterprise: one \(\beta\) chain of haemoglobin contains 146 Amino Acids. Each Amino Acid requires three bases. Therefore the \(\beta\) chain is coded by a chain of 438 nucleotide bases on the DNA. This is equivalent to finding a 1.5mm segment within two kilometers!

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As part of the Licentiate, he has presented a short thesis under the guidance of Professor Brian Johnstone C.Ss.R. The title of this dissertation is: The Human Genome Project in the Light of a Christian Anthropology (Rome 1993). The essay presented here could be considered to be a summary of this thesis.

1. \(1 \text{nm}=10^{-9} \text{m}\)
2. This sequence would fill one thousand five hundred volumes of one thousand pages each with two thousand characters in each page.
The Human Genome Project would necessarily require complex techniques for: the gradual identification of all the human genes, the mapping of their precise locus on the chromosomes, and sequencing all the DNA contained in all the chromosomes. Up to the year 1989 only four thousand five hundred genes of the circa one hundred thousand genes have been identified. One thousand five hundred of these have been localized in their respective chromosomes. Only six hundred of these have been cloned and sequenced.

Pinpointing the precise location of a gene on a specific chromosome is called mapping. Two kinds of maps are made, which could then be superimposed: Genetic linkage maps, which locate DNA markers or signposts at closely spaced intervals along the chromosomes and Physical maps which indicate the exact distance between these markers.

The tendency of some genes to be inherited together led to the discovery that these genes are linked. This means that they are found close together on the same chromosome. Until recently, only genes responsible for readily visible traits could be mapped by linkage. The recent discovery of markers, called RFLPs has made it possible to identify genes even when the symptoms of a disease which they cause are not present.

Linkage mapping relies on a biological process known as “crossing-over” or “recombination”. When recombination takes place, genes that are far apart from each other are more likely to become separated from each other, while genes that

8. Linkage is the association of two or more non-allelomorphic genes, so that they tend to be passed from generation to generation as an inseparable unit and fail to show independent assortment (as held by Mendel’s second law). This is due to the fact that they are on the same chromosome.
9. Restriction fragment length polymorphisms (RFLPs). These are due to variations in DNA nucleotide sequences which are apparently without phenotypic effects. These RFLPs occur very frequently — about once every two hundred base pairs, throughout the whole human genome. These changes in base sequence mean that the fragments produced by a particular restriction enzyme will be different lengths in different people. Cf Alan E.H. Emery/Robert F. Mueller, Elements of Medical Genetics, (Churchill Livingstone; Edinburgh, London, Melbourne and New York, 1988) 41.
are closer together are more likely to travel together. Thus the linkage between genes or between genes and RFLPs can be described as distances between them.\textsuperscript{10}

Physical maps give the actual distances in the numbers of nucleotide bases between genes on a chromosome. The map with the highest resolution will show the location of genes or markers on the chromosomes as measured by the number of nucleotide bases between them.\textsuperscript{11}

With all the information obtained from Genetic linkage maps and Physical Maps a "Genome library" may be compiled which can be used to identify and localize specific genes within the chromosomes.

Sequencing involves the decoding of the biochemical information down to the so-called "letters" of inheritance. This information is encoded in the sequence of the nitrogenous basis along the DNA. Several sequencing methods have been developed and others are being developed every day.\textsuperscript{12} Sequencing DNA manually is very expensive, time consuming and tedious.\textsuperscript{13} Recently automatic DNA synthesizers and sequencers have been developed. Japan has taken the lead in developing this technology.\textsuperscript{14}

The costs of the project depend on the way it would be programmed and the development of new technologies. With the technology available today the cost of sequencing the entire genome is estimated to be around six thousand million U.S. dollars, about one dollar for every base pair. This may be reduced to one thousand million dollars using the automated sequencers and synthesizers developed in Japan.

\textit{How ought the H.G.P. be evaluated?}

The objective which scientists usually put forward to justify the HGP may be summed up in the following statement:

...to create an encyclopedia of the human genome — a complete map and

\begin{enumerate}
\item \textsuperscript{10} Cf Louis W. Sullivan, \textit{The Human Genome Project: New tools for tomorrow's health research}, (Bethesda 1991) 7.
\item \textsuperscript{11} \textit{Ibid}, 8.
\item \textsuperscript{13} The time required and the cost of the Human Genome Project, as well as the accuracy of the sequencing process, greatly depends on the development of these technologies.
\item \textsuperscript{14} Cf David Swindbanks, "Japan keeps its options open on the genome sequencing project," \textit{Nature}, 334(7 July 1988) 5.
\end{enumerate}
sequence...the sequence will be the ultimate map, a tool useful for all time and a source book for biology and medicine.\textsuperscript{15}

Although this objective is praiseworthy it is insufficient to justify the project. I believe that the ultimate criterion on which the project ought to be evaluated and the knowledge from it applied is whether or not man’s dignity is respected not only in the working out of the project itself but also in all its applications. So that those applications which do not help man or go against individual people are immoral, while those which respect the dignity of all the human beings involved are moral and laudable.

