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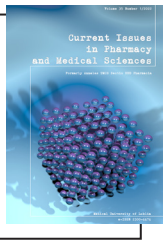

Legal protection of the human genome – excessive or disproportionate?

Bozena Plonka-Syroka[1], Marek Stych[2*]

[1] Department of Human Sciences, Faculty of Pharmacy, Wroclaw Medical University, Poland
[2] Institute of Law and Economics, Pedagogical University of Cracow, Poland

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ABSTRACT

Throughout the 20th century, life expectancy in developed countries considerably increased from 40 to 70 years. This stemmed mainly from the technological advancements in medicine that have been taking place since the end of the 19th century. Although the development of contemporary clinical medicine is undoubtedly beneficial to public health, it may also entail certain hazards. Hence, both in international and national law systems one can find legal regulations setting the limits of this development and taking into account the principles of public safety in its contemporary understanding. Both the human being as a whole and the human genome are protected *de jure*. The article attempts to answer the question whether the currently binding genome regulations are excessive or disproportionate. The authors use the dogmatic-legal and the theoretical-legal methods in the study. The article ends with conclusions.

INTRODUCTION

The provisions of generally applicable law constitute a certain kind of barrier against the unlimited development of medicine [1]. Examples of such a barrier are the legal boundaries set for research in the field of genetic engineering[1]. Medical considerations apart, genetic engineering consists in manipulating the genomes of organisms in order to give them new, desirable characteristics. The genes that are harmful to human health can be eliminated and the beneficial can be added. In the long term, leaving more freedom for manipulating the human genome may contribute to the creation of a so-called "better world", where severe genetic diseases are eliminated or reduced, parents might make their dream to have healthy children come true (advocates of genetic manipulation claim this possibility should not be criticized), and humans will have the opportunity to improve the quality of their lives, regardless of age (treating age-related diseases through genetic engineering is believed to bring tangible results particularly in this group of people).

The question arises whether it is necessary to introduce legal regulations in this area, and also whether a legal system providing for the genome's protection is autonomous in this respect or whether it is an instrument of a specific policy. It is impossible to give unequivocal answers to these questions, as many scientists hold a diversity of opinions here. This stems not only from their identification with a specific ideal of science, but also from a given political ideology or worldview influenced by one of the religions.

The authors of this paper advocate the concept of autonomy in lawmaking. Modern lawmaking is undoubtedly autonomous. It has its own specificity, principles and basic concepts on which it bases its norms. Moreover, it is strongly embedded in society, which is obvious to historians dealing with political and legal doctrines. Throughout history, lawmaking has been a tool for creating statutory foundations allowing for the implementation of specific policies, the general direction and assumptions of which are considered appropriate by the entity ruling in a given area. Historically shaped legal systems are based on different foundations, and so are their regulations pertaining to specific issues. The purpose of this article is to analyze contemporary Polish legal provisions regulating genetic research and therapies. It attempts to answer the basic question: Can excessive legal protection of the human genome halt further rapid development of medicine? The article employs the dogmatic-legal and the theoretical-legal research methods. The article ends with conclusions.

Legal protection of the human genome

The human genome is legally protected at three different levels: political, legal and ethical. This order is not

[1] The first success of genetic engineering was the transfer of the human gene encoding insulin into bacterial cells performed by Stanley Cohen and Herbert Boyer in 1973, as a result of which bacterial cells began to produce the human hormone

* **Corresponding author**
e-mail: bozena@plonka-syroka.pl

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accidental, as the shape of the law passed at a given time in a given country is basically influenced by a specific system of political relations. The adoption of particular regulations depends on a parliamentary majority willing to pass laws in line with the program assumptions of only their party. The parliament's effectiveness in making laws depends on its ability to pass them in both its chambers (in most countries, legislative chambers are divided into lower and higher ones), the ability to convince the head of state to sign them, and the constitutional court's unwillingness to deem them unconstitutional.

In this article, we will look at the legal regulations regarding genetic research through the prism of positive law.

It seems justified to start from the international law followed in Poland that outlines the protection of the genome [2]. It includes the following recommendations [3]:

- Recommendation of the Parliamentary Assembly of the Council of Europe No. 934 of 1982 on genetic engineering; it introduces the right to life and human dignity and the right to inherit a genetic structure that has not been artificially altered;
- Recommendation of the Parliamentary Assembly of the Council of Europe No. 1046 of 1986 on the use of human embryos and fetuses for diagnostic, therapeutic, scientific, industrial and commercial purposes; Point 10 – "embryos and human fetuses must be treated in all circumstances with the respect due to human dignity";
- Recommendation of the Parliamentary Assembly of the Council of Europe No. 1100 of 1989 on the use of human embryos and fetuses in scientific research – "the human fetus and the embryo are treated in conditions appropriate to human dignity" (Point 3).

