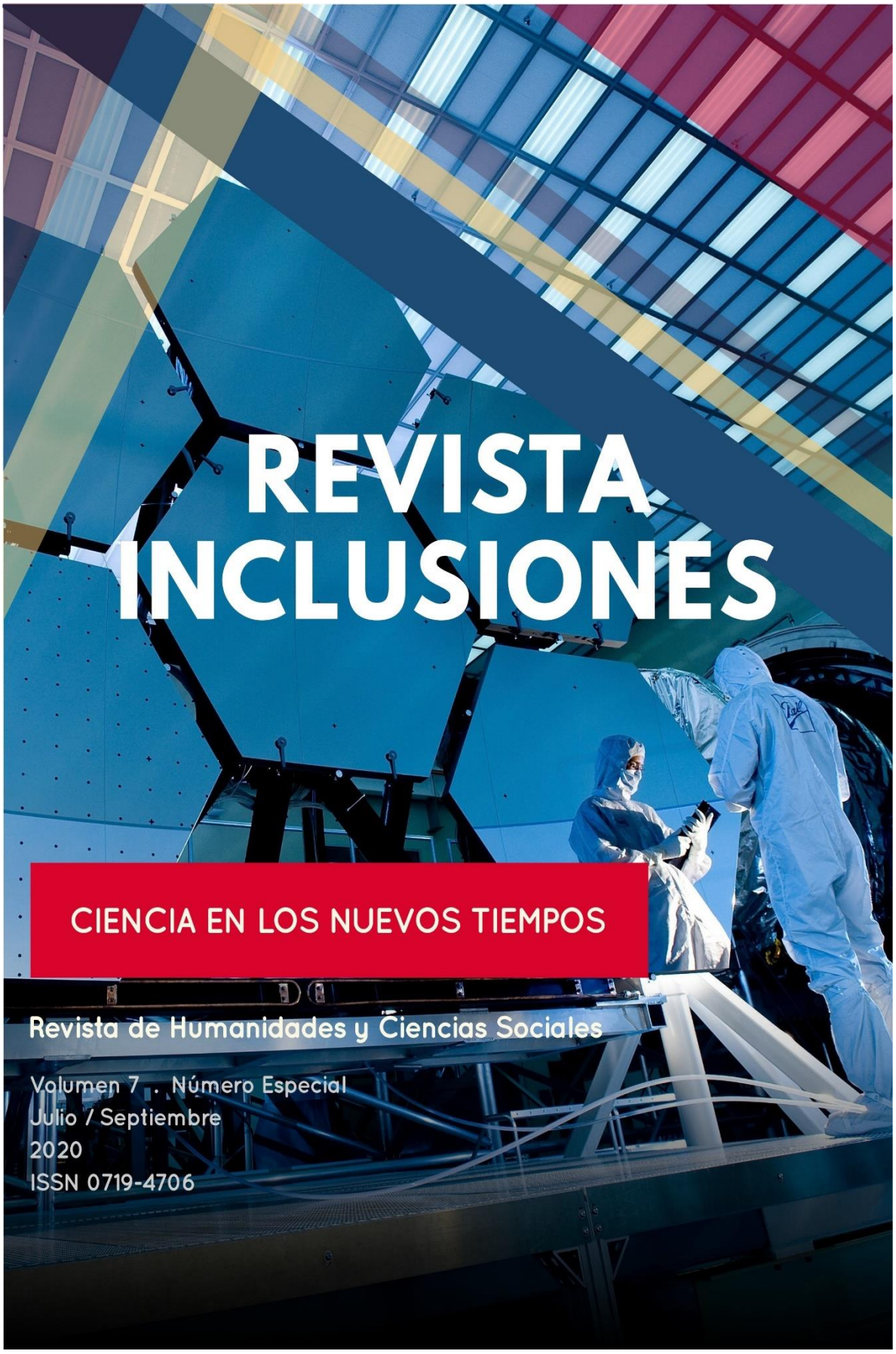

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PRINCIPLES AND TENDENCIES IN PERFORMANCE OF PRENATAL DIAGNOSTICS: INTERNATIONAL EXPERIENCE AND POSSIBILITIES OF ITS APPLICATION

