

Cancer: Genetic testing can save lives

Track chances of having a tumor's tendency among people with a history of disease cases in the family, but practice should not be trivialized and hurt even more who have excessive fear of developing diseases

By Valeria Mendes - **Full Health**

Publication: 25.09.2015 10: 00 Atualização: 09/28/2015 10:38

(original website, published in Portuguese:

http://sites.uai.com.br/app/noticia/saudeplena/noticias/2015/09/25/noticia_saudeplena,155218/cancer-teste-genetico-pode-salvar-vidas.shtml)

It is estimated that between 5% and 10% of malignant tumors are hereditary in origin, that number is one in ten of the 576,000 new tumor cases planned for this year in Brazil. It is now possible to predict whether a person will develop a malignant tumor and act before it manifests itself. The challenge now is to get genetic testing reach in every corner, people who can benefit from this technology. The survey, which can be done with saliva or blood, it is important to discover the changes in genes related to rare tumors and the development of cancer due to the high incidence of the disease in the family.

It was the news of the removal of the breast, ovaries and fallopian tubes of the actress Angelina Jolie - who lost his mother, grandmother and an aunt to cancer - which brought the issue of oncogenetics to the media. The impact of the Hollywood star's decision is so great that, on the one hand, the wife of Brad Pitt brought to public knowledge the possibility to do a genetic test to identify the mutation of the BRCA1 and BRCA2 genes, which predisposes to the development of tumors already mentioned, it also promoted a 'race to the clinics' patients in search of tests that give them an answer on the probability of developing cancer in the future and when the act before.



Selma Gromboni (esquerda) é mãe de Marcelo, 24, e Mayara, 19 diagnosticados com Síndrome de Li-Fraumeni

The nonsense of this whole story is that the disclosure of genetic testing can predict malignant tumors did not impact just who is considered a risk group, but fueled the search for those suffering from cancer phobia, an exaggerated fear of being affected by a tumor. A person with no family history of cancer or rare tumor does not enter the screening criteria and can fall victim to bad practices. Thus, a breakthrough medicine as genetic counseling may fall into mere guesses, no scientific evidence of benefit for the person or technical support. Coordinator of the Center of Oncology Network Mater Dei Health, Enaldo Melo de Lima remember that most cancers are sporadic, without any inherited factor.

On the other hand, the history Jolie failed was distribute information that is already known in medicine several other genes (other than BRCA1 and BRCA2) which, when altered, increase the risk of development of various types of cancers as the colon (intestine), melanoma, pancreas, prostate, and a variety of rare tumors. According Enaldo Melo de Lima, have been identified and cataloged about 200 genes and 30 cause inherited cancer syndromes.

More chance of cure

In 2002, Carlos Alberto de Oliveira discovered a tumor in the kidney. He lived with his wife and two children in Jau, São Paulo. The father of Mayara student, now 19, and the journalist Marcelo, now 24, underwent surgery and for the assessment of the doctors who accompanied him, were not required chemotherapy or radiotherapy. Carlos was 41 years old. After the first tumor, others emerged until a cerebral metastasis killed him at age 44.

Soon after the first surgery of Carlos, his wife, Selma Gromboni, dealer, now 50, says that the family lived one second blow with the news of a lymphoma in the youngest, who was 7 at the time. The girl also received treatment in Jau, but in the year of his father's death in 2005, Mayara appeared with a second tumor. And there was given the plot twist that family.

That year, Selma Gromboni decided to take her daughter to a center specializing in oncology, AC Camargo Cancer Center, in São Paulo, and after genetic counseling, was suspected of Li-Fraumeni syndrome. The inherited disease characterized by high risk for the development of various types of tumors, especially breast cancer, sarcoma, central nervous system tumors and adrenal tumors. This syndrome is caused by mutation in the TP53 gene, which - in its unaltered state - acts as the primary tumor suppressor in the body.

Until 2003, Li-Fraumeni syndrome was considered as a rare syndrome, with only 280 cases identified, according to the World Health Organization (WHO). However, it has high incidence in Brazil. The country conducted studies show that there is a mutation in the TP53 gene founded that is specific to the Brazilian population, p.R337H.

Confirmation of the diagnosis of Mayara came from a genetic test that, at the time, nor was held in Brazil. It has gone through 13 surgeries to remove malignant tumors and 14 benign seven. The Li-Fraumeni syndrome also affects the eldest son of Selma. Marcelo had a brain tumor in 2010, but thanks to early diagnosis is cured. Besides him, a sister of 52 years and a brother of 58 Carlos were diagnosed

with the disease.

Since then, Mayara Marcelo and make periodic follow - her every three months and he, every six - so that, in case of a new tumor, the diagnosis be given early. "The fact that we know the syndrome clarified much. My children face head disease erected and deal very well with the whole situation. Mayara doing prep school to go into medicine and specialize in oncology. If we knew the syndrome at the time my husband discovered the tumor in the kidney, the story could have been different. All you can discover when it is still small, the chance of cure is greater, "said Selma.