\textit{General criterial with which the HGP ought to be evaluated.}

A. Each human being ought to be treated as an end and never as a means. This implies that the benefit which can be obtained by one or any number of persons can never justify the killing or any other kind of harming of another or any other number of other human beings. Special concern must be given to protect the good of the weakest persons involved.

B. Scientific and medical knowledge obtained through the Genome project must be used to promote and not inhibit the self-transcendence of the subjects (the scientists) and objects (the persons on whom experiments are carried out) of the research. The same holds for all the persons who would ultimately be affected by the results obtained through the research.

C. The medical technology which results through the HGP can never be used to change essentially the reality of any human being. It may only be used to cure human beings or improve their humanity. Where “improve” means promote their self-transcendence.

D. The human body of each person involved must be respected as an integral dimension of the person. This means that it may never be manipulated as a means towards an end but always for its own good which is the good of the person.

E. Human beings are of equal value and dignity therefore the technologies derived from the HGP can never be used when their use leads to the creation of an elite class of human beings or causes essential division among men. These divisions would lead to further injustices and evil.

\textsuperscript{15} McKusick, “Mapping and sequencing the human genome,” 913.
How the HGP could enhance human dignity

The HGP will greatly increase man's knowledge of the human body and therefore of human nature. This understanding may be used to enhance human dignity in two ways. It may lead to a greater appreciation of the greatness of the human mystery and therefore inspire people, especially those involved in health care, to respect each human being more by treating each person's body with the dignity it deserves. Secondly the knowledge reaped by the project promises to form the basis of great advances in medicine and thus enable cures to be found for diseases which oppress man. These diseases are the cause of suffering and therefore are a threat to his dignity.

The biological data on man obtained from the project could provide:

a. Further evidence of the complex union of body and spirit of the human being.
b. Man with a stronger awareness of what human freedom is.
c. People with a greater awareness of man's intimate relationship with nature.

The analysis of the human genome may also bring into sharp focus the reality of man's uniqueness and his special place in nature. He has a genome which is different from the genome of any other species. Related to this is the fact that this analysis will probably provide more biological evidence of the special place which human beings have in nature.

Knowledge obtained through the HGP could help to remove racial prejudice and racial discrimination. Every human being of whatever race is shown to have the same basic genetic structure so that all share in the same basic common human nature. No human being can have grounds for dismissing another as non-human or essentially inferior.16

Another prejudice which the HGP may help to remove, albeit with far greater difficulty, is the view that the human zygote, is not a human individual. Present knowledge in human genetics and embryology is already indicative of the fact that the zygote and early embryo is a human individual and therefore must be treated with the respect worthy of human beings. The likelihood is that the vast amount of knowledge which is expected to be obtained through the HGP will further strengthen the evidence behind this view and therefore could help to generate respect for the early human embryo.17


17. Unfortunately what is known through the HGP is often used for prenatal diagnosis with the express
The HGP and advances in Medicine

Some of the advances in medicine which the HGP has made possible or will make possible include:

1. Important mendelian disorders have been mapped, including Huntington’s disease on chromosome 4, adenomatous polyposis of the colon on chromosome 5, cystic fibrosis on chromosome 7 ...

2. Several diseases are already understood at a gene (DNA) level. In these cases the nature of the genic lesion that produces the clinical disorder is identified. These include: sickle cell anemia, fragile X syndrome, cystic fibrosis, duchenne muscular dystrophy.

3. While gene discovery does not directly lead to treatment or cure, it always means better gene and disease understanding as well as more accurate diagnosis.

4. Without a means to identify the defective gene responsible for a monogenetic disease a genetic counsellor would have to rely on imprecise methods in order to calculate the risks which other members of the family have of being carriers of the disease.

5. It is theorized that, in the future medicine will be molecular and genetically based. Preventive medicine is likely to expand greatly.

6. Mapping and sequencing studies have provided extensive support for Theodore Boveri’s theory of cancer. It is probable that specific changes in the DNA of tumors will be the basis of tumor diagnosis, staging, prognosis and therapy.

