

Strategies for identification of familial and hereditary colorectal cancer

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In 10-15% of all colorectal cancers (CRC) genetic factors play a role in the etiology. The most common inherited form of CRC is the Lynch syndrome which is responsible for about 3-5% of all cases. Various studies have shown that surveillance of individuals at high risk for CRC prevents the development of CRC and improves the survival.

About one million individuals in Europe (population 730 million) have Lynch syndrome and five million have familial CRC. The main challenge is to develop appropriate tools to identify all individuals at high risk. Such strategies include those directed at the general population and those that aim to identify families through case finding. We performed a survey to evaluate the currently used strategies in twelve European countries. Questionnaires were distributed among members of the Mallorca group, a multidisciplinary group of experts in hereditary CRC with representatives from most European countries.

The survey showed that in all countries family history followed by referral to clinical genetics centres of suspected cases, was the main strategy to identify hereditary CRC. In five out of seven countries with a CRC population screening program, attention was paid in the program to the detection of familial CRC. Special campaigns to increase the awareness of familial CRC in the general population are organized in only two countries. In almost all countries, a family history is taken when a patient visit a doctor. However, all responders indicated that the quality of the family history was poor to very poor. Microsatellite instability testing (MSI) or immuno-histochemical analysis (IHC) is usually recommended as a tool to select high risk CRC-patients for genetic testing. The survey showed that the Bethesda criteria are used in most countries to select CRC-patients for MSI- or IHC-analysis. In one country, IHC was performed in all cases of CRC. In almost all countries, specialists have guidelines for referral of patients suspected of inherited CRC to clinical genetic centres. However, general practitioners have such guidelines in only about half of the countries. In eight of the twelve countries general patient information pamphlets on CRC also include information on hereditary CRC. In most countries there are no specific programs on cancer genetics in the curriculum of medical doctors.

In conclusion, the outcome of this survey may be used to improve the strategies to identify all individuals at high risk of CRC. First of all, more attention should be given to the awareness of hereditary CRC in the general population. IHC-analysis of all CRCs may be an effective tool to identify all Lynch syndrome families. However, the cost-effectiveness of this approach should be evaluated before introducing this approach in all European countries. All countries with a CRC population screening program should pay attention to the family history within the program.

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