

The Role of Genetic Testing in Cancer Prevention

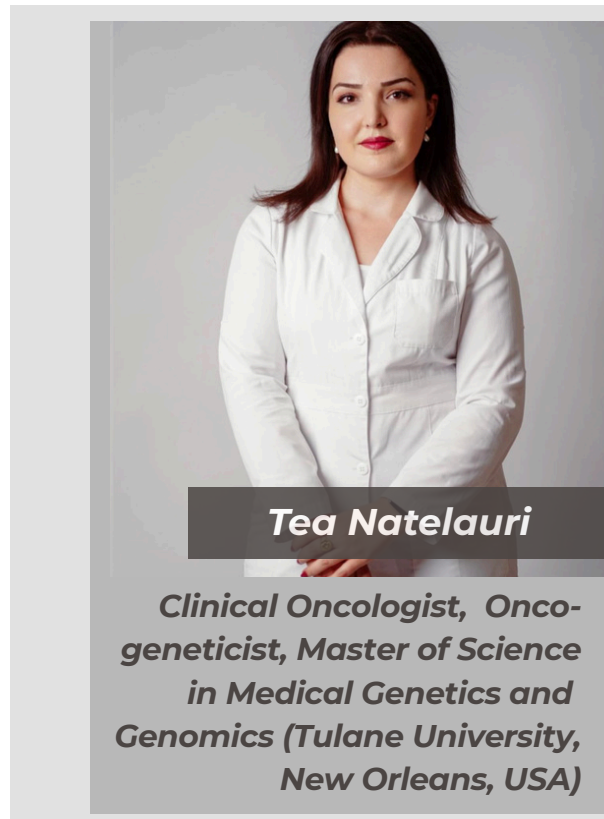
Dr. Natelauri, thank you for taking the time to speak with us. Please share your perspective: What role does genetic testing play in the prevention and early detection of cancer?

The role of genetic testing in modern medicine, including oncology, has increased significantly. Through genetic testing, it is possible to assess the risk of developing oncological diseases. Hereditary cancer syndromes, which account for approximately 10% of all cancer cases, can be detected through genetic testing even before the disease manifests clinically. Identifying a genetic predisposition to disease (for example, BRCA1/2 mutations or Lynch syndrome) enables both clinicians and patients to plan preventive measures. These may include intensive screening or other types of interventions, such as prophylactic surgical procedures or chemoprevention (in specific cases).

Furthermore, when a pathogenic variant is detected, genetic testing often helps protect the patient's family members as well—it makes cascade testing necessary, which is crucial for determining their individual risk. In addition, genetic testing plays an important role in treatment personalization: certain genetic mutations determine the effectiveness of specific therapeutic agents, such as in the use of targeted therapy.

How easy is it to integrate genetic testing into everyday clinical practice, and what challenges do you see in this process?

There are quite a few problems; however, the main barrier is the high cost of genetic testing



and the fact that insurance companies do not cover these expenses.

An additional challenge is the limited laboratory infrastructure and the reliability of local testing — there is a shortage of highly reliable, internationally accredited laboratories. A significant problem also remains the shortage of specialists: the number of genetic counselors and oncogeneticists in the country is very limited.

Moreover, another difficulty lies in the proper interpretation of genetic testing results by other medical professionals.

How informed are patients in Georgia about genetic testing, what is the level of

their engagement, and what steps could be taken to increase awareness?

Unfortunately, the level of awareness among Georgian patients remains low; however, we are making efforts to raise awareness of this issue.

It is also noteworthy that limited financial accessibility leads to lower levels of patient engagement.

To increase awareness, we carry out various activities in collaboration with MegaLab. These include presenting our services to colleagues at scientific conferences so that they can share this information with patients, as well as conducting public information campaigns, distributing brochures, and sharing videos through social media.

The active involvement of foundations and partner civil society organizations is also crucial.

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Are there ethical or social challenges associated with the use of genetic testing, and how can they be addressed?

One of the main challenges is ensuring confidentiality and preventing discrimination, which remain significant concerns globally. It is essential to establish a clear legal framework that explicitly prohibits insurance companies and employers from discriminating on the basis of genetic testing results.

In addition, the psychological impact must be taken into account: if a pathogenic variant is detected, the patient may experience uncertainty and fear, making psychosocial

support particularly important.

Furthermore, genetic testing—or its misinterpretation—can influence a patient’s decisions regarding family planning or create a negative perception of their existing risk, which constitutes a significant social and ethical concern.

What future innovations or advances in genetic testing do you expect to enhance the effectiveness of cancer prevention?

In the future, we anticipate that the issue of funding for genetic testing will be reconsidered, which would help increase demand for local testing—particularly for next-generation sequencing (NGS) in Georgia—eliminating the need to send samples abroad.

As a result, the cost of locally conducted NGS testing is also expected to decrease. However, at this stage, the price of such tests unfortunately still reaches four-digit amounts.

In the long term, in addition to NGS panels, we plan to introduce the use of polygenic risk scores (PRS)—a multi-gene risk assessment tool that, when integrated with clinical factors, can be translated into personalized recommendations.

The use of liquid biopsy is also expected to expand. Currently, it is primarily employed for disease monitoring—in cases of metastasis and minimal residual disease detection—but in the future, it could also serve as a screening tool. Detecting early traces of neoplasia in the blood could become a simple and potentially widely accessible method of early detection.

There will also be greater automation in result interpretation and increased support from artificial intelligence (AI), particularly in variant classification, clinical significance assessment, and the development of risk-based

recommendations.

In addition, tele-genetics and remote consultations are expected to become more widely accessible, expanding the availability of genetic services to a broader population.