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Abstract

The relevance of this topic is based on the research on fundamental legal and medical postulates and the study of the sequence of trends in the regulation of prenatal diagnostics in systems of justice. This study is determined by the need for the application of this technology and its subsequent implementation in the Russian legal regulation. The purpose of this analysis is the projection of foreign legal maxima of this mechanism through the prism of national characteristics and, as a result, their adaptation to Russian realities. To achieve this goal, the authors have formulated the research objectives to identify legal principles and patterns of their occurrence, formation, development, and functioning in prenatal diagnostics of other states.

Keywords

Principles – Patterns – System of justice – Prenatal diagnostics – Genetic screening

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Introduction

The fundamental task of any state represented by the relevant authorities and officials should be to ensure the birth of a healthy population and the prevention of genetic diseases in the early stages of pregnancy. It considers both a more multifaceted differentiation of the principles of legal regulation into international and national ones, as well as a classification characterized by the specifics of a particular sphere of legal regulation.

Some scholars have positioned their point of view regarding the effect of international legal maxima on such an uncertain in its legal nature quasi-subject as an embryo in a somewhat peculiar manner, reserving the prerogative to give a non-national character to this legal effect. This paradigm is based on its dichotomy, which consists of the unconditional quintessence of the interaction between international acts: The Universal Declaration of Human Rights (adopted by the UN General Assembly on December 10, 1948), the fundamental principles of bioethics, and the moral component. The convergence of the above phenomena either guarantees a positive and healthy intrauterine development of the fetus or, on the contrary, this can lead to a formation of a different mode of behavior, aimed at aborting the fetus in case of danger for the mother, as an objective component of considering her natural right to life and health. In this regard, various disputes arise regarding the admissibility of the use or non-use of prenatal diagnostic measures.

Based on statistical data, it can be stated that over the past 20 years, a significant breakthrough has been made in the field of genetic screening, expressed in the identification of more than 95% of affected fetuses by a false-positive result of less than 3%[1].

While the fact of revealing a genetic predisposition to certain diseases is a positive thing, at the same time, one should not forget about the inherent human subjectivity and lobbying for the idea of forming a personality constitution in the form of predetermining the sex of an unborn child or its social and psychological characteristics. For example, the University Hospital Erlangen in Germany practices scientific research in the field of human reproduction and its possible disorders at the stage of embryonic development. This resulted in the birth of the first child with the help of assisted reproduction in 1982.

In particular, Austria and Switzerland have a regulatory array that is advisory in the field of genetic diagnostics and treatment of repeated spontaneous abortions. The main trend being promoted in these systems of justice is the analysis of chromosomes in both parents after repeated abortions. At the same time, these countries adhere to the direction expressed in the counter-effect of screening to detect such a defect as thrombophilia. Further, in these states, scientists developed a method for karyotyping women who have had two or more miscarriages, which allowed analyzing nucleoprotein structures for the detection of chromosomal aberrations. In this regard, geneticists note an increase in the probability of optimal fetal development and, as a result, the possibility of having healthy children. Subsequently, this concept (the implementation of the microscopic chromosome analysis) was supported and accepted as a basic standard by other states.

[1] KO. Kagan; J. Sonek; P. Wagner y M. Hoopmann, “Printsipy skrininga pervogo trimestra v vozraste neinvazivnoi prenatalnoi diagnostiki: skrining khromosomnykh narushenii”, Archives Gynecology Obstetrics. num 296 (2017): 645-651.