The above recommendations provide for special protection through the employment of the notion "human dignity". It does not follow, however, that research on the human genome has to be prohibited, especially that in the future genes responsible for severe hereditary diseases might be eliminated. The benefits for future generations are obvious.

Another act of particular importance is the Universal Declaration on the Human Genome and Human Rights, adopted unanimously by UNESCO member states at the 29th session of the General Conference on 11 November 1997 [4]. An analysis of this international document's provisions allows for the conclusion that it combines the idea of respect for human rights and fundamental freedoms with another important issue, that is the freedom of scientific research. The provisions of Article 11 and 12 are crucial in this respect. Article 11 lays down the extent of the prohibition of "practices that are contrary to human dignity, such as reproductive cloning of human beings (...)". The quoted provision generally prohibits practices contrary to human dignity, which may be interpreted in various ways. This prohibition is further clarified by the prohibition of reproductive cloning [5]. Thus, the Declaration allows so-called "therapeutic cloning". Therapeutic cloning means "the use of cloning techniques to create human embryos in order to use the therapeutic potential of their stem cells" [6]. The interpretation of Article 11 may bring different results, depending on the views of the person interpreting its provisions. For conducting medical research, Article 12 Letter b) of the Declaration plays a particularly important role. The provision emphasizes the need for freedom of research for the progress of science [7]. However, it is essential to outline the purpose of this freedom, that is "to offer relief from suffering and improve the health of individuals and humankind as a whole". Such an idealistic goal as "the health of individuals and humankind" must justify the permissibility of extensive research on the human genome, the aim of which is to eliminate genetic diseases. Another relevant international act is the Convention for the Protection of Human Rights and Dignity of Human Beings with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine[2], which provides for the protection of the human genome in Articles 11-14 of Chapter IV. The act also emphasizes the priority of human rights [8]. As regards the subject matter discussed in the paper, Article 13 of the Convention lays down the limits of permissible intervention on the genome. According to the above provision, a manipulation aimed at altering the human genome may only be carried out for:

1. Prophylactic purposes; the goal of primary prophylaxis or prevention is to take medical measures so that the emergence of defective genotypes does not occur. In the prevention of genetic diseases, it is particularly important to identify the factors that cause mutations in genes, e.g. ultraviolet radiation, high temperature etc. Secondary prophylaxis, on the other hand, consists in detecting a genetic disease as early as possible, i.e. before the first symptoms appear. Prophylaxis can take one of two forms[3]:
   - screening tests – carried out on healthy people at high risk of developing a genetic disease; their aim is to detect possible mutations at an early stage;
   - counselling in genetic clinics – in Poland, there are several dozen genetic clinics. These centers provide both primary and secondary prevention.

Therapeutic purposes; for instance gene therapy [9,10]. Gene therapy consists in introducing foreign nucleic acids (DNA or RNA)[4] [11] into the cells of the body in order

[2] Adopted by the Committee of Ministers on 19 November 1996; the so-called Oviedo Convention. Open for signature on 4 April 1997 in Oviedo, entered into force on 1 December 1999. Protocols:
- Additional Protocol Prohibiting the Cloning of Human Beings, opened for signature at Paris on 12 January 1998 (effective since 1 March 2001);
- Additional Protocol on Transplantation of Organs and Tissues of Human Origin, opened for signature at Strasbourg on 24 January 2002 (effective since 1 May 2006);
- Additional Protocol on Biomedical Research, opened for signature at Budapest on 25 January 2005 (effective since 1 September 2007);
- Additional Protocol on Genetic Testing for Health Purposes, opened for signature at Strasbourg on 27 November 2008 (effective since 1 July 2015)
https://www.coe.int/t/dg3/healthbioethic/texts_and_documents/ETS164Polish.pdf

[3] Agencja Oceny Technologii Medycznych i Taryfikacji Wydział Świadczeń Opieki Zdrowotnej [Agency for Health Technology Assessment and Tariffication, Department of Healthcare Services], *Genetic research using next-generation sequencing technology* (*NGS*): • *clinical exome study* (*panel > 4 500 genes with well-documented clinical significance*), • *whole exome sequencing* (*WES*) – *in the diagnostics of genetically conditioned diseases*, Report on the evaluation of health care services, 5 February.2020, http://bipold.aotm.gov.pl/assets/files/zlecenia_mz/2018/030/RPT/WS.430.4.2018_WES_CES_raport_zaczern.pdf

[4] In the years 1902–1909, A. Garrod discovered that genetic defects resulting in the loss of an enzyme caused hereditary metabolic diseases. S. Cohen and H. Boyer developed the foundations of genetic engineering.

to achieve a certain medical effect. The main goal is to remove the harmful effects of mutations, hence gene therapy could possibly have a positive effect within the upcoming generations;

2. Diagnostic purposes; in this case, the diagnosis and treatment of genetic (rare) diseases is a specific medical problem[5].

