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Screening and Survival in Familial Adenomatous Polyposis

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The key issue for all cancer screening efforts is: Does screening improve survival? If a screening approach for a particular cancer is found not to improve survival then screening cannot be recommended. Other screening approaches must be found. In many types of cancer screening with presently available tools simply has not been found effective and therefore is not recommended. For colon, breast, cervical, prostate and melanoma, however, screening has been found to improve survival in the general population and is therefore a recommended. For each of these, however, caveats have arisen as screening efforts have been applied. For example, there are differing ages to begin and end for each and annual PSA for prostate cancer is no longer recommended as a general approach.

Screening approaches in less common but higher risk populations are often even more effective than those for the general population. This is particularly true for colon, breast and melanoma cancers where inherited genetic syndromes have been identified and genetic testing can be done for diagnosis. Those thereby found to have one of the inherited conditions should be involved in specifically tailored screening strategies to maximize cancer prevention and mortality reduction.

The study in this volume by Koskenvuo, et al, demonstrates that screening in those with FAP who are called up as known relatives of a proband with FAP improves survival at 20 years following diagnosis equivalent to that of the general population! (1) This is a rather astounding finding considering the malignant potential of this disease. Without therapy the risks of colon cancer in FAP patients is 7% by age 21 years, 50% by age 39 years and near 95% by age 50 years. Even with modern colon cancer therapies, survival after cancer diagnoses in FAP would be considerably lower compared to the general population. A number of previous studies have demonstrated that improved survival in FAP can be accomplished with screening. But this study is quite unique in that it finds that screening of the call-up group leads to a survival equivalent to a comparative general population at 20 years from the FAP diagnosis. It is uncertain how long this survival equivalence will persist as serious problems related to rectal cancer and desmoid tumors in particular are known to be frequent causes of morbidity and mortality following colectomy.

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Nonetheless, the results of this study are incredibly important as they encourage attending physicians to be certain to have family members of a person known to have FAP notified and screened for colon cancer prevention. The findings can also be of substantial encouragement to persons and families with this condition with the knowledge that proper surveillance and therapy will allow survival similar to the general population for at least 20 years.

But how does screening in call-up relatives of those with FAP improve survival. The first reason is intuitive. In a disease with such extreme colon cancer risks at young ages, identification of relatives, most of which will be younger than the proband, will allow premalignant screening and appropriately timed colectomy for most. Colon cancer is thereby prevented. This is clearly borne out in the findings of the present and other studies. The second reason may not be as obvious. When a registry effort promotes a call up strategy for FAP relatives, those identified become part of that registry. They are made aware of all aspects of the disease. They learn appropriate prevention guidelines. And they in turn will often notify other relatives and further improve outcomes. They also will have regular communication with the registry that initiated the call-up for any questions concerning symptoms and/or management. Attending physicians outside the registry or center of excellence will likewise be made aware of management guidelines and will be in contact with that center when needed.

In summary, this study in general is further demonstration of the importance of rare disease management by a registry or coordinating center with the needed expertise. A center of excellence in FAP will in turn guide attending gastrointestinal physicians in proper patient finding, recommended screening guidelines, surgical timing, optimal surgical procedures and updates of disease management as they develop. The center can assist patients, families and physicians in critical areas of decision making and even see patients for particular needs in consultation with managing physicians. And finally, the rather remarkable mortality improvement demonstrated by this study provides considerable motivation for affected persons and attending physicians to be certain that those with FAP and their families receive appropriate knowledge and care to decrease and often eliminate colon cancer risk.

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