At the same time, one needs to focus on the moral and ethical side of the issue. Researchers conducting stem cell research occupy a position on the reflection of the main factor of ethical problems, which is expressed in the safety aspect of this procedure. It is interesting that in 2002, the British Council on Bioethics illustrated recommendations that reproduced the idea of investigating the genetic structure of criminal defendants before a court ruling, as well as determining their genetic predisposition to criminal behavior[2].

One's attitude to bioethical values manifests itself differently in this state at a particular period[3]. It seems optimal to ensure a harmonious combination of moral and regulatory principles during the genetic screening procedure. Thus, we find the experience of Switzerland and Western Australia interesting, with the Academy of Medical Sciences developing the proposed guidelines. As a result of the situation, expressed in an independent assessment performed by patients of their circumstances and possible consequences manifested in large-scale refusals of intensive treatment for serious illnesses, local medical societies developed wish forms for patients, and visions for patients regarding possible treatment and further correction of such events from the medical staff.

Given the foregoing, the aim of studying this problem is to construct national as fundamental principles of legal regulation in prenatal diagnostics, as well as local principles, unique to this procedural tool, determining the specifics of this area. In this regard, an additional objective was to study the legal and ethical array based on the foreign experience of various systems of justice and for the most part demonstrating an effective and productive result in the field of genetic screening.

Methods

Using the historical-legal method, we analyzed the occurrence, formation, and development of the phenomenon of prenatal diagnostics in the context of the permanent transformation of the political, economic, and spiritual sphere of life in a society of a particular system of justice. The main part of our study is based on the use of comparative-legal means, due to the diversity and inconsistency of doctrinal research, judicial practice and the legal framework both within a particular state and on the global scale as a whole. Using formal legal tools, we ascertained an indifferent attitude of Russian legislators to this issue, expressed in the transcendental and amorphous legal regulation of genetic screening in Russia.

When writing the article, we analyzed more than 20 normative legal acts in the field of genetics and bioethics and some documents of a recommendatory nature. We also analyzed the legal and medical doctrine reflecting the views of medical scientists and lawyers regarding the ethical side of the issue, the patterns of genetic screening conditions, the presence of limitations in prenatal diagnostics, as well as the fundamental guidelines functioning in this area which should serve as a guide for medical personnel during the application of such procedures.

[2] V. Tarantul, The human genome: a four-letter encyclopedia (Moscow: Languages of Slavic culture, 2003), 721.

[3] M. Sleeboom-Faulkner, Sotsiologicheskie perspektivy v bioetike: prognoznoe geneticheskoe testirovanie (PGT) v Azii", Bioethical Inquiry. num 4 (2007): 197-206.

Results

The study of foreign experience in the range of basic settings within the framework of the prenatal diagnostics procedure helped to identify several principles and patterns. It should be emphasized that the screening procedure is not aimed at leveling inherited pathologies in general. The main goal is to prevent the reproduction of the fetuses with serious diseases that are incurable or difficult to treat.

After analyzing the points of view of German geneticists, we can identify three key areas that should be considered when forming approaches to developing legal maximums in genetic screening:

1) *political correctness*, which appears in the reproduction of children and genetics, which has a predominant effect on the relevant state bodies and officials of a particular state, as a result of which, for example, Germany had adopted the Law on the Protection of Embryos;

2) *protective ethics*. Lobbying ethical principles inhibit development and intentionally ambiguously evaluates genetic and reproductive tools to promote public health policies that focus on budget savings;

3) *service ethics.* Willingness to provide risk in the application of new technologies to facilitate the introduction of innovations in the healthcare sector, ignoring the aspect associated with certain costs[4].

In this regard, we can conclude that the *principle of confidentiality* has an ambiguous position when considering this phenomenon. Prenatal screening protocols can be applied on a large scale to identify women at risk of having a baby with a common genetic disease to counter the spread of genetic disorders[5].

