opción

Revista de Antropología, Ciencias de la Comunicación y de la Información, Filosofía, Lingüística y Semiótica, Problemas del Desarrollo, la Ciencia y la Tecnología

Año 35, 2019, Especial N° 21

Revista de Ciencias Humanas y Sociales
ISSN 1012-1587/ ISSNe: 2477-9385
Depósito Legal pp 198402ZU45

Universidad del Zulia
Facultad Experimental de Ciencias
Departamento de Ciencias Humanas
Maracaibo - Venezuela

Opción, Año 35, Especial No.21 (2019): 703-718
ISSN 1012-1587/ISSNe: 2477-9385

Legal and ethical aspects of development of molecular genetic technologies

Lyudmila Voronina[1]
[1]Ural Federal University, Yekaterinburg, Russia
l.voronina@yandex.ru

Екaterina Zaitseva[2]
[2]Ural Federal University, Yekaterinburg, Russia
e.zaitseva2009@yandex.ru

Andrey Nechkin[3]
[3]Ural State Law University, Yekaterinburg, Russia
a.nechkin@yandex.ru

Irina Tuzankina[4]
[4]Institute of Immunology and Physiology, Yekaterinburg, Russia
i.tuzankina@yandex.ru

Abstract

This research aims to understand the concept of molecular genetic technologies and other types of studies relating to innate immunity errors by using the comparative method. The authors borrowed the results of the sociological researches conducted by Russian scientists in the framework of the international research program Genetics and Ethics as well as the expertise of their own specialists of genetic laboratories in Yekaterinburg (Russia, n = 16). In conclusion, it is also advisable to prohibit an unborn child genetic test for the identification of the diseases that may appear in adulthood.

Keywords: Legal, ethical, standards, molecular, genetic.

Aspectos legales y éticos del desarrollo de tecnologías de genética molecular

Resumen

Esta investigación pretende comprender el concepto de tecnologías genéticas moleculares y otros tipos de estudios

Recibido: 10-03-2019 •Aceptado: 15-04-2019

relacionados con errores de inmunidad innata mediante el uso del método comparativo. Los autores tomaron en préstamo los resultados de las investigaciones sociológicas realizadas por científicos rusos en el marco del programa internacional de investigación Genética y Ética, así como la experiencia de sus propios especialistas en laboratorios genéticos en Ekaterimburgo (Rusia, n = 16). En conclusión, también es aconsejable prohibir una prueba genética de un niño no nacido para la identificación de las enfermedades que pueden aparecer en la edad adulta.

Palabras clave: legales, éticas, estándares, moleculares, genéticas.

1. INTRODUCTION

The research topic is determined by the following theoretical and practical problems:

- The underdeveloped concept of molecular genetic technologies in the theory of Russian health management;

- A more specific concept of molecular genetic technologies in terms of innate immunity errors.

The theoretical underdevelopment of the concepts discussed above leads to the underdevelopment of concepts such as the consulting technology based on the diagnostics of severe combined immunodeficiency disorder (SCID) and therapy technologies for SCID confirmation. One of the reasons these concepts are not formulated properly by both Russian theorists and practitioners is their inadequate experience in molecular genetic studies. The authors of the research

reveal the other reason that is the absence of clear and complete formulations of the concepts themselves and the corresponding procedures or actions in Russian legislation.

At the same time, the critical review of the foreign legislation in gene diagnostics and its variations provides the understanding of the list of specific procedures (actions) required for genetic research. The most important thing is that evaluating the foreign experience allows us to determine the possible ways to improve the Russian legislation in the sphere of federal law on the study of the complex human genome. At the same time, the study of ethical standards for molecular genetic studies in different countries, as well as of doctors' and patients' interaction, makes it possible to clarify the existing standards and to realize the necessity of new standards' development and the appropriate means of preventing ethical conflicts.

Thus, the purpose of the current research is the following:

-To study the concept of molecular genetic studies technologies and their variations;

-To analyze the legal and ethical aspects providing the implementation of these technologies;

-To introduce the recommendations to the Russian legislative establishment and geneticists' professional community.