7. Data obtained on genetic polymorphism could also be used to identify persons through the so called “DNA test”. This would be useful, for example, in the course of criminal investigations.

8. Work on the genome project has already contributed to the invention of therapy aim of selective abortion. Those embryos found to have a genetic defect are often aborted. This is far from taking HGP results as the basis for a greater respect for the human embryo.


19. Including prenatal diagnosis for embryos with a high risk of a genetic disease. A discussion of the moral value of this diagnosis is presented below.

techniques for some somatic cell genetic diseases.\textsuperscript{21}

9. At present there is clear evidence that genetic factor(s) are major components in the mechanism of the following: birth defects, aging and several complex diseases such as schizophrenia. The HGP could provide the necessary genetic information to test this evidence and possibly, in the longer term, to work out new cures.

\textit{These advances in Medicine and human dignity}

It is interesting to note that most authors who write about the HGP discuss the medical advances of the project and hardly ever link these advances to man and the promotion of his dignity. This is not surprising since the methodology of modern medicine is restricted to the empirical observation of diseases, the scientific research of their causes, and the development of therapy to overcome or control these causes. However, the subject of the disease, the person who suffers the disease, is often not considered.\textsuperscript{22}

As has been pointed out above a human person has “absolute” (unconditional) value and therefore demands unconditional respect. This means that medicine, if it is to be human, must primarily seek to help the person who suffers the disease and only on account of this motivation strive to find the causes underlying the disease involved.

In this context it is clear that the advances in medicine, which the HGP promises, have far more value than that constituted by great advance in science or in the fight against disease as such. Their fundamental value is surely derived from the fact that through it many human beings now and in the future are offered the means to be freed from the oppression and dehumanization which genetically related diseases threaten them with.\textsuperscript{23}

\textit{How the HGP could endanger human dignity}

Scientists involved in the project are often afraid of exposing the real dangers


of the project and they attempt to play down the real grave dangers to individuals and to humanity if the knowledge obtained by the project were abused.\textsuperscript{24} While it is true that unfounded apocalyptic fears may be propagated from various quarters, there is ample and objective evidence which points to real dangers to man's dignity if the findings of the HGP are abused. It would be most irresponsible to ignore these dangers.

A. The biological data derived from the HGP could be misused to enhance distorted views of man.\textsuperscript{25}

Genetic \textit{reductionism} and genetic \textit{determinism} are both views which go against human dignity because they present a distorted understanding of man. They do not take human transcendence into account. The first involves the assumption that the human person can be reduced fundamentally to sets of genetic structures and nothing more.\textsuperscript{26} Genetic determinism implies the theory that the kind of person one is and everything one does is all determined by his or her genes.\textsuperscript{27} Man is a transcendent being and therefore much more than his genes. A basic dimension of his transcendence is his freedom, although this freedom is the freedom of a creature. If man did not have human genes he would not have the potential for human freedom at all and yet these same genes limit human freedom.

B. The danger of misuse of the knowledge gained through the HGP to the detriment of individual persons.

\textit{Patenting the human genome}

Is it illicit to patent human nucleotide sequences? The HGP, and the information from it, must serve the well being of each individual human being and humanity. If it is used to satisfy the interests of a few individuals or nations it would contribute to the already very unjust distribution of wealth within a country and between countries. Furthermore, the knowledge that would be monopolized if patenting would be allowed, is very intimately human: the human genome, which constitutes a second human anatomic map. Surely it is by nature the property of the whole of humankind.\textsuperscript{28} The practice of patenting the human genome is the first step

\begin{itemize}
\item \textsuperscript{24} Cf B.F. Mackler, et al, Ibid, 154.
\item \textsuperscript{25} Cf Johnstone, "Genetics from now to 2020," 5-6.
\item \textsuperscript{27} Cf E.O. Wilson, \textit{On Human Nature}, (Harvard University Press; Cambridge 1978).
\item \textsuperscript{28} Cf Marga Vicedo, "The human genome project," \textit{Biology and Philosophy}, 7(1992) 262.
\end{itemize}
to misusing this knowledge since it gives too much power to the few.

