Bioinformatics and Issues of Conclusion
a Contract for Provision of Medical Services

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Abstract. The article analyzes the possibilities of commercialization of scientific research results of the human genome and its positive impact on research activities, provided that the information is properly protected. As shown by the legal regulation of scientific research of the human genome in certain states, the secret of private life (privacy) is the main content of the contract practiced in the field of medical services. Genetic information, although similar to medical secrecy when entering into a contract for the provision of medical services, differs in a public element in connection with the use of digital technologies in scientific research. The requirement to protect genome privacy is a mutual obligation of the parties in the contract. This parties do not always give proper attention to such issue. Legal regulation of genomic research will help to formulate a pragmatic attitude to the problem and find a balance between the risk and benefits of human knowledge.

Keywords: Biomedicine · Contract for the provision of medical services · Genome privacy · Protection of genome privacy

1 Introduction

The new service contracts are related to the research of the human genome in biology through biotechnologies. “One of the main features of our genome is its amazing regulating. Just as some computer folders are inside other folders, and those are in the next ones, and so on, the human genome consists of three billion so called building blocks, which are called nucleotides (there are four types A, C, G and T)” [11]. The comparison of nucleotides with computer folders used by Spencer Wells is not accidental. The fact is that genomic research has helped many sciences, including bioinformatics in developing. The capabilities of biological engineering can be applied to almost all types of cells, providing the creation of genetically modified plants or animals, as well as the creation of cells of adult organisms, including humans. This significantly differs from modern genetic engineering. The practice of research in the eighties of the last century differs in increased accuracy, efficiency and use simplicity. The list of potential applications of biological engineering is almost unlimited: from the ability to modify animals to grow them on a more economical diet adapted to local conditions, to the creation of food crops that can withstand extreme temperatures or drought.
As research in genetic engineering progresses are developing (for example, the development of the CRISPR/Cas9 method in gene editing and therapy), delivery and specificity constraints will be overcome. We will only have to find an answer to the most difficult question from the point of view of ethics, namely: how does gene editing revolutionize medical research and treatment procedures?

Different technologies are merging and enriching each other, and new ways are being developed to implement and use technical devices that track our activity levels and blood chemistry, as well as the impact of these factors on a person’s physical condition, mental health, and productivity at work and at home. According to this statement, 3D production can be combined with gene editing to produce living tissues for the purpose of their recovery and regeneration (this process is called “bioprinting”). This technology is already used to create skin, bone, heart and muscle tissue. One day, printed cell layers of the liver will be used to create organs for transplantation [3].

Researchers have already started creating pig genomes for the purpose of growing organs for human transplantation (a process called “xenotransplantation”, which could not be considered before due to the risk of immune rejection by the human body and transmission of the disease from the animal to the person). Plants and animals can be engineered to produce medicines and other forms of treatment. The day when a cow will be created that produces milk containing the element of blood coagulability, which is absent in people with hemophilia, is on the horizon. In fact, science is developing at such a pace that it is no longer technical, but legal, regulatory, and ethical constraints that stand in the way of progress.

The demand for genomic research in the field of law is evident in the commercial sphere and in the field of criminology. However, the search areas often complement each other. In addition to law enforcement, biological research has become widely used in commercial activities. Commercial genomics is a rapidly growing consumer services industry. It offers its clients a sample of their DNA (a little saliva) to restore their origin, find distant relatives, assess the risks of various diseases, choose a diet and sport. This innocent activity, however, can be a powerful force and lead to violations of human rights. People’s genetic differences are quite diverse, but two main parameters are used to compile databases. The first of them is SNP, or “single-nucleotide polymorphisms”, they are simply “substitutions”. There are about 3 million points in the human genome where different individuals may have different “letters” (nucleotides). The totality of data on these points is a unique genetic portrait of a person. This portrait is made up when genotyping using a DNA chip. Such data is collected in the databases of commercial companies.

In order to avoid human rights violations, according to Fyodor Konovalov, head of the “Laboratory for clinical bioinformatics” (a Russian company engaged in medical genomics), genetic information should be subject to the law on personal data. According to this law, personal data is what makes it possible to identify a person. Previously, it was not possible to identify an individual by genotype, but if such a possibility exists, a legal problem may arise. This opinion, however, is not shared by all participants in commercial relations. So, Colin Fitzpatrick, director of the DNA Doe Project California company, believes that the fears are exaggerated: genomics data is not fundamentally different from all other information that is legally used by law enforcement, and there is no reason to treat genetic tests more scrupulously than, for
example, to publications in social networks. As the number of genetic tests for relatives searching increases, the threat to human rights in the forensic use of genomic research results increases. In this regard, the question of forming the Institute for the protection of genome privacy is increasingly raised [12].