2. LITERATURE REVIEW

The selection of theoretical literature and regulatory legal acts is determined by the goals and objectives of the study, the results of which are presented in this article. For the selection and analysis of the literature, the following databases were used: Scopus, Web of Science, Google Scholar, WHO IRIS (the World Health Organization's Institutional Repository for Information Sharing), and the Russian Science Citation Index database (scientific electronic library: elabrary.ru). To clarify the concept of molecular genetic studies technology and its variations, namely, newborn screening technology and the technology of early diagnostics of severe combined immunodeficiency in newborns, the authors analyze the Russian works devoted to the theoretical substantiation of various medical technologies.

These are works by Pecherskih (2009) (research on the process of medical care for children with malignant neoplasm); Pozdnyakova (2011) (some aspects of the technologies involved in cancer patients' treatment); and Aksenova et al. (2007) (algorithms and sequence of actions in the differential diagnostics of BCG complications, including the modern informative diagnostic methods in vivo and ex vivo). The analysis of the research led to the conclusion that only specific methods or factors influencing the medical treatment results are currently being studied. At the same time, there are no Russian authors' works where these technologies are considered as the set of elements.

Nowadays, the technology of molecular genetic studies for forensic activities is being developed. In 1999, the Ministry of Health of Russia introduced the methodology instructions containing the description of the technological components of the forensic examination of personal identification and kinship determination. The results of the analysis of the theoretical works and regulations provided the opportunity for theoretical justification of the following technologies: newborn screening and the consulting contributed by the general pediatricians to identify or exclude patients with severe combined immunodeficiency. To prove the significance of these technologies, the works of Russian and foreign researchers are analyzed, namely Zakharova (2006), Baranov et al. (2012); Vanzelm et al. (2011); Mahlaoui et al. (2014) and others.

The components of these technologies (newborn screening, the consulting to identify or exclude patients with severe combined immunodeficiency), legal and ethical (recommended) regulations form the basis for the doctors' geneticists and pediatric physicians' actions. The authors of the article analyze the works of such foreign researches as: (Vanzelm et al., 2011; Mahlaoui et al., 2014). In their investigations, they highly appreciate the system of rules, standards, and security means for organizing and conducting the genetic screening programs, which were presented by the Public and Professional Policy Committee of the European Society of Human Genetics.

Tuzankina et al. (2018) declare that one of the most dangerous forms of the primary immunodeficiency, namely severe combined immunodeficiency disorder (SCID), matches the suggested criteria. The analysis of the researches of Tuzankina et al. (2018) led to the conclusion that the newborn screening for severe combined immunodeficiency, as well as the patients with such a diagnosis, consulting, should be regarded as the technologies. In addition, for their realization, the corresponding legal and ethical regulations accepted by the professional community should be formed; furthermore, the appropriate financing for equipment and consumables must be available.

The authors of the article appeal to the study of such federal laws including the Russian Federation federal law on federal regulation in the area of genetic engineering activity, the State registration of genome in the Russian Federation and on personal data, and on consumer protection to estimate the possibilities for molecular genetic studies, including newborn screening. In addition, the works devoted to the study of the legal regulation of genetic research in Russia and abroad conducted by the following researchers are analyzed (Romanovskiy, 2016). The analysis of the legal regulations and theoretical works allows for a conclusion that the laws adopted in the Russian Federation cannot meet the needs in the legal regulation of gene diagnostics and the implementation of the necessary technologies.

To study the experience of legal regulation of gene diagnostics and related molecular genetic studies, the authors of the article refer to

the laws of the United States of America, Germany, and the Republic of Latvia. They conclude that the legislation of these foreign countries provides an integrated approach to gene diagnostics, while at the same time, various approaches to the implementation of different types of gene diagnostics exist that allowed or prohibited ones. The integrated approach allows the participants of the molecular genetic studies to conduct legitimate actions and to avoid unethical ones. The analysis of the fundamental work of Wertz & Fletcher (2012) and Izhevskaya (2006) allow the authors of the article to identify current ethical standards that determine the actions of geneticists, including when interacting with the patients. The development of this theory is presented in the results of an expert survey conducted by the authors of the article.

