

GENETIC TESTING AND PERSONAL CHOICE OF THE PACIENT

Sevdalina Alekova *, **Veselina Petrova-Tacheva ****, **Borislav Popov ****,
Svetlana Georgieva***, **Neli Dimitrova****, **Katya Dimitrova******

* *Department of General medicine and Ophthalmology, Faculty of Medicine , Trakia University, Stara Zagora, Bulgaria*

** *Department of Molecular Biology, Immunology and Medical Genetics, Faculty of Medicine, Trakia University, Stara Zagora, Bulgaria*

****Department of Genetics, Animal Breeding and Reproduction, Agricultural Faculty Trakia University, Stara Zagora, Bulgaria*

**** *ESTRE- OOD, Sofia, Bulgaria*

GENETIC TESTING AND PERSONAL CHOICE OF THE PACIENT

Sevdalina Alekova *, **Veselina Petrova-Tacheva ****, **Borislav Popov ****,
Svetlana Georgieva***, **Neli Dimitrova****, **Katya Dimitrova******

* *Department of General medicine and Ophthalmology, Faculty of Medicine , Trakia University, Stara Zagora, Bulgaria*

** *Department of Molecular Biology, Immunology and Medical Genetics, Faculty of Medicine, Trakia University, Stara Zagora, Bulgaria*

****Department of Genetics, Animal Breeding and Reproduction, Agricultural Faculty Trakia University, Stara Zagora, Bulgaria*

**** *ESTRE- OOD, Sofia, Bulgaria*

ABSTRACT

Implementing a genetic tests, the perception of the received results and taking further action based on the patient's own choosing, raises a number of moral, ethical and psychological dilemmas. In our study we surveyed 140 men and women of reproductive age in Stara Zagora region about their attitudes for adoption of genetic prevention and related new genetic technologies and the presumed risks. In the current report we present part of the processed and analyzed study results.

Keywords: genetic testing, ethics, individual choice

Introduction:

In their own daily work medical workers meet often with hereditary diseases and predispositions: monogenic diseases, congenital disorders, socially significant multifactorial diseases, etc.. In the clinical practice, due to the hereditary nature of the disease and predispositions, while working with patients, can arise moral and ethical, psychological, social and legal issues and problems. Studies and discussions concerning medical ethics are always important in many aspects of the health care system, but they have particular importance in medical genetic consultation. The actual implementation of genetic testing, the perception by the patient of the result, the questions about the probability of developing a serious hereditary disease, and the probability of transmission of hereditary disease in generation, raises a number of ethical questions for people-carrying the mutant allele.

Goals and purposes:

The submitted by us study has the following goals:

To explore and analyze the personal position of the patient to predictive medicine and the application of genetic testing in the early detection of diseases.

Material and Methods:

It was implemented a sociological study of 140 people from Stara Zagora region between September and December 2012, including method direct individual poll in compliance with the principles of anonymity and voluntary.

It was used a questionnaire with seven point scale. / Likert type /.

When processing a data, there was applied advanced mathematical and statistical methods, which makes it possible to clarify the personal position of the surveyed group on genetic testing.

Discussion and results:

The total number of participants who completed a thorough and responsible poll is 140.

The largest age group is the category- 20-22 years- 92/ 65%/

In the sample are included 44 - /31%/ men and 96 - /69%/ women.

Genetic consultation and how to present information from a test of specific disease hides a social, psychological and ethical risks. On the one hand, the physician must take into account the patient's right to confidentiality and informed consent, but also he must be able to predict the potential for social harm and psychical stress.