Protecting the confidentiality of medical information is the most important legal basis, which is designed to guarantee respect for the boundaries of the patient's intimate sphere and trust in the medical profession. This circumstance was established on February 25, 1997, in a judgment in the case of Z v. Finland[6]. The applicant's medical information had not been properly protected, which resulted in unauthorized access to her personal information, and thus, in neglect and violation of both the norms of Finnish national legislation and the norms of the international act, art. 8 of the Convention of Human Rights and Fundamental Freedoms of November 4, 1950.

Besides, there are many misconceptions regarding the principles of screening: this phenomenon is sometimes erroneously confused with prenatal diagnostics and, as a result, there is a substitution of legal constructions at the stage of application of certain procedures.

[4] H. M. Sass, Bioethics in the German-speaking countries of Western Europe: Austria, Germany, and Switzerland. In: Lustig B. A., Brody B. A., Engelhardt H. T., McCullough L. B. (ed.). Textbook of bioethics Vol: 2 (Dordrecht: Springer, 1992).

[5] P. Gambhir, “Basic principles of prenatal aneuploidy screening”, Molecular Cytogenetics. num 7 (2013): 115.

[6] Z v. Finland: Reports of Judgments and Decisions 1997-I. February 25, 1997. Retrieved from: http://europeancourt.ru/resheniya-evropejskogo-suda-na-russkom-yazyke/z-protiv-finlyandii-postanovlenie-evropejskogo-suda/

Thus, it should be noted that there is a large-scale and diverse classification of these legal maxima. It seems that the most optimal and transparent is the differentiation of principles on:

1) principles regulated in the norms of international legal acts;

2) principles regulated by the rule of law of a particular state (general and specific).

The general legal principles include the principles of legality, equality, and equal rights, justice, respect for the honor and dignity of the individual, humanism. For example, the fundamental principle of law, which is non-national, represents the *principle of legality*. The economic situation has a significant impact on this basic setting, resulting in stagnation in the medical field, expressed in the conservation of various innovations in the field of genetic screening. Some experts have interpreted the term as "a state of security, stability, and public health of the country"[7]. This interpretation is based on the ideological component, which serves to ensure the achievement of the proper result. Alternatively, the provision of paragraph 84 of the European Convention for the Protection of Human Rights and Dignity of the Person in Connection with the Application of Biology and Medicine, is aimed at preventing genetic testing in any other interests besides protecting the health of the person.

Some of the above principles are at the crossroads of special principles that absorb the specifics of this field of activity, namely the implementation of the genetic screening procedure.

First of all, it is necessary to focus on such a principle as the *professionalism and competence of the medical personnel.* Alternatively, if a balanced chromosomal pathology is found by the patient, it should be remembered that although the genetic material is completely normal in the number of copies and functionally intact, meiotic disturbances in the distribution of chromosomes are possible at the same time, which increases the risk factor for miscarriage or birth of a child with a hereditary defect. The diagnosis of this fact radically changes the treatment strategy while continuing further pregnancy. Statistical indicators demonstrate an increased level of aberrations: 40-60% of early embryos are abnormal in chromosomes[8]. Given this, it seems that to avoid negative consequences, the geneticists need to be qualified enough to identify and diagnose genetic defects at an early stage. The *principle of efficiency and timeliness of identifying a hereditary disease* shall be considered logical and consistent. The technique of prenatal diagnostics consists of sampling chorionic villi or amniocentesis in a patient. The former procedure has a significant advantage, due to its implementation in the early stages, occurring at 11-12 weeks of gestation, rather than such an invasive method as amniocentesis, which is practiced only at 16-18 weeks of gestation[9].

[7] V. I. Shind, Some questions of management in Prosecutor's offices (Methodological aspect). Problems of the theory of legality, methodology, and methods of Prosecutor's supervision: A collection of scientific papers (Moscow, 1994).

[8] T. Hassold y P. Hunt, “To err (meiotically) is human: the genesis of human aneuploidy”, Nature Reviews Genetics. num 2 (2001): 280-291.

