

To the bank for DNA

What opportunities does genetics give us, and how can we manage information about ourselves?

Today, a huge number of medical and scientific organizations around the world are engaged in genetic research. All together they generate a huge amount of data, which must somehow be systematized and analyzed. This information is accumulated in banks of genetic information, access to which is regulated by the researchers themselves. And then a whole series of problems of a legal and ethical nature arise, for which there are no unambiguous solutions yet.

It is true that the regulation of this process should be based on the opinion and preferences of people who, in fact, are carriers of this information. Meanwhile, as the first results of a large-scale international research project "Your genes are for you to solve," only slightly more than half of people (54%) have any idea about DNA, genetics and genomics. However, only one in two is willing to share their genetic information for scientific purposes.

What are the public risks of DNA testing technology? Do I need to give medical advice to patients before carrying out a genetic test? Are there any legislative norms that regulate this sphere? This is our conversation with the associate professor of the Department of Medical Genetics with the course of prenatal diagnosis of the Russian Medical Academy of Continuing Vocational Education of the Ministry of Health of Russia, PhD Elena BARANOVA.



- Explain, what are the genetic data itself and why is it necessary to collect such information?

- Now we are available and widely distributed genetic testing for diagnosis of hereditary diseases, for the selection of drugs or their dosage, genealogy (when people want to know their "origin"). DNA is excreted and analyzed. As a result, we do not get the DNA itself, but its decoding - the code letter, that is, information about DNA. It is these "letter chains" that get into different data banks. The presence of such bases is very important for doctors and scientists, since only an analysis of a large amount of material can help to reveal some patterns, to obtain information about the purity or pathogenicity of genetic changes, their role in the functioning of the organism.

- What perspectives of these researches do you see in terms of improving approaches to treatment, developing new methods of therapy and drugs?

- Even if we do not take absolutely stunning results in the diagnosis of hereditary diseases, most breakthroughs in medicine are somehow connected with genetics and molecular biology. This primarily drugs for the treatment of cancer, the selection of an individual dose of drugs for cardiovascular diseases, there are genetically engineered drugs that help to avoid amputation with extensive lesions of the vessels of the lower extremities, etc. It is difficult to imagine the field of modern medicine, where genetic technologies would not be used.

Today there is a process of accumulating knowledge about genetic mutations. The bulk of information is contained in foreign data banks, in Russia in this direction the first steps are being taken. Information is constantly updated, due to this, researchers are able to diagnose new rare diseases, to seek approaches to their treatment. That is, little to know the diagnosis, the main thing is to have information about what kind of mutation in the gene there is in the patient and what needs to be done to level out the "gene breakages". This is a huge achievement, because only recently we could only state the disease at the stage of prenatal diagnosis and recommend the patient to terminate the pregnancy. Now in a number of cases there is the possibility of therapy of such conditions.

- How well is the data interpreted adequately? Do they have a share of subjectivity?

- Indeed, at the moment we know quite well the "anatomy of the genome" and much worse its "physiology", that is, what is responsible for this or that change in the genome, is this change by normal variability, or it is pathogenic. Therefore, the availability of databases is so important - precisely because of the need to analyze a large amount of information from many people to assess the pathogenicity of various changes. To date, algorithms have been developed, according to which one can interpret certain data, and with each new scientific publication they are supplemented and refined. This opens up great opportunities in terms of reassessing the results obtained earlier, and adjusting them.

Of course, much, if not all, depends on the qualifications and professionalism of the specialist-bioinformatics. Genetic changes are very "subtle spheres", and there are often situations where it is difficult to interpret whether they are benign or pathogenic. Unfortunately, we still do not know too much about the nature of these changes and are forced to leave these data "for later" so that after a while they can be returned to them and analyzed again, taking into account the information that has appeared.

- In recent years, even a term such as genetic discrimination has appeared. For example, abroad, particularly in the United States, there are precedents, when people who have a genetic predisposition, say, to Alzheimer's disease, insurance companies change the cost of long-term medical insurance, depending on genetic factors.

- Yes, moreover, there were cases when employers found out about the genetic predisposition to oncological diseases, in particular to breast cancer, and took steps to dismiss the employee. In some situations, the employer, without obtaining an informed consent from the employee, tried to find out, on the basis of genetic tests, his involvement in some incidents. Since 2008, the United States has passed a law on non-discrimination in genetic information (GINA), according to which employers and insurance companies can not request a DNA test or medical data about a family from a person. Thus, the law at the federal level prohibits genetic discrimination in health insurance and at work, and does not affect other areas of life. Real court examples show that the law works.

- Today, some research centers offer services to create a so-called genetic passport. You can even make such a "gift" to your relatives or relatives. Is there any practical benefit from this kind of research?