78% of the surveyed group declare, that they exercise over a right to be informed about their health./ fig.1/

fig. 1

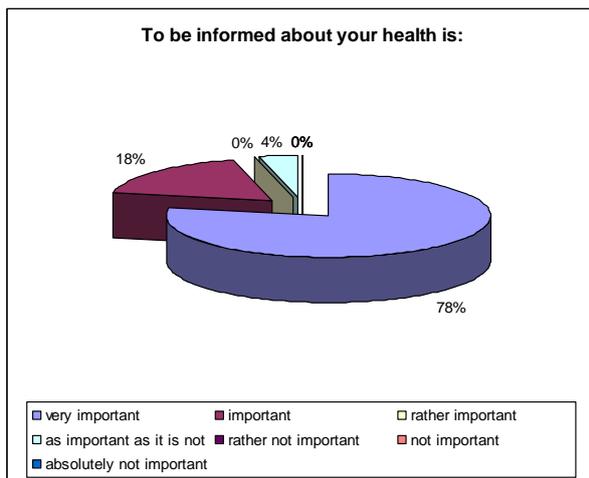
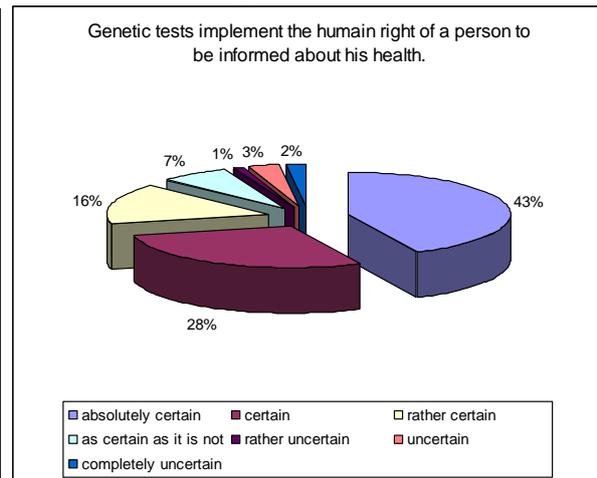


fig. 2



Genetic testing and analysis of results is another safe option for 43% of respondents to receive expert assessment of their health./ fig.2/

The mere submission of a genetic test may expose the patient to a heavy moral choice, to prompt rethink its future / fig.3/

fig. 3

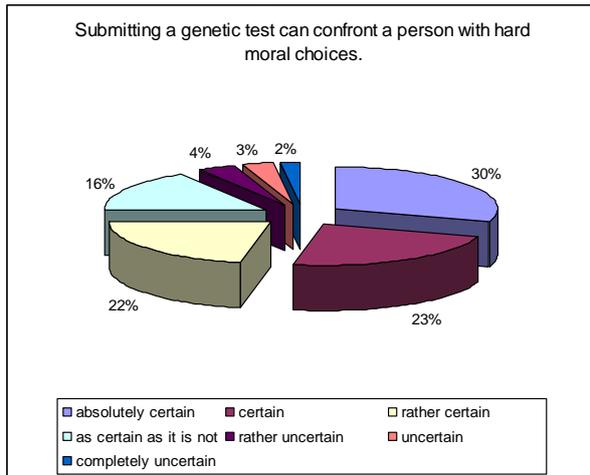
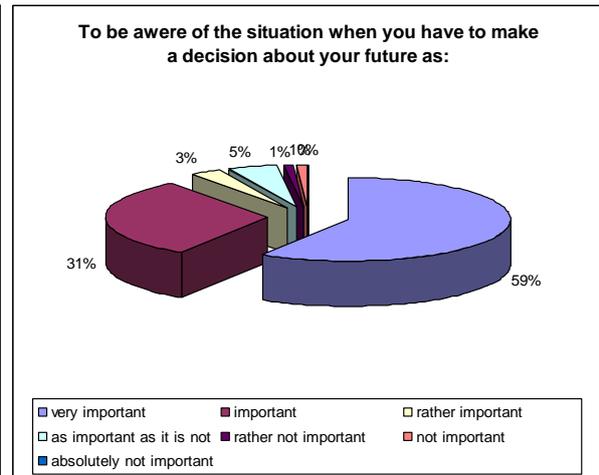