[9] C. Kähler; U. Gembruch; K-S. Heiling; W. Henrich y T. Schramm, “DEGUM Recommendations for amniocentesis and chorionic villus selection”, Journal of Ultrasound in Medicine. num 34 (2013): 435-440.

Further, it is appropriate to disclose the *principle of informing the client about the goals and results of the medical examination.* It is indisputable that any genetic diagnostics should be preceded by patient awareness of this procedure, which is factual according to the legislation and internal instructions of a particular institution. At the same time, the doctor should obtain the patient's signature on familiarization with the above documents, as well as their consent to prenatal diagnostics.

The *principle of the configuration of ethical and moral factors*, which manifests itself at the time of exposure of hereditary defects or the presence of medical indications for abortion, is also very important. Thus, the German law prohibits abortion starting from the end of the 14th week of gestation, the Swiss law allows it until the end of the 12th week of gestation, and in Austria, this issue is determined by ideas of the doctrine based on the maximal possible time (until the end of the 16th week of gestation). The ethical side of the issue is related not only to the philosophical aspect but also to the problem of moral choice[10].

As an option, the content of the moral component has been disregarded in the PRC law and order, where the consideration of family values dominates international norms that prohibit any invasion of the genetic structure of the embryo[11]. Besides, in 2014 in Germany, the PID control came into force, which allows this procedure to be carried out if the carrier's genetic disease can be transmitted to the unborn child. It is advisable to add that the PID procedure is authorized only in specialized centers, by a positive vote by the ethics committee.

Speaking about the proper implementation of genetic research, a discussion of financing issues should be raised. In this connection, the administration of budgetary funds concerning Russia is a cornerstone in the system of the prenatal diagnostics mechanism. This state of affairs allows us to talk about the *principle of necessary funding*.

The provision of paragraph 72 of the European Convention on Human Rights and Biomedicine (1996) reflects the consolidation of structural elements of genetic research, including medical examination by analyzing the patient's genetic heritage. It seems appropriate and justified to introduce a license to carry out this type of procedures, as illustrated, for example, in the experience of the United Kingdom, which has a special institution (Human Fertilization and Embryology Authority, HFEA) working on issues of issuing permits to specific medical organizations for genetic screening.

The best idea is to create committees that would have the opportunity to work out in a multidimensional plan the possibility of any biomedical research. Thus, a research ethics system is successfully functioning in the Scandinavian countries (Denmark, Norway, and Sweden)[12]. Accordingly, it seems necessary to consolidate the *principle of compulsory licensing* in the design of special legislative acts.

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[10] J. C. Fletcher, Moral Problems and Ethical Guidance in Prenatal Diagnosis. In: Milunsky A. (eds) Genetic Disorders and the Fetus (Boston, MA: Springer, 1986).

[11] J. J. G. Gietel-Habets; C. E. M. Die-Smulders; V. C. G. Tjan-Heijnen; I. A. P. Derks-Smeets; R. Golde y E. Gomez-Garcia, "Professionals' knowledge, attitude and referral behaviour of preimplantation genetic diagnosis for hereditary breast and ovarian cancer", Reproductive Bio Medicine Online. Vol: 36 num 2 (2018): 137-144.

[12] R. K. Lie y J. E. Paulsen, Bioethics in Scandinavia: 1989–1991. In: Lustig B.A., Brody B.A., Engelhardt H.T., Mccullough L.B. (eds) Bioethics Yearbook. Bioethics Yearbook, vol 2. (Dordrecht: Springer, 1992), 280.

In the context of discussing the dilemma of prenatal research, one should pay attention to human subjectivity and the possibility of errors on the part of medical staff. To correct medical errors, based on the presumption of guilt, the principle of *full and affordable compensation* should be implemented.