2 Methodology

The methodological apparatus of the presented research consists of the following general and special methods of scientific knowledge successfully tested in the course of fundamental and applied research: philosophical method based on the dialectical method of knowledge; formal logical method that includes analysis and synthesis, induction and deduction, abstraction and generalization, analogy and comparison; system-structural method; historical method; formal legal method of analysis; dogmatic method (study of the dogma of law based on the provisions of existing legal norms, legislation, by-laws and international treaties); methods of interpretation of law, including lexical, systematic, teleological, doctrinal and authentic methods of interpretation; content analysis. The use of philosophical, formal-logical, system-structural and historical methods was a necessary condition for the scientifically justified and methodically verified implementation of scientific research. It was impossible to conduct scientific research of a legal nature without using the formal legal method of analysis, dogmatic method and method of interpretation of law.

3 Results

Bioinformatics is usually understood as the use of computers to solve biological problems. Nowadays, this is almost exclusively the task of molecular biology. The reason for this is that over the past 20–25 years, a truly colossal experimental material has been accumulated on the structure and functioning of biological molecules (proteins and nucleic acids), as an example, it is enough to cite the human genome [17]. Bioinformatics in its current state is a set of quite voluminous sections, as in any other science. If you compare it, for example, with physics, it is quite obvious that a specialist in theoretical mechanics is likely to have some difficulties in understanding the latest articles on quantum physics, and moreover, he probably will not have time to read these articles. The situation is similar in bioinformatics. There are a lot of topics in bioinformatics: 1) the evolution (and not only in the form of “first there were the pithecanthropuses,” but also lesser known issues, such as developments in cancer); 2) search for genetic variants that lead to diseases; 3) fabrication and selection of drugs that bind to certain types of “hazardous” proteins; 4) to study the genes functions, their annotation; 5) structural bioinformatics (manipulation of 2D and 3D structures, such as, for example, proteins or RNA); 6) assembly of genomes; 7) mapping of how all this aspects of proteins/RNA/DNA/fat/clever ideas/workout/Kremlin diet and other react between each other; 8) modeling of complex systems (such as the development of an organism from embryo); 9) neurobiology (or rather the analysis of the data collected by neuroscientists).
The development of bioinformatics as a new branch of knowledge was facilitated by the creation of an international research project that brought together the efforts of scientists from various countries, just as the “Apollo” program provided space research. Laboratories all over the world worked to create a genetic map of a person, to determine the sequence of nucleotides that make up DNA [13]. The project began in 1990, under the leadership of James Watson under the auspices of the US National Health Organization. A working draft of the genome structure was released in 2000, and the full genome was released in 2003, but even today additional analysis of some sections is still incomplete. Two main tools were used to implement the project: DNA sequencing and computer technologies for combining and analyzing (interpreting) of the received information in a database system [14].

A private company Celera Corporation launched a similar parallel project that was completed a little earlier than the international one. American researcher Craig Venter and his firm Celera Corporation launched a similar study in 1998. Unlike the international project, the Craig Venter project was funded by private capital. The parallel project was only for commercial purposes. The firm used a more risky variation of the genome fragmentation method (the shotgun method), which has previously been used to sequence bacterial genomes up to six million pairs of nucleotides in length, but never for anything as large as the human genome, consisting of three billion pairs of nucleotides. Additional legal support was required to achieve commercial goals. The firm has filed preliminary patent applications for 6,500 whole or partial genes. Celera Corporation has also promised to publish the results of its work under the terms of «Bermuda Declaration» of 1996, releasing new data quarterly (the “Human genome” project released new data daily), but, unlike a publicly funded project, the firm does not grant permission for the free distribution or commercial use of its data. In March 2000, US president Bill Clinton stated that the genome sequence could not be patented and should be freely available to all researchers. After the president’s announcement, shares of Celera Corporation fell sharply, dragging down the entire Nasdaq biotechnological sector, which lost about $50 billion of market capitalization in two days.

Competition in researches between the private and public sectors, as shown by the example of genomic research, has had a very good effect on the obtained results. In a public-funded project, competition has forced participants of the public project to modify their strategy to speed up progress. Firstly, the competitors agreed to merge the results, but the union broke up after Celera Corporation refused to make its results available through the GenBank public database with unlimited access for all users. Celera Corporation included data from the “Human Genome” project in its own sequence, but prohibited attempts to use its data for all third-party users. The main sequencing was performed at universities and research centers in the United States, Canada, and the United Kingdom. In addition to its obvious fundamental importance, determining the structure of human genes is an important step in the development of new medicines and other aspects of health care. The results of scientific research conducted within the framework of the international project gave an impetus to the development of a huge segment of market relations, covering various types of medical services. The existence of an active demand in this market raised the question of the
need to form a contract for the provision of medical services using genetic information, as well as a special type of contract for the protection of the genome privacy.