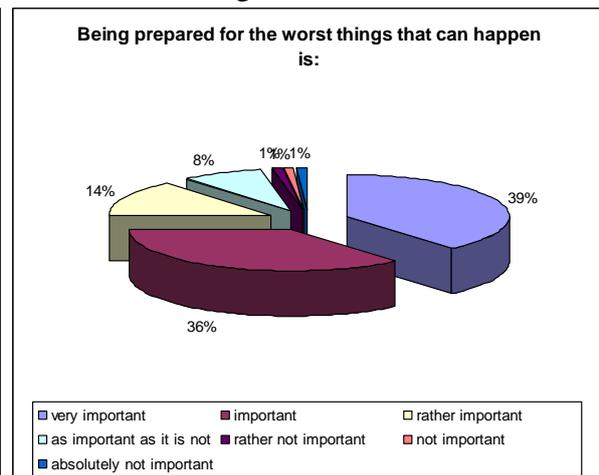
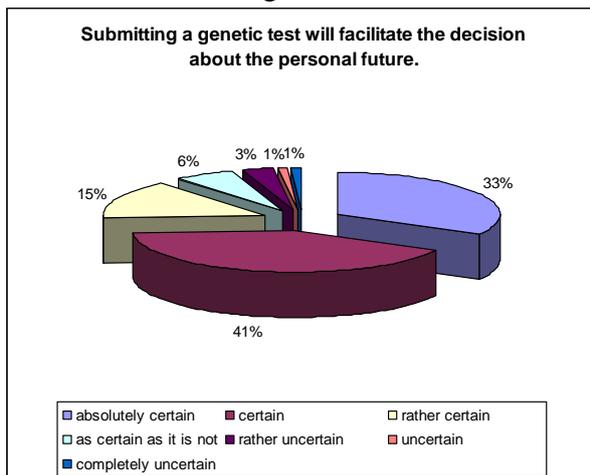
fig. 4



Being aware of the situation, when it is necessary to take an important decision about your full life, considered for extremely important, 59% of respondents. / fig. 4/

fig. 5

fig. 6

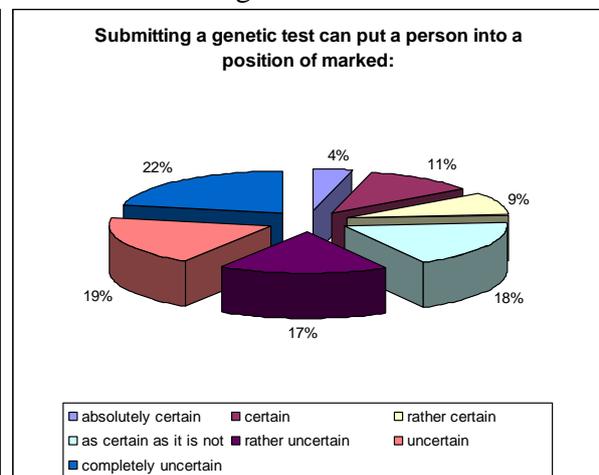
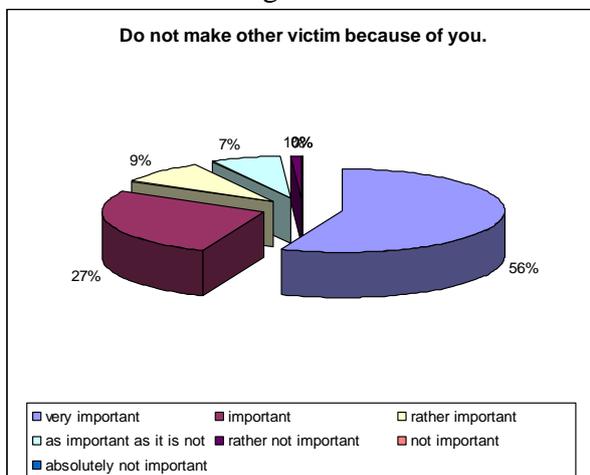


A significant percentage of them consider that the analysis of the results of a genetic test will also help facilitate their individual choice for a "quality" life. /fig.5/