Laying the foundation in the mechanism for the implementation of legal maxima, the need for the *principle of providing state guarantees to ensure the rights of a person who has consented to undergo prenatal diagnostics and mandatory monitoring of their compliance* should be emphasized. It seems that this legal framework is an effective tool for the successful and enhanced functioning of each basis individually, as well as for ensuring the interaction of all principles as a whole.

Discussion

The correlation of the facts of a wide range of hereditary diseases, both already known in genetic engineering and the ones that are still at the research stage, as well as their constant modification and modernization, allow us to ascertain the severity of proper monitoring and supervision of the patient's treatment to apply procedures for examining the health status of both the subject and unborn baby[13]. A study of statistical data shows that in 30-50% of individuals and their families, classic hereditary defects such as monogenic diseases or chromosomal aberrations are identified[13].

First of all, it is necessary to ensure the rational functioning of the relevant committees in the field of healthcare represented by qualified personnel that will implement recommendations in the field of prenatal diagnostics, and, as a result, be responsible for interim control. It also seems necessary to us to establish a clear list of criteria for performing genetic screening. An exhaustive list of grounds for admission to the genetic screening procedure should be developed to implement the industry *principle of the inadmissibility of abuse of the right by officials.* It must be remembered that the relevance of the situation lies in the essence of prenatal diagnostics: namely, in the complex of medical measures by which hereditary diseases are identified at the molecular level at the stage of fetal development[14].

We find it unfortunate that there is no model law serving as a guideline for different states to adopt an intra-national act by incorporating legal norms that provide legal regulation of prenatal diagnostics. At the same time, at the international level, during a WHO consultative conference in 1997 on the topic "Ethical Research in Medical Genetics", a discussion was held on the ethical principles of the prenatal genetic service[15]. From the analysis of some principles voiced at this conference, one should refer to the debatable nature of such a legal attitude as the *principle of notification of diagnostics to the patient's family* and, as a result, leaving the final decision to the family. As it seems, no other person, except a professional geneticist, has the opportunity to qualify properly the diagnosis and

[13] M. S. Kondratev y E. V. Zakharova, "Virtual screening of thiol reducing agents for peroxiredoxin 6", Biophysics. Vol: 63 num 5 (2018): 844-849.

[14] V. I. Saburova, Ethical problems of prenatal diagnostics. Church and public Council on biomedical ethics. December 22, 2009. Retrieved from: http://bioethics.orthodoxy.ru/analitika/zashchita-zhi. DOI: 20.03.2020

[15] E. V. Perevozchikova y E. A. Pankratova, "The Constitutional right to life and the legal status of the human embryo", Medical Law. num 2 (2006): 33-34.

the measures prescribed under it, a priori, not having the necessary sufficient knowledge and skills to operate and interpret the medical conceptual and categorical apparatus.

We would also like to demonstrate another gap on which one needs to focus. Thus, the norm of paragraph "b" of Art. 8 of the 2003 International Declaration on Human Genetic Data, a situation has been resolved in which the patient cannot provide informed consent to permit genetic testing. In this case, the patient's legal representative is vested with the right to make a decision. In the absence of conventional regulation regarding the case of making specific requirements for the status of such a person, fixing legal maxima at the international level in the field of genetic screening, we would like to illustrate one of the possible conflicts. Under the Federal Law "On the Basics of Protecting the Health of Citizens in the Russian Federation", minors who have reached the age of fifteen years have the right to give informed consent to medical intervention or to refuse it. It seems that a minor subject does not have sufficient resources to make an adequate, constructive and objective decision to provide consent to abortion or continuation of pregnancy if a hereditary disease is detected at the stage of embryonic development. At the same time, one cannot agree with the fact that the legal representatives of such a person are informed about the potential threat to the life or health of both the pregnant woman and the child, since this conflicts with the provisions of Clause 9, Article. 4, Art. 13, paragraph 2, part 2 of Article 73 of the aforementioned law, which reflects the implementation of the fundamental principle of health protection, namely *respect for medical confidentiality and confidentiality of personal data*.