3. RESEARCH METHODS

The selection of the scientific methods used to achieve the goals of the study is determined by the object of the research, namely the molecular genetic studies technologies in terms of the innate immunity errors. The authors of the article analyze theoretical works devoted to the study of the concept of gene diagnostics and the technology of molecular genetic studies, specifically newborn screening and early diagnostics of severe combined immune deficiency of the newborns. Secondly, the selection of the scientific methods is determined by the subject of the research, namely the analysis of the legal regulations of the molecular genetic research technologies in such countries such as

Germany, the USA, the Republic of Latvia, and the Russian Federation.

The use of the comparative analysis allows us not only to analyze the legal norms in these countries, but also to reveal the experience that can be applied in regulating the specific procedures of the genetic research, such as informing the patients by the geneticists, the consulting doctors' decisions in accordance with the genetic research results, the subsequent use of the genetic testing samples, and the related information.

4. PRACTICAL SIGNIFICANCE AND RESULTS

The analysis of the Russian scientific researches leads to the conclusion that only a few of them provide and implement the theoretical basis for the technologies necessary for molecular genetic research and the subsequent treatment of the patients. There are almost no Russian works in which such technologies are considered as a combination of elements. Basically, scientists analyze particular elements or factors that influence the results of medical treatment. More often, the technology-related concepts are analyzed by the authors studying the specific issues of healthcare management. For example, Pechersky investigates the process of providing medical care to children with malignant tumors. Pozdnyakova (2011) analyzes particular elements of medical technology, especially diagnostic

methods used in the treatment of cancer patients such as complex chemical prophylaxis, chemical prophylaxis with immunomodulators.

In addition, the authors analyze the methods and individual resuscitation techniques which are not considered as the components of the system, but as specific factors influencing the effectiveness of the patient's treatment. Thus, Aksenova et al. (2007) draw the conclusions about the significance of the specific algorithms that are analyzed as a sequence of actions including the modern informative diagnostic methods in vivo and ex vivo. At the same time, specific medical techniques have been adequately analyzed as a set of medical instruments and actions. In the Russian state program, Healthcare Development technologies are mentioned only in the context of medical equipment necessary for treatment, or in the context of the development and implementation of innovative diagnostic methods, prevention and treatment.

Currently, the molecular genetic research techniques used in forensic activities is the most developed and justified. The Ministry of Health of Russia in its methodical guidelines presented in 1999, develop the components of the structure of the technology for conducting a forensic examination of personal identification and kinship determination, namely the local act and the relevant rules that allow conducting the appropriate actions or procedures. It also contains the specified subject of forensic molecular genetic expert studies. The requirements for places for molecular genetic studies are featured. The same document describes the precautions for the working process

biosecurity. In fact, there is a prohibition of particular actions. Instructions are developed for the implementation of the particular method, namely, the enzymatic amplification of DNA molecules. At the same time, there are no Russian researches, where the technology is regarded as a subsystem of the healthcare management and the combination of the specific elements.

The decision on the need to develop and apply a particular technology should be accompanied by a comparative assessment of the risks and benefits that direct participants (the scientific community, doctors, patients and their families) and indirect participants (society and the state in general) can take. The term stack holders, which is widely spread in international practice, can be used for the participants' identification.

The results of the theoretical works and regulations analysis provided the opportunity for theoretical justification of the following technologies: newborn screening and the consulting contributed by the general pediatricians in order to identify or exclude the severe combined immunodeficiency. Newborn screening has been carried out in different countries for almost half a century, it confirmed the effectiveness in preserving the population's health, in many institutions a scientific and production base has been created and there are distinct diagnostic programs (Khorrami et al, 2015).

According to Tuzankina et al. (2018), these criteria are fully consistent with one of the most dangerous forms of primary

immunodeficiency, such as the severe combined immune deficiency (SCID). That is a group of genetically determined syndromes based on molecular defects that cause the cascade of the immune reactions violations, proliferation processes, differentiation and immune competent cells functions. This researcher in cooperation with the research team of the Russian scientists, studying the various aspects of newborn screening for the severe combined immune deficiency for a long period, proves the possibility of considering it as a technology.

