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Critical Reviews in Oncology/Hematology 83 (2012) 329–340

CRITICAL REVIEWS IN
Oncology
Hematology
Incorporating Geriatric Oncology

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Surveillance for hereditary cancer: Does the benefit outweigh the psychological burden?—A systematic review

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Accepted 25 January 2012

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Abstract

Individuals at risk for developing hereditary cancer are offered surveillance in order to improve the prognosis. An important question is whether the benefit of surveillance outweighs the psychological burden. In this review, we evaluated all studies that investigated psychological distress and the quality of life in individuals under surveillance for hereditary cancer of the breast, ovarian, prostate, pancreas, colorectum, melanoma, and various rare syndromes such as familial adenomatous polyposis, Li–Fraumeni and Peutz–Jeghers syndrome.

Thirty-two studies were identified. Surveillance for most hereditary cancers was associated with good psychological outcomes. However, surveillance of individuals at high risk for developing multiple tumors appeared to be associated with increased distress and a lower quality

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1040-8428/$ – see front matter
doi:10.1016/j.critrevonc.2012.01.004

of life. Common factors associated with worse psychological outcomes included a personal history of cancer, female gender, having a first degree relative with cancer, negative illness perceptions and coping style. The use of a simple screening tool to identify distressed individuals is recommended.

Keywords: Hereditary cancer; Familial cancer; Genetic risk; Surveillance; Screening; Psychological burden; Distress; Quality of life

1. Introduction

The clinical relevance of hereditary cancer lies in the fact that family members with an increased risk may be helped by surveillance and by advice about prevention.

Inherited factors are predominant in 5–10% of cancers, which includes, for example, hereditary breast, colorectal (Lynch syndrome) and prostate cancer as well as rare tumor syndromes. Rare tumor syndromes characterized by the development of multiple tumors at various sites are, for example, Von Hippel Lindau syndrome (VHL), familial adenomatous polyposis (FAP), and Li–Fraumeni syndrome (LFS) which account for a small proportion of cancer incidence.

In the 1990s, the underlying gene defects have been discovered for most forms of hereditary cancers including mutations in the *BRCA-1* and *2* genes associated with hereditary breast and ovarian cancer, mutations in the mismatch repair genes associated with Lynch syndrome, mutations in the *APC*-gene responsible for FAP and many other gene defects. The identification of these gene defects allowed a presymptomatic diagnosis and differentiation between carriers and non-carriers. A major advantage of genetic testing is that surveillance can be focused on the carriers and that the non-carriers can be re-assured and refrain from follow-up.

In 1968, a publication of the WHO formalized criteria for population screening programs that should be met before a screening program for a specific condition is instituted [1]. Over the years, the criteria have been extended and updated [2]. According to these criteria, the condition should be an important health problem in the target group, the natural course of the disease must be known, a sensitive and specific screening test should be available, there should be an accepted treatment, there should be evidence for improvement of the prognosis due to surveillance and the costs should be acceptable. Another important criterion is that the benefit of surveillance should outweigh the physical and psychological harm associated with the program. The psychological harm may include an increased awareness of being at high risk of developing cancer, the need for periodic examination, the stress before and after the surveillance examination and the burden of the examination itself.

Many studies have been performed that evaluated the clinical benefit of surveillance for several inherited forms of cancer in terms of early detection or improved prognosis. In some hereditary cancer syndromes, such as hereditary breast cancer (HBC), Lynch syndrome, FAP and familial melanoma, there is substantial evidence that surveillance improves the prognosis; however, the benefit of surveillance is largely unknown for familial pancreatic cancer, hereditary prostate cancer, ovarian cancer and most of the rare tumor syndromes such as LFS, VHL and Multiple Endocrine Neoplasia (MEN) syndrome type I [3–10].

In recent years, also an increasing number of studies have been reported on the psychological impact of surveillance. The aim of this review is to investigate the current knowledge on the psychological burden of surveillance in various forms of hereditary cancer and to discuss whether the clinical benefit of surveillance outweighs possible psychological disadvantages.

2. Methods

2.1. Search strategy and selection criteria

To perform a systematic literature search the databases PubMed, Web of Science, Current Contents and Medline were used covering a period between 1985 and June 2011. To search for studies that reported on psychological aspects of participants in surveillance programs for hereditary cancer, search terms were: “hereditary cancer”, “familial cancer”, “genetic risk”, “high risk”, “screening”, “surveillance”, “distress”, “stress”, “psychological”, “psychosocial”, “burden”, “pain”, “quality of life”, and “mental health”. The authors (JG, HV) reviewed the abstracts of the search results and selected the articles relevant for the present review.

Inclusion criteria were: (a) empirical studies published in scientific journals in the English language; (b) studies evaluating the psychological burden of surveillance and various psychological aspects related to surveillance (for example, an increased awareness of being at high risk, the need for periodic examination, the stress before and after the surveillance examination and the physical burden (e.g. pain) of the examination itself); (c) studies concerning individuals being affected/at risk for hereditary cancer for melanoma, cancer of the breast, ovaries, prostate, pancreas as well as individuals at risk for Lynch syndrome, and the rare syndromes LFS, VHL, FAP, Peutz–Jeghers and MEN-1; (d) studies including carriers of a mutation or untested individuals from families with a known genetic defect or members of families that met clinical criteria for familial or hereditary cancer; (e) cross-sectional as well as prospective studies were incorporated. References from the retrieved articles were scanned to further identify more studies.