Currently, the list of services provided to consumers in the field of biological information includes services for preimplantation genetic testing, prenatal diagnostics, paternity testing, oncodiagnostics, diagnostics of inherited diseases, chromosomal matrix analysis, and other services. Commercial genomics is a rapidly growing consumer services industry. The basis for providing such services is the relationship between the doctor and the patient. There is little change in the legal regulation of contractual relations between two subjects of civil law. However, the emergence of genomic research introduces its own peculiarities in terms of the formulation of rights and obligations, as well as responsibility for their implementation.

The provision of medical services is associated with obtaining the patient’s consent to perform certain operations, the success of which is associated with the preparation and correctness of decisions made by the doctor. Personalized medicine sets out its requirements for the contractual relationship between the doctor and the patient. One of these requirements is to obtain informed consent from the patient to conduct certain genomic studies [15].

The doctrine of the informed patient consent, developed in UK jurisprudence provides for the doctor’s liability of physical harm caused to the patient if he did not warn the patient of a specific risk of adverse consequences. Traditional civil law institutions are used to ensure responsibility to the patient. For example, the basis of liability is the doctor’s fault in the form of negligence and the relationship between awareness of the risk and the occurrence of the risk. The so-called “causation” of adverse consequences and lack of warning are the criteria for determining the doctor’s negligence. When assessing the availability of informed consent, the English court is guided not only by the fact that the doctor must warn the patient of possible consequences, but also by the fact that the patients must fulfill certain requirements for providing their consent in accordance with the established procedure, i.e. the procedure for providing informed consent by the patient. The emergence of the procedure for obtaining informed consent of the patient is due to the fact that the increasing complexity of medical research changes the criteria for assessing guilt in the form of negligence in medical practice. The change in the assessment of guilt in medical practice is due to a change in judicial practice regarding the standard of information disclosure requirements. Thus, the right, based on the principle of patient autonomy in making decisions and expressing consent to certain actions from the doctor, has become more focused on the details of obtaining a safe and effective medical service. If previously the procedure for obtaining the patient’s consent was based on a simple agreement with the doctor’s recommendation, now this is no longer enough. For a patient to make an informed decision, they need to make an informed decision on all the issues raised as a result of their dialogue with the doctor. This helps to form a contract for the provision of medical services of a new type.

The patient’s obligation to comply with the procedure for providing informed consent to the actions of a medical professional corresponds to the duty of the medical employee to maintain confidentiality with respect to information on the patient’s health obtained as a result of treatment. Judicial practice has introduced the concept of “reasonable expectation” in terms of ensuring the human right to privacy. A reasonable
expectation of patient privacy has become a kind of criterion for evaluating the behavior of a doctor who violated confidentiality and a way to protect the patient’s rights. The duty of a doctor is to keep medical privacy. Only the patient’s expressed or implied consent to the disclosure of information about their health status. The patient’s consent may be the basis for excluding such medical obligations under the terms of the contract [12]. In the practice of concluding contracts for the study of the human genome, the question of the patient’s implied consent arises most often in connection with the widespread use of computer technologies and modern communication technologies, including the internet.

The legislation of some countries where the commercial practice of providing genetic information has been the most developed is aimed at protection of genome privacy. This applies, in particular, to US legislation: in the field of health insurance and in terms of protection against discrimination based on genetic information.

National legislation on the protection of the patient’s genome privacy in the provision of medical services. The law on identity and liability in health insurance [6] and the law on non-discrimination based on genome privacy [7], adopted in the United States at the beginning of the 21st century, indicate the creation of control over genetic information present in commercial circulation. The literature notifies that the law pays little attention to the protection of genome privacy in the database of commercial organizations. The diversity in the categories of actors in “genetic” relationships and the goals pursued by participants in these relationships makes it difficult to work on general principles for the protection of genome privacy [4]. In the United States, where the volume of accumulated information allows you to formulate specific requirements for participants in the commercial turnover of genetic information, two laws were adopted at the federal level, the analysis of which showed that for the purpose of individual protection of the patient’s genome privacy, the requirements for the protection of ordinary medical information are equally applicable. At the same time, it was noted that the peculiarity of genetic information is the presence in it of not only the interest in its protection of individuals, participants of commercial relations, but also public services and society as a whole. “DNA is the concept of a unique human identifier, and on the other hand reveals information on biological relatives” [2].