The question arises how one should understand the above-cited provision of the Convention and why it is prohibited to manipulate the human genome in such a way so as the effects of such an intervention would be hereditary. First of all, it should be noted that some hereditary genetic changes in offspring caused by manipulating the genome may be beneficial, which seems to be ignored in the Convention. The following may serve as examples:

- change of complexion inherited from ancestors, which may prevent severe skin diseases,
- elimination of birthmarks – subsequent generations have a birthmark in the same place, which is the result of genes; this in turn may contribute to the inheritance of cancer.

Other benefits of altering the genome include the elimination of hereditary diseases passed, for example, from father to son, which include:

- fragile X chromosome syndrome [12] – inheritance of moderate intellectual disability which affects 1 in 1500 men. The inheritance of this disease is accompanied by a phenomenon known as the Sherman paradox. This means that the probability of developing the disease depends on the position in the family tree – in the case of brothers of a healthy male carrier it is 9%, whereas in the case of his grandchildren it is 40%,
- prostate cancer [13] – men from families with hereditary breast and ovarian cancer syndrome (if mothers, sisters, aunts or aunts on the paternal side have suffered from this type of cancer) who have been diagnosed with BRCA1 or BRCA2 mutations.

A linguistic interpretation of the Convention's provision leads to a conclusion that the analyzed restriction means, from the medical point of view, that such an alteration is possible in specific cases. From the patient's point of view, it can be assumed that a desired intervention on the human genome should be aimed at introducing changes that improve genetic characteristics [14]. Hence, it seems correct to assume that there are no clear, objective reservations as to such changes in the genome that would be hereditary in subsequent generations. The benefits for future human beings must be taken into consideration. It is commonly believed that potential parents are particularly interested in having children with favourable genetic characteristics [15]. At this point, it is necessary to mention the two features that are undoubtedly desired by parents:

- child's intelligence – dependent on genes in 70%; American scientists have identified the genes responsible for intelligence – the first gene discovered is on the 6th chromosome [16];
- limiting (or eliminating) the tendency towards the so-called "pathological behaviors", which also has a high degree of inheritance. The gene that has been identified as controlling mental traits is the gene coding for monoamine oxidase A (MAOA) [17]. Thus, mutations in this gene lead to disturbances in the activity of neurotransmitters, which causes rapid and violent responses to stress. Nowadays, medicine associates individual variants of the MAOA gene with schizophrenia, alcohol and nicotine addiction, a tendency to depression, or excessive and unjustified aggression [18].

Article 14 of the Convention, as well as the Polish Act of 25 June 2015 on the treatment of infertility [19] (Article 25), lay down specific prohibitions pertaining to the protection of the human being and the genome itself. It is prohibited to:

1. create human embryos for purposes other than medically assisted procreation;
2. create chimeras[6] and hybrids[7] using medically assisted procreation techniques;
3. make heritable alterations to the human genome'
4. create embryos whose genetic information in the cell nucleus is identical to the genetic information in the cell nucleus of another embryo, fetus, human, cadaver or human remains.

While the prohibitions 1, 2 and 4 seem reasonable, the hereditary changes in the genome, as it has been said before, may bring certain benefits for certain people and larger populations. The complete elimination of genes responsible for severe genetic diseases will undoubtedly bring certain benefits to all mankind and improve the quality of human life – and quality of life is an element of human dignity. As inferred from the linguistic interpretation of the analyzed legal provisions, the use of genetic engineering techniques in order to select the child's sex is forbidden[8]. The question whether an exception resulting from one's desire to have a healthy child can be allowed in this case seems perfectly justified. The answer is yes if the choice of sex allows for preventing a serious hereditary disease that affects only:

Its advent allowed for the treatment of this type of diseases at the very root level, i.e. at the DNA sequence level

[5] Regulation (EC) No 141/2000 of the European Parliament and of the Council of 16 December 1999 on orphan medicinal products (OJ L 18, 22.1.2000, p. 1). A rare disease is a disease (or medical condition) that affects 1 in 2,000 people or less in a population. In 2014, in Regulation 536/2014 of the European Parliament and of the Council (EU) of 16 April 2014 on clinical trials of medicinal products for human use, and repealing Directive 2001/20/EC (OJ L 158, 27/05/2014), the definition of an ultra-rare disease was introduced - it is a disease affecting no more than 1 in 50,000 people. The above morbidity rates, as well as the definition itself, should not be treated as universal, e.g. in the USA, this definition was introduced as early as in 1983 and its morbidity rate for rare diseases is <1: 1250 people. In Poland, rare diseases affect approximately 1.9 million patients (which is about 5% of the population). In the whole EU, there are between 27 and 36 million people suffering from rare diseases. The Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02) recommended the member states to develop and implement plans for the treatment of rare diseases by the end of 2013 at the latest