The problem of privacy

Through the HGP knowledge of an individual’s genetic defects which are the direct cause of a genetic disease or at the base of a predisposition for a disease will be easily available. However respect of the dignity of the individual person involved implies that:

a) Each individual has the right not to be compelled to know about his/her own genetic defects. The right of self-determination of the subject ought to come before economic considerations.

b) The communication of such knowledge of these defects without the free and informed consent of the individual/s concerned amounts to the breaking of “a natural secret” since it touches the physical basis of one’s personality, and, if possessed by others, could expose the individual to exploitation.

c) Any genome analysis must be used for the well being of the patient and his/her family.

d) Personal genetic identity cards ought not to be required by the State or private companies. 29

While the privacy of the individual ought to be respected, genetic information always has implications for other peoples’ welfare, including spouses, children, and extended family members. 30 At times there may be legitimate cases in which the welfare of these persons may imply a certain obligation for the person with a serious gene defect to disclose his situation for the good of these persons.

Discrimination based on genetic risk may occur in two ways: i) The information may be used unfairly to deny otherwise eligible individuals opportunities for adequate health care coverage, life insurance 31 or employment. 32


31. Insurance companies trample on the rights of their clients when they attempt to eliminate the risk involved when the clients concerned have the predisposition of a disease. Having a genetic disposition to a disease does not necessarily mean that one will get the disease. Insurance companies are morally bound to cover the risk a client may have of contracting a disease. They have no right to require information on his/her predisposition to a disease.

32. While employers have the duty to ensure that workers are not given a job which is directly
ii) It may stigmatize individuals socially. All forms of discrimination must be avoided at all cost. Persons who suffer from genetic diseases, indeed any person suffering from whatever disease, must be treated with unconditional respect. In fact the graver the disease the person suffers the more respect he/she is due as a person. Eliminating misinterpretation of genetic defects and diseases which often occurs by patients themselves, their families and the public in general undoubtedly would help reduce discrimination. However, this is certainly insufficient. There is the more fundamental need that peoples’ attitude change. And that laws be made which reduce the discrimination. Everybody must do his/her utmost to respect the persons who suffer from genetic defects and diseases. In the concrete, all must do what they can to ensure that society does not discriminate against them.

The HGP, Prenatal Diagnosis and Selective Abortion

Prenatal diagnosis in itself does not constitute a threat to the respect due to the embryo, except that a small risk of spontaneous abortion is always present. When there is a real risk that the fetus is suffering or could suffer from a genetic disease and the diagnosis shows that this is not the case, it is certainly very helpful to tranquilize the parents’, especially the mother’s, anxiety. When, on the other hand, the disease is shown to be present it could help the parents to prepare themselves for the defective child in the best possible way. In certain cases it would probably be possible to commence therapy a little after birth thus preventing the baby’s death or the deterioration of its health.

There is strong biological evidence to suggest that the human embryo from the moment of conception is a human individual and must be treated with the respect due to a human person. This attitude of reverence towards nascent human life is unfortunately lacking in contemporary western society. Prenatal diagnosis has
detrimental, they have no right to demand genetic screening which would individuate genetic traits in prospective workers who are prone to disease not correlated to the work in question. Cf Pascali/D’Aloja, “Il progetto genoma e le conoscenze sui geni normali e patologici dell’uomo,” 231.

33. By promoting better understanding among the public about the true nature of human genetics and involving the public in deliberations over its use. Cf Ruab, The Ethical, Legal and Social implications of Human Genome research, 8.


35. Cf ibid, 323,324.
already been extensively used with the specific end of terminating those pregnancies in which the fetus has some genetic disease. The prevailing mentality exerts enormous pressure on women to have prenatal diagnosis in order to terminate the pregnancy when there is a sign of abnormality.

**The HGP and Eugenics**

The new possibilities of eugenic control offered through the great advances in genetics has already generated a great deal of fear among many people, including scientists working on the HGP. In the light of Nazi atrocities these fears are not surprising. Daniel Callahan is convinced that researchers have a duty to “use moral imagination” to predict what might be the application of their findings.

**The danger presented by racial discrimination**

Some would dismiss Nazi science as merely mad science. However the involvement of so many a mainstream authorities and leaders in medicine, public health and science in a technologically and scientifically advanced nation such as Germany cannot be dismissed as merely ‘fringe’ or ‘peripheral’. The horrors of Nazi Germany were not enough to dissuade important figures in the scientific world from continuing to voice their recommendations for eugenic programmes. The civil war in Bosnia and other parts of the ex-Yugoslavia and the ex-Soviet Union, their horrendous crimes of genocide and the overt practice of a systematic policy of “ethic cleansing” have blown up the myth which has up to recently been strong in the West: that racism is a thing of the past or at least that this form of inhumanity

36. Indeed in some countries prenatal diagnosis is used to show whether the fetus is male or female and the perfectly normal fetus with the unwanted sex (usually female) is selectively aborted.