Despite constantly facing challenges, Selma Gromboni states that the children want to marry and have children and ensures that, despite not having the disease, keeps their exams on time. "If I'm not well, I can not help my daughter and my son. Do my preventive examinations annually, "he says. For her, it is very important to disclose not only the Li-Fraumeni syndrome as any other to show that, with proper treatment, you can overcome cancer.



Maria Isabel Achatz

Cancer Tree

The medical geneticist and director of the Department of Oncogenetics the AC Camargo Cancer Center, created in 2001 and a pioneer in Brazil, Maria Isabel Achatz explains that the purpose of genetic counseling is to try to identify who are the individuals who have some hereditary change and define whether this person has a mutation that may explain the onset of the disease. According to her, although the genetic code is unique, science and medicine know the basis of this code that allows the specialist to investigate and specific regions of the DNA sequence.

Genetic counseling begins with a consultation at the Department of Oncogenetics where the expert will address information which refer to living habits and health of family members of first and second degree of the patient as parents, siblings, grandparents, uncles and cousins. Achatz explains that this consultation provides a survey of the entire history of cancer in a family: the type, at what age

manifested in individuals, in which relatives, if the tumor has been passing from generation to generation. "With this information we construct what is called heredograma, which is nothing more than a family tree of cancer," explains.

Faced with this information you can identify the possible hereditary syndrome that should be investigated. "Some stories are so typical that only the diagnosis already shows that that family has a greater clustering a type of cancer that is seen in the general population," he says.

When there is an indication, the next step is the molecular diagnosis through genetic testing, which will say whether or not the patient has inherited a genetic changes related to cancer. If, with this examination, is identified a mutation linked to cancer is possible to evaluate the risk that this person has of developing certain tumors and also investigate which family members may be inserted in the risk group.

Initially, this whole process is performed in a patient who has had or has a tumor. It is from the result of the examination of this individual who will identify the mutation in other family members. A simple way to explain, used by Maria Isabel Achatz, is to think of a code with 5000 letters. "If the expert has detected the exchange of a letter of this code in the patient with the tumor, he will not need to look at the 5000 letters of other family members. He will direct the change to see who inherited and who does not," explains. This also impacts the cost of the examination, since it is not necessary to search the entire DNA sequence. The genetic test to identify the mutation in the BRCA, for example, ranges from \$ 5 thousand and R \$ 15 thousand.

Preventive examinations or preventive surgery

With this information, according to the Cancer Center's coordinator of the Network Mater Dei of Health, the expert will offer the patient the performance of screening tests in order to diagnose the disease at an early stage or withdraw preventively a body or bodies as was the case with Angelina Jolie. "The surgeries represent a minority of the indications," said Enaldo Melo de Lima.

The decision of the risk reduction strategy is always based on studies conducted on thousands of people to find the best route to take. It is important to know that the patient follow-up in which the genetic alteration was detected allows the tumor to be identified so early it is sufficient that it be removed to eliminate the disease without even the need for chemotherapy or radiotherapy.



Enaldo Melo de Lima

Enaldo Melo de Lima also noted that genetic testing should be offered to the patient's family on the following criteria: positive family history, diagnosis of cancer before age 50, two cases of different cancers in the same individual, impaired cognition, history of cancer unusual gender (male breast cancer). The result of molecular analysis takes, on average, 45 days. The expert also explains that every patient who will make a genetic counseling consultation is required to sign a consent form established by the Ministry of Health and the National Health Agency (ANS).

The Network Mater Dei Health created the Department of Oncogenetics in 2013 and, according Enaldo de Melo Lima, demand is growing for genetic counseling consultations. "This is due largely to the orientation of the conduct of treatment ranging from the possibility of very early detection of disease to operation. You can make a more refined and personalized treatment for the patient with the tumor and preventive for family members," he considers.

Access

In Brazil, the Public Health System (SUS) does not offer molecular analysis broadly. The Ministry of Health states that the SUS guarantees full treatment to cancer patients. "With regard to tests for genetic counseling, research has been done and funded by MS and the National Council for Scientific and Technological Development (CNPq), through the National Network of Familial Cancer (RNCF)," said in a note to the entry.

He said "it is precisely the need for more scientific evidence on the subject which justifies funding research on familial cancer area. The incorporation of technologies - such as genetic testing in general, oncology - the health system should be made after evaluation studies of health technologies, in which the risks and benefits for the population should be assessed. "

Currently, even according to the MS, "the sequencing of the coding regions of the genes BRCA1 and BRCA2 is offered under the SUS, by health facilities to develop research in this area. These tests are done in women with a history of cancer in the family to identify increased risk of some specific types of breast and ovarian cancer, with medical advice and follow-up will be guided by this professional.

According to the National Health Agency (ANS), 45.6% of the population has health insurance, but even in the health insurance, access to genetic testing need to follow some rules laid down in guideline number 876 2013: it is mandatory that the prescription is made by a geneticist, specialty not widespread in the country, and comply with the rules established, which includes the screening and treatment of genetic disorders 29 and each of them has a different policy.