[6] Article 2 Paragraph 4: chimera – a group of cells composed of cells with different genotypes, coming from more than two individuals of the same species or from different species, where one of the species is human

[7] Article 2 Paragraph 11: hybrid – a cell or group of cells formed from a human reproductive cell and an animal reproductive cell

[8] For example, due to the possibility of choosing the child's sex in the medically assisted procreation (*in vitro*), male children prevail. The ratio of newborn girls to boys has already been disturbed in the People's Republic of China (since the beginning of the 21st century, the ratio has been 117 boys to 100 girls, while the world average is 103-105 boys to 100 girls), mainly due to the opportunities offered by medically assisted procreation (*in vitro* fertilization)

- boys – e.g. Hunter syndrome[9],
- girls – e.g. Turner syndrome[10].

In the EU, one can observe quite varied approaches to the development of modern medicine. The EU states divide into the ones that have and have not ratified the Convention[11]. The latter group includes the states whose national legislation is already more liberal (e.g. the UK), or the states that perceive the Convention's provisions as a potential threat to the values professed by a large part or the majority of their citizens or supported by the ruling political parties[12]. Such is the case in Poland, where it is difficult to conduct advanced medical research.

To sum up this part of the deliberations, it can be said that the protection of the genome discussed above does not pertain to the rights of individual persons. The regulations are intended to protect the rights of the "community of human individuals", i.e. the human species.

CONCLUSIONS

The intervention on the human genome still remains an open question and it is difficult to predict the further effects of the development of genetic engineering. Legal issues and research safety are of particular importance in the sequencing of the human genome. It seems justified that the development of genetics should be accompanied by specific regulations in the statutory law – which is not questioned by the authors of this paper. Based on the above analysis of the legal acts, a thesis can be formulated that not only the human genome itself, but also the research on the human genome needs to be legally protected. The sequencing of the human genome undoubtedly constitutes a milestone in the development of biological sciences and medicine. However, the effects of the research on the human genome extend beyond natural sciences. For this reason, it should be guaranteed, just like every human individual, respect and legal protection due to the potential it carries. This is what the legislator believed when introducing protective regulations. Research on the genome with the use of genetic engineering has been being conducted on an unprecedented scale and is still a novelty in science. This state of affairs is undoubtedly natural, and results from the development of civilization, which prompts researchers not only to use new techniques in their research, but also to define new goals to be pursued. The employment of the latest technological advancements does not have to entail the violation of human dignity, both individual and collective. Regarding law as the guardian of the human genome and invoking the argument of protecting its dignity from violation or loss might lead to the abandonment of research on the practical applications of genetics. Protection needs to be carried out within the legal framework outlined above, which is not questioned by doctors. However, before introducing further restrictive legal regulations, legislators should bear in mind the consequences of their decisions. They should make sure that the introduced bans do not stop the development of science. They should consider whether the benefits resulting from their decisions do outweigh the possible negative effects, and whether such effects always occur. The legal solutions to be introduced should be consulted with a wide group of doctors and specialists in the field of genetics and other basic sciences. This way the protection of patients' rights will apply not only to the human genome, but also to real people who are legally entitled to proper medical care provided in accordance with the highest and latest standards.

In the authors' opinion, regulations pertaining to genome research should be less restrictive. Further attempts to limit human genome research in legislation will result in limitation of research in practice.

[9] Hunter syndrome is mucopolysaccharidosis type II (MPSII). It is a genetically determined metabolic disease. It has a different way of inheritance. It is sex-linked (X chromosome-linked) and inherited recessively. This means that girls who receive the defective chromosome are only carriers and do not have the symptoms of the disease, but they will pass it on to their sons. It is boys that mainly suffer from MPSII

[10] Turner syndrome is a congenital genetic defect that is caused by an abnormality in the structure of the chromosomes. Healthy girls have two X chromosomes - one from their mother and one from their father. In this disease, some (or even all) of the body's cells lack one chromosome. This outcome results from a faulty division in the formation of sperm cells and ova. Even if the second X chromosome is present in the cells, it is usually defective. A genetic error occurs with the appearance of reproductive cells

[11] Poland signed the Convention with the only protocol that existed at that time (concerning the prohibition of cloning human beings) on 7 May 1999. So far, this Convention has not been ratified in Poland, and the other 3 protocols concerning transplantation of human organs and tissues, human research and genetic tests for health purposes have not been signed by Poland

[12] It has been ratified by 22 out of 47 member states of the Council of Europe. Another 12 countries have signed it but not ratified (as of 07/24/2009, source: Council of Europe)

ORCID iDs

Bożena Płonka-Syroka https://orcid.org/0000-0001-8743-4634
Marek Stych https://orcid.org/0000-0003-4359-1085

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