Being informed and prepared for the difficulties that have to stand up and fight, say 39% of participants in the statistical study. / fig. 6. /

fig. 7

fig. 8



Not to be a burden to the family, not encumbered with extra care and cost their relatives, do not suffer another because of their health condition, disturb a large percentage of the respondents. /fig.7/

fig. 9

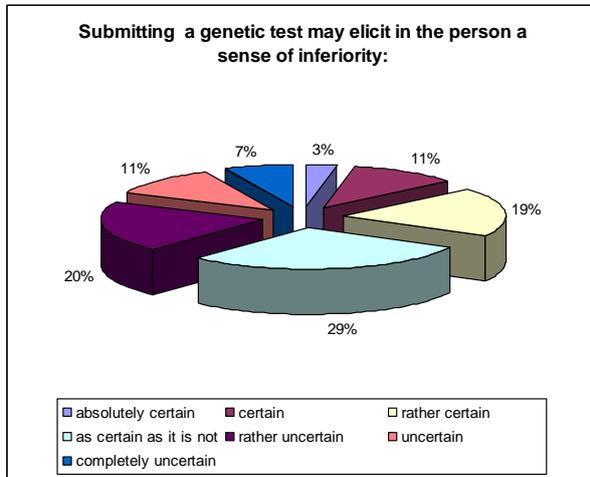
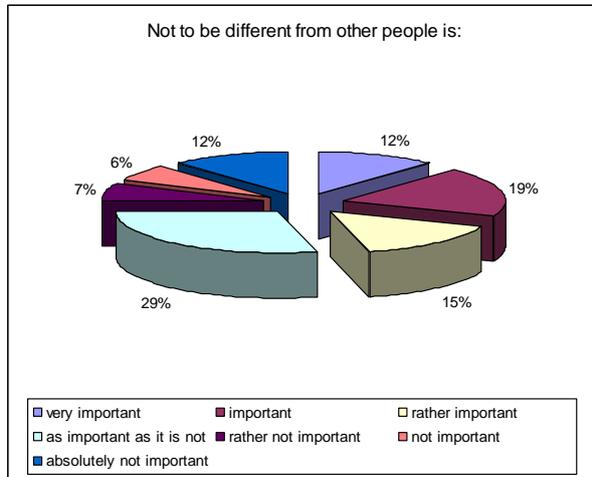


fig. 10



The results of genetic testing may cause deep psychological trauma for the patient, to awake a sense of inferiority, depression, alienation from family and friends./ fig. 8, 9 and 10/

The result of a genetic test can be used for decision of discontinuation of high risk pregnancy, family planning and creating a healthy generation. Results that predict the probability of developing an inherited disease may influence the choice of education, career, raise a family, conclusion and dissolution of marriage. The decision to undergo genetic testing and subsequent interpretation affect the privacy of the test person, but also on other members of the family. Psychological consequences of testing are coming especially with the results.

They can change the way that the patient thinks about himself, to influence his relationship with family and friends. In such a situation it is possible to create a sense of isolation, alienation, discrimination by individual mutation carriers. Conversely, other genetically unencumbered family members to feel blame for having escaped the gene defect.

Conclusions:

Genetic testing is part of the prophylactic activity in clinical genetics. The results of genetic testing are not only important for the health of the individual, but have a significant impact on his psychological and even social status. Adopting the presented genetic information, the patient determine the further behavior and chooses one or another course of action, measures for the treatment, prevention and lifestyle.

Reference:

1. Halsey Lea D., Williams J., Donahue P., Ethical Issues in Genetics Testing, Journal of Midwifery & Women's Health, 2005; 50 (3): 234:240
2. Norrgard k., Ethics of Genetic Testing: Medical Insurance and Genetic Discrimination. Natural Education1 (1), 2008
3. Burgess M., Laberge C., Knoppers B., Ethics and genetics in medicine, Bioethics for Clinicians, Journal of Canadian Medical Association /CMAJ/, may 19, 1998, 158 (10) 1309
4. Dimitrova I., Prenatal Diagnosis and Biopolitics in Bulgaria, Sofia, 2012