- Let's just say, genetic passports in healthy people have rather limited clinical application. For example, it makes sense to determine some changes in the planning of pregnancy, the appointment of certain drugs. Of course, all this should be done only after preliminary medical and genetic counseling, during which the goals of testing, patients' expectations will be revealed and stipulated what he wants or does not want to know. This is important, because with full-genomic methods, for example, we can accidentally identify a predisposition to a disease with a late onset, in which there is no effective prophylaxis, or to oncological disease. With regard to such a proposal as "to give a genetic test" ... Think about whether you want to make such a gift to your relatives and friends? What do they personally want to know?

- How and for what purposes can this information be used? What are the public risks of DNA testing technology?



- I would like to emphasize at once that in this case we are not talking about some fantastic situations with the development of biological weapons, persecution by the police or the government, but about more real ones. Again, in the genetic databases are not stored the DNA itself, and information about it - the same code, without specifying the personal data of a person. Nevertheless, in the modern world it is quite possible, with the help of social networks and forums, to find information about a person with a rare pathology and to link it with data on the genome. For example, many faced the problem when their phone and the information that the policy is ending was in the hands of insurance companies offering insurance for cars. This leads to a large number of calls with the sale of services. There may be suggestions about drugs and so on. Besides,

- When a person gives his biological materials for the analysis of the genome, he not only learns the results of the research, but also provides information to the company that conducts the testing. De facto, it can provide this information to the state or to someone else who can ask for it and obtain this information ...

- In our country, this sphere is clearly regulated. Thus, in 2006, the Federal Law No. 152-FZ "On Personal Data" was adopted in Russia. There is an article in which it is written that the biometric personal data of a person on the basis of which it is possible to establish his personality can be processed only with the written consent of the person. But there are a number of exceptions. For example, this rule does not apply to terrorists, people who committed serious crimes - recidivists, rapists, etc.

- Your professional position is clear. And how does society treat the problem of collecting genetic information? What results were obtained during the study "Your genes - it's up to you"?

- The main goal of this international project, developed by a group of scientists from the Global Alliance for Genomics and Healthcare (GA4GH) and the Wellcome Genome Campus (Cambridge, UK), is to find out what people in different countries think about this problem. In Russia, the study was initiated by the Russian Society of Medical Genetics, more than 200 people have already taken part in it, it will last until the end of 2017.

So far, preliminary results on Anglo-speaking countries - the USA, Canada, Australia and Great Britain - have been summarized. In total, at the beginning of September 2017, 9742 people took part in the study. As it turned out, 46% of respondents do not know the information about DNA, genetics and genomics, while every second thinks that this is a "special type of medical information". For the overwhelming majority of people (79%), the protection of banking data is of the greatest importance, in second place - medical data and information about DNA; 12% gave an affirmative answer to the humorous offer to maintain confidentiality regarding their "vacation photos"; 45% of respondents are ready to provide their medical data for scientific purposes, if this is not related to their commercial use (only 28% agree). Notably, that they are ready to sacrifice their DNA for research precisely those people who have a certain idea about DNA and consider it a special kind of medical information. In addition, they expressed interest in providing them with the results of the study. And absolutely mirror situation with respect to those who do not own the "theme". These people in most of the negative attitude to this kind of research and do not want to take part in them. The main concerns of the study participants are related to the fact that their DNA can be copied and placed on the crime scene, with discrimination by health insurance companies and obsessive advertising of drugs by pharmaceutical companies. they expressed an interest in providing them with the results of the study. And absolutely mirror situation with respect to those who do not own the "theme". These people in most of the negative attitude to this kind of research and do not want to take part in them. The main concerns of the study participants are related to the fact that their DNA can be copied and placed on the crime scene, with discrimination by health insurance companies and obsessive advertising of drugs by pharmaceutical companies. they expressed an interest in providing them with the results of the study. And absolutely mirror situation with respect to those who do not own the "theme". These people in most of the negative attitude to this kind of research and do not want to take part in them. The main concerns of the study participants are related to the fact that their DNA can be copied and placed on the crime scene, with discrimination by health insurance companies and obsessive advertising of drugs by pharmaceutical companies.

- And in conclusion, what do you think will be the further development of this direction?

- Progress in genetics and genomics is far ahead of other areas of clinical medicine. These changes are so rapid that it is becoming more difficult to control this sphere. It is clear that the absolute anonymity of genetic data is unlikely to be achieved in view of all those problems that we voiced. All participants in this process need to learn to listen and hear each other, to develop common ethical principles by analogy, for example, with those that exist in the field of clinical drug research. This task was also put in the basis of the British study. Its organizers hope that the opinion of the most important part of the process - our patients - will further stimulate the development of international and national documents regulating legal,

The conversation was conducted by

Irina Stepanova,

corr. «MG» .

[<N 92 of December 6, 2017](#)

[Top](#)

 [Login or register](#) to post comments |  [: Science and practice](#)

[Healthcare](#) [Medicine and society](#) [Focus on](#) [clinical recommendations](#) [Pharmacist page](#) [Science and practice](#) [Peace and health](#)
[Science abroad](#) [Time and we](#) [Autograph for "MG"](#)



[About the newspaper](#) | [Subscription](#) | [Archive](#) | [Contact](#)
Us Copyright © 2006