The dualism of the implementation of this provision is also manifested in the regulation of the norm-obligation, which consists in the obligation of the medical worker to disclose information to the relatives of the pregnant woman who are at risk of being BRAC gene carriers to prevent serious harm[16]. Given that medical counseling is becoming more and more relevant and in demand, due to the risk of relapse of genetic diseases, it is necessary to continue research in this area not only from a doctrinal point of view but also due to the manifestation of legislative interest in this area of public relations, to ensure relief and harmonious legal regulation of prenatal diagnostics in Russia.

Now, the key regulatory legal act in Russia is Order No. 457 of the Russian Ministry of Health "On improving prenatal diagnostics in the prevention of hereditary and congenital diseases in children" dated December 28, 2000, along with relevant instructions. This order acts as a rudimentary document incapable of regulating this spectrum of relations, which have been changing and modernizing over two decades; therefore, the above norms should be reconstructed.

We would like to note the following: in Russia as of 2017, hereditary aberrations accounted for 6.8 thousand people with mutations, which is about 320 people with metabolic disorders per year[17], even though the functions of preventing and leveling such adverse effects should be provided by instrumental norms.

[16] YAK. Skittles, A new bioethics framework to facilitate more effective decision-making about genetic information. In: Boylan M. (ed.) public health Policy and ethics. International library of ethics, law and new medicine (Dordrecht: Springer. 2004).

[17] E. Gubernatorov y E. Kostina, Region with a high level of genetic disease was detected in Russia. November 24, 2017. Official website: RBC. Available at: https://www.rbc.ru/rbcfreenews/5a181d139a79471217133d36 DOI: 20.03.2020.

For example, the legislation of the Anglo-Saxon legal family based on case law, in particular the United States, demonstrates the general legal message of regulating genetic screening at the federal level, leaving the right for each state to regulate prenatal diagnostics, a key feature of which should not be an *antagonistic* contradiction to bioethical teachings[18]. It is noteworthy that only in 2007 the first recommendations on large-scale non-invasive prenatal testing were developed in the USA, as a result of which this technology was cultivated and, as a result, the invasive method of genetic screening was abandoned[19].

It seems necessary to indicate that in most European countries, such as Belgium, Greece, Ireland, Spain, Italy, Slovakia, Ukraine, the Czech Republic, and Russia, the prenatal diagnostic procedure has not received proper legal regulation in a multidimensional plan, unlike, for example, France, where the prenatal diagnostics technology is determined by an independent special law "On the donation and use of elements of the human body, assisted reproductive technologies and prenatal diagnostics", which reflects fundamental legal maxima that guide geneticists in implementing these procedures.

Conclusion

The study identified the following constitutive general and specific principles of prenatal diagnostics: confidentiality; legality; professionalism and competence of medical staff; efficiency and timeliness of identifying hereditary diseases; informing the patient about the goals and results of the medical examination; configurations of ethical and moral factors; necessary funding; compulsory licensing; full and affordable compensation; providing state guarantees to ensure the rights of the person who had given their consent to undergo prenatal diagnostics and mandatory supervision of their observance; inadmissibility of abuse of the right by officials; diagnostic notifications for the patient's family; medical confidentiality and confidentiality of personal data.

A prenatal diagnostic is a productive tool that allows minimizing hereditary aberrations, using a variety of tools and technologies that allow timely identification of the possible presence of deviations at the stage of embryonic development and predicting a potential favorable or unfavorable outcome of events. At the same time, realizing the objective and technical side of the issue, we should not forget about deontological ethical tools, despite the ambiguity and binary approaches regarding the onset of the doctor's responsibility for the diagnosis, surgical intervention, indications for termination of pregnancy or the onset of other consequences for the person who undergoes the genetic screening.

An analysis of scientific research in the field of prenatal diagnostics has led to the conclusion that it is necessary to adopt a special federal law in Russia that provides stable and effective legal regulation of genetic screening.

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