

CANCER GENETIC COUNSELING

ASESORÍA GENÉTICA EN CÁNCER

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Dear Publisher:

Cancer is a disease characterized by cell's malignant transformation and uncontrolled growth caused by multiple alterations in their genome¹. It is considered a public health problem as it affects millions in the world and causes death, mostly because it is not detected on time.

Cancer can be classified as: **1) Sporadic (~70%)**: usually affecting people 50 years old and older and caused by spontaneous mutations in an organ cell's genome due to ambient factors, exposure to carcinogens and aging, among others; **2) Familial (~20-25%)**: it is caused by mutations in low penetrance genes, where an hereditary pattern is not observed; and **3) Hereditary (~5-10%)**: it is caused by germ-line mutations in genes that are transmitted in an hereditary pattern, it can be inherited by descendants and increases the risk of developing cancer at a younger age compared to the sporadic cases.

So far, 200 hereditary syndromes that predispose cancer have been described², including breast/ovary cancer, Lynch syndrome, and hereditary diffuse gastric cancer, among others. If the doctor believes a patient's diagnosis is one of these cases, it is paramount to run a genetic test, seek genetic counseling, and later request genetic studies following certain instructions³ (Table 1).

Tabla 1: Instructions to refer patients suspected to have cancer

1.	Two or more members of the family with cancer
2.	Various members of the family affected by the same type of cancer
3.	Early apparicion of cáncer (younger tan 50 years old)
4.	Bilateralism when the affected organs are even and/or multifocality
5.	More tan one primary cáncer in one individual
6.	Cancer associated to defects of development

Genetic counseling in cancer provides: 1) an evaluation of the patient whose results include personal and familiar oncologic antecedence through the elaboration of a family tree.; 2) establishment of an hereditary cancer diagnosis following the clinical and genetic criteria for each syndrome; and 4) a complete and objective explanation of the results, the genetic causes that lead to the development of the cancer, as well as its implications. Also, due to the emotional impact a patient suffers after knowing the results, it is paramount to refer him/her to make a psico-oncologic evaluation.

The main objective of a genetic test, in these cases, is to identify the alteration responsible for the cancer. The American Society of Clinical Oncology⁴ recommends that doctors request genetic studies when a personal or familiar history suggests hereditary cancer, and when results can help doctors provide a diagnosis or influence the medical handling. Furthermore, there are criteria for choosing the ideal person for this study⁵: 1) to choose the person suffering cancer over the rest of the family members; 2) if there are few affected people, choose the one that suffered cancer at the youngest age or the one that has more than one cancer.

The results of the genetic study can be: 1) Negatives: when genetic alterations are not detected or benign variants or likely to be benign are detected. 2) variants with unclear meaning: when we identify variants that so far have not been linked to a higher risk of developing cancer, but whose classification can vary in time; 3) Positives: when pathogenic or likely to be pathogenic variants are identify, allowing us to establish the patient's risks, the multidisciplinary handling and the possibility of searching for that variant in their families.

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In conclusion, cancer genetic counseling helps reduce the morbidity and mortality in patients with hereditary cancer and their family through a multidisciplinary handling that establishes preventive measures, early detection and risks control.

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