The presence of a public element in the information itself that allows the use this information for the bad purposes not only of the owner of such information, if it is provided without his permission, but also of a wider range of persons, primarily law enforcement agencies and the entire commercial turnover of genetic information in general. This creates a positive perspective for the legal regulation of scientific research of the human genome through the improvement of the system for collecting, storing and protecting of genome privacy. As stated in foreign literature, “in our media age, the very concept of legal protection of information secrets, or “Privacy”, has changed and become more intense [12]. The Internet does not require people to consent to the dissemination of information about them, but it is thanks to new communication technologies and the activity of citizens, using these technologies, the information is widely distributed. New theories regarding the definition of “Privacy”, as the right of an individual to control the use of personal data in commercial circulation, is only a prerequisite for the formation of contracts for the provision of medical services that include genetic information on the patient.
Since we are talking about contracts for the provision of medical services, the question arises on the nature of people right to control the use of their genetic information. State common law courts analyze claims for infringement of the right to protect genome privacy either as a claim for infringement of proprietary rights or as a claim for damages, but the public nature of the offense is not excluded.

The experience of legal regulation accumulated in foreign legislation is useful for the construction of a new medical care system, which was launched in Russia due to the danger of a pandemic that occurred on the basis of COVID-19. The program for the development of genetic technologies in Russia includes scientific developments that will prevent serious diseases, increase life expectancy, improve the environment, etc. To fulfill these tasks, three world-class genomic centers are being created, representing a consortium of universities, research institutes, and manufacturing innovation companies. Medicine is one of the main directions developed by genomic centers. The question of the legal regime of accumulated genetic information is certainly a central issue in the development of a promising direction of genomic research.

4 Discussion

The problem of involvement of the researcher in the process of conducting scientific work is very important in the regulation of scientific research. Solovyov drew attention to the fact that “the mind or meaning of knowable things and phenomena can be known only by intellect or sense of the cognizing subject, the relation of the subject to everything can exist for us only because we ourselves have an intellect [10]. Otherwise our knowledge would not differ from animals, affecting us reality of private objects and phenomena [10]. To a certain extent, the idea of Solovyov was reflected in a thought experiment of the Austrian scientist Schrödinger, described in the framework of the work “The current situation in quantum mechanics” [9] published in 1935. This work was devoted to the study of the problem of so-called “quantum entanglement” (a term introduced by Schrodinger himself), which allowed the simultaneous existence of several realities.

The uncertainty of quantum reality in a certain extent makes the problem of the cognizing person relevant, since “a person perceiving or passively experiencing this reality as a subject of sense, when a subject of reason determines the meaning of this reality, evaluates it in relation to the principle of unity that he has in himself as his mind” [10]. The Schrodinger cat experiment draws attention to the problem of observer morality in physics, but these principles are even more important in research of biology and in genetics, in particular. Compliance with ethical principles in scientific research should be reflected not only in legislation as formal requirements established by mandatory legal norms, but also in contractual practice (genomic contracts).
5 Conclusion

In 1866 Mendel [8] suggested that the characteristics of organisms are determined by inherited units, which he called elements. Later they were called “factors” and, finally, genes; it was shown that genes are located in chromosomes, with which they are passed from one generation to another” [5]. In the early decades of the 20th century, geneticist Thomas Morgan, who worked in a laboratory in Chicago, used fruit flies as an experimental model for his groundbreaking study. The result of scientific research is the conclusion that the genes are located in the chromosomes located in the insect germ nucleus. By the early 1930s, biologists and medical researchers were able to claim that genes were physical objects, the chemical information blocks strung on chromosomes like beads on a fishing line. The current level of genomic research allows us to suggest a program for the development of scientific research in this area based on the best practices of international research organizations, which meets the requirements of the new concept of the precision medicine development [1].

The US program for the development of research in the field of genomic medicine, presented by Jim Vaught, the president of the International Society for Biological and Environmental Repositories (ISBER), at the 3rd International conference on genomic medicine in Jeddah (Saudi Arabia) on November 30-December 3, 2015, provides a systematic approach to the collection, processing, storage and analysis of bio-samples [15]. The program is particularly important in the transition from the health care system to the precise medicine, because it is with no doubt associated with the transition from the study of diseases as such to the study of specific features of the body of a particular patient [16]. The conditions for the development of modern medical services set the legal experts the task of forming a commercial contract for the provision of medical services that meets the requirement of protecting the individual genome privacy of the patient, which is the basis of the contract.

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