But, despite the scientific and practical achievements, the lack of legal regulation for public health institutions and doctors to diagnose severe combined immune deficiency naturally leads to the fact that regional health programs do not have financing for the implementation of such a technology. Most parents cannot afford such a diagnostics. The next technology is consulting based on the diagnostics of severe combined immunodeficiency disorder. For the evaluation of the technology, the author conducted an expert survey of the managers and specialists of genetic laboratories in Yekaterinburg (Russia).

We should note that the current federal law On State Genomic Registration in the Russian Federation is severely limited by a purely criminological purpose: to identify a person (an unidentified criminal or unidentified corpse), which requires a minimum amount of received and stored genetic (genomic) information, and also a limited circle of subjects entitled to its legal use (courts, preliminary investigation bodies, inquiry bodies and bodies conducting operational-search activity). Accordingly, this law is not able to solve the problems of the

current legal regulation of gene diagnostics in the Russian Federation. There is an understanding in Russian society as to the necessity to change the situation connected to the collection, processing, use and storage of genomic information and samples obtained from the gene diagnostics.

The authors analyze the experience in the legal regulation of the gene diagnostics in the United States of America (USA), Germany (Germany) and the Republic of Latvia. In particular, in the federal United States, the issues of legal regulation of gene diagnostics are solved at the level of the subjects of the federation (states), which provides various approaches to the legal regulation of this issue. Germany, like the United States, is a federal state, but with the Roman-German legal system similar to the Russian one, where gene diagnostics issues are regulated by a single comprehensive. In the Republic of Latvia there is a general approach to the legal regulation of gene diagnostics, which indicates an active reception of the experience, providing the development of a single comprehensive law on this issue.

At the same time, there are various approaches to different types of gene diagnostics, allowed or prohibited ones, in the legislation of the above-mentioned foreign countries. In Germany, secret genetic tests for the paternity determination and for early determination of a child's gender are prohibited; in addition, it is prohibited to examine fetus for the identification of the diseases that may appear in adulthood. Besides, the molecular genetic studies on the identification of diseases that cannot be prevented or cured in accordance with the currently available scientific and technical knowledge are prohibited.

US law (depending on the state) provides specific requirements for actions with the information obtained from the molecular genetic research. It is determined that such information is the confidential and exclusive property of the inspected person, thus, the information cannot be revealed without the consent of the person who was tested. But some states have exceptions, for example, in Maine, Missouri, Indiana, Minnesota, and Michigan there is an obligatory coding of genetic information (Marbán & Mulenga, 2019).

In the legislation of foreign countries, considerable attention is paid to the actions with genetic samples remaining after the gene diagnostics and obtained genetic information. In Germany, this kind of information is stored for ten years and is destroyed, if there is no consent to the other. If the person requires, the results can be destroyed immediately. The genetic sample can be used only for the purposes for which it was obtained and must be destroyed immediately if the goal has been achieved. In the United States, there is almost a complete solidarity of approaches on this issue; the only exception is the question of the possible anonymous use of samples for scientific purposes.

5. CONCLUSIONS

In the Russian Federation, there is a significant gap in the legal regulation of both gene diagnostics and its varieties, including neonatal screening, which has no legal definition. Gene diagnostics on the territory of the Russian Federation requires an urgent legislative regulation, taking into account the permitted and prohibited types, ensuring the

confidentiality of the information received and the subsequent actions, including those with remaining genetic samples. At the same time, one of the conditions for achieving the regulatory impact is the implementation of the systemic approach: namely, regulation of the analyzed issues not discretely (with separate regulatory legal acts of a sub-legal nature), but with a federal law on gene diagnostics, which could be called a federal law On the human genome study, taking into account the legislative experience of Germany and Latvia.

According to the authors of the article, it is advisable to include in such a law some prohibitions, similar to prohibitions in the analyzed countries. For example, a prohibition on the genetic tests of early determination of a child's gender, that can contribute to the gender imbalances among the Russian population. It is also advisable to prohibit an unborn child genetic test for the identification of the diseases that may appear in adulthood. In addition, it seems fair to prohibit the genetic tests of a deceased person, if this is contrary to his will, expressed during his life. The necessity of these regulations implementation is caused by the correlation of the legal norms with ethical ones. Such unity is a remarkable criterion of the expected legal norm, which is characterized by expediency, humanity, compliance with both national interests and the interests of the citizens, and as a result, the absence of the conflict between the public good and the restriction of the personal freedom.