37. Cf Angelo Serra, *La Consulenza Genetica. Lecture Note*, (Università Cattolica del Sacro Cuor; Roma) 4.

38. Cf section below on racial discrimination.


is on its way out. In line with Daniel Callahan's view\textsuperscript{42} it is relevant to ask: how could contemporary racists use the knowledge which the HGP would soon make available if they had the necessary technology?\textsuperscript{43}

Even if the international community were to find a solution or at least prevent the spreading of the nightmares of Bosnia many precautions would have to be taken so that the genetic knowledge, which would be available in "civilized" societies, would never be used for racial or ethnic discrimination.

Temptations for the abuse of the genetic knowledge obtained through the HGP for racial discrimination with the excuse of social community or public policy goals would get stronger as more knowledge of the genome becomes available to governments. To safeguard against such temptations Caplan\textsuperscript{44} suggests the following practices:

1. Genetic screening and testing should be avoided except in so far as it is undertaken with the goal of benefiting the individual being tested or the group of which that person is a member at present or in the future.

2. Each person tested must do so freely, consenting to do so after being adequately informed on how the material would be used and on the risks involved that the information might be misused if it got into the wrong hands.

3. In line with the sole purpose of seeking the good of the group in question. The traits to be selected for identification and classification of a group must be those traits which are incapacitating, disabling or damaging to the members of the group rather than merely what is characteristic, distinctive or typical of the group in question.

4. The rights, interests and dignity of individuals must, at all costs, be protected against the desire of the community or the State to obtain information in order to create social policies which would be of benefit to the majority but at great cost to the few.\textsuperscript{45}

\textsuperscript{42} Cf section above.

\textsuperscript{43} Cf Caplan, "Race, Ethnicity and Heredity," 14.

\textsuperscript{44} Ibid, 12-14.

\textsuperscript{45} This is in line with criterion A, presented on page ? above. Each human being ought to be treated as an end and never as a means.
The danger of destroying human nature

The dream of producing a super race carries with it the danger of manipulating human beings, transforming them into "objects" planned and controlled by others. This would go against man's dignity as a "subject" with a capacity for self-determination. Man's personal identity is intrinsically linked with his biological nature. His biological nature is the indispensable foundation of his own personal individuality and singularity and an expression of his interior and external liberty. This means that if his psychosomatic identity is modified and manipulated man's personal identity is distorted or destroyed.46

Would it be licit to modify characteristics found in man which are not part of his essential biological design?

Pope John Paul II has on several occasions, supported, in principle, strictly therapeutic interventions aimed at correcting a defect or at healing a malady if the risk taken is reasonable.

Pope John Paul II does not rule out in principle interventions aimed at improving the human biological condition.47 However he affirms that the following conditions must be respected:

1. The biological nature of every human being as a single substance composed of two principles body and soul should be respected. For example it would be unacceptable to attempt to attach human brain to a totally synthetic body.48

2. The developing human embryo may not be used experimentally. Any intervention on the human embryo is legitimate only if it is directed towards the therapeutic benefit of the embryo concerned.

3. Modifications carried out on the genome may not be directed at creating groups of different people49 at the risk of provoking fresh emarginalizations in society.50

48. The body-soul union is an essential part of man's identity. It would therefore be a grave violation against human dignity to manipulate this union in any way.
49. An essential change in man's psychosomatic nature would provoke such emarginalization (cf above).
Conclusion

The scientific knowledge derived from the HGP will give those who possess it great power. This power may be used to promote human dignity or it may be used to its detriment. Has man got the wisdom and the virtue necessary to apply this knowledge only for the promotion of human dignity of each human person and of humanity as a whole? I believe that while the dangers stemming from the abuse of the power gained by the project are very real, and cannot be overcome by wishful thinking or an unrealistic optimism, the HGP ought not to be abandoned for this reason. Instead the best way to ensure that this power is used for the integral good of man, is to create the right conditions for it to be channeled in the right direction. These conditions involve two main areas: the personal moral strength of the scientists working on the programme and the development of institutions which are capable of guiding practice and restricting abuse. 51

In his book *Science, faith and Society*, the eminent scientist and philosopher Michael Polanyi points out that the ultimate safeguard of freedom (from totalitarianism) is conscience and faithfulness to tradition. Polanyi challenges scientists to form their consciences by aligning their transcendent beliefs to their rightful object: love, justice and truth.

Good institutions presuppose a broad basis of thrust; pursue the common good and promote justice as participation. 52

What kind of international institutions could be created in order to prevent the HGP from being dominated by the interests of single powerful nations? How could these institutions protect the interests of developing nations?

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