Analyzing critically the foreign experience, it is necessary to note the necessity of legal regulation of the obligatory coding of any genetic information, as well as its storage in a single national database that is created and served only with a specified federal institution. In addition, it

seems obligatory in the Russian Federation to regulate legally the procedure of handling the genetic samples remaining after the gene diagnostics and the genetic information obtained.

It is also advisable to develop the regulations that the genetic information should be stored for ten years and destroyed afterward, but at the request of the subject or his legal representatives could be destroyed immediately. A genetic sample can only be used for the purposes for which it was obtained and should be destroyed immediately as soon as the goal was achieved, except if it is used for anonymous scientific research and if there is the permission from the subject or his legal representatives. The introduction of the necessary law and the development of the corresponding legal regulations will create the necessary conditions for such technologies as newborn screening and early diagnostics of severe combined immune deficiency in newborn children, as well as their financing.

REFERENCES

AKSENOVA, V., LEVI, D., & KLEVNO, N. 2007**. Child and Adolescent tuberculosis**. Geotar-Media. Russia.

BARANOV, A., ALBITSKY, V., & IVANOVA, A. 2012. **Morbidity trends and health status of the child population of the Russian Federation**. Russian Journal of Pediatrics. N^{o} 6. pp. 4-9. Russia.

IZHEVSKAYA, V. 2006. **Russian medical genetics' ethical settings liberalization.** Bioethics Workbooks. Bioethical problems of genomics and ethnogenetics (collection of scientific articles). pp. 33-42. USA.

MAHLAOUI, N., GATHMANN, B., & KINDLE, G. 2014. **The European Society for Immunodeficiencies (ESID) Registry:**

recent advancements in the epidemiology of Primary Immunodeficiencies and how does that translate in clinical care. Rare diseases and orphan drugs. Vol. 1. N° 4: 25-27. Netherlands.

PECHERSKIH, M. 2009. Mana**ging the process of medical care providing to children with malignant tumors**. Doctor Thesis in Medical Sciences. Voronezh. Russia.

POZDNYAKOVA, A. 2011. **Child and Adolescent tuberculosis: features of the disease, new diagnostic technologies and prevention peculiarities**. Doctor Thesis in Medical Sciences. Moscow. Russia.

ROMANOVSKIY, G. 2016. **Legal regulation of genetic research in Russia and abroad**. Lex russica. N° 7. pp. 93 - 102. Russia.

TUZANKINA, I., DERYABINA S., & BOLKOV, M. 2018. **Primary Immunodeficiency at an Early Age**. Moscow. Russia.

VANZELM, M., VANDERBURG, A., & LANGERAK, J. 2011. **PID comes full circle: application of V (D) J recombination excision circles in research, diagnostics and newborn screening of primary immunodeficiency disorders**. Front Immunol. Vol. 2. P. 12. Switzerland.

WERTZ, C., & FLETCHER, H. 2012. **Ethics and Human Genetics: Cross-Cultural Perspective**. Springer Science & Business Media. Germany.

ZAKHAROVA, E. 2006. **Mass screening programs: technical, social and ethic issues**. Medical genetics. N° 3. pp. 21-23. Netherlands.

Marbán, J. M., & Mulenga, E. M. (2019). **Pre-service Primary Teachers' Teaching Styles and Attitudes towards the Use of Technology in Mathematics Classrooms**. International Electronic Journal of Mathematics Education, 14(2), 253-263. https://doi.org/10.29333/iejme/5649.

Khorrami, F. T., Fallah, M. H., & Abadi, H. Z. M. (2015). **The Effect of Unconscious Influences of Satellite Channels on Attitude of Using Satellite**. UCT Journal of Social Sciences and Humanities Research, 3(1), 61-67.

UNIVERSIDAD DEL ZULIA

opción

Revista de Ciencias Humanas y Sociales
Año 35, Especial N° 21, (2019)

Esta revista fue editada en formato digital por el personal de la Oficina de Publicaciones Científicas de la Facultad Experimental de Ciencias, Universidad del Zulia.
Maracaibo - Venezuela

www.luz.edu.ve

www.serbi.luz.edu.ve

produccioncientifica.luz.edu.ve