



<b>Title</b>	<b>Genetic-guided screening programme for familial adenomatous polyposis: result of a regional registry</b>
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**Conclusion:** Transrectal ultrasound-guided trucut biopsy of prostate is a safe and effective method for diagnosing carcinoma of prostate. With the help of Digital rectal examination and PSA level it may help in the early detection of curable cases.

### 10.9 Genetic-guided screening programme for familial adenomatous polyposis: result of a regional registry

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Familial adenomatous polyposis (FAP) is an autosomal dominant condition due to germline mutation of the APC gene resulting in hundreds to thousands of adenomatous polyps in the colorectum with inevitable malignant transformation if left untreated. Offsprings of affected individual have a 50% chance of inheriting and developing the disease. Screening of these at-risk first-degree relatives (FDR) is advocated to allow presymptomatic diagnosis, prevent malignant transformation and reduce colorectal cancer mortality. We report the result of our Registry's genetic-guided screening programme. For FAP families with surviving index patients, blood were taken for genetic diagnosis using a combination of protein truncation test and direct nucleotide sequencing. In those families with positive genetic diagnosis, blood were taken from at-risk FDR for genetic screening. Of the 17 eligible families tested, mutations were detected in 14 families to date (rate: 82.4%). There were 8 deletions, 2 insertions and 4 nonsense mutations detected in the APC genes. Twenty-four at-risk FDR (14 male and 10 female) at a mean age of 32.6 years (range: 13-84) were subjected to genetic screening. Three FDR were found to be mutated gene carriers and 21 were normal gene carriers. The genetic screening result corresponded completely with the baseline endoscopic screening result. Mutated gene carriers were offered prophylactic surgery when polyps develop. The normal gene carriers were discharged from further clinical screening. As compared with the traditional clinical screening programme, we were able to avoid 187 unnecessary sigmoidoscopies in normal gene carriers. Although genetic diagnosis and screening may be expensive initially it saves money in the long term by avoiding unnecessary sigmoidoscopies. In our families, this represents 52.3% of cost reduction. Furthermore, genetic-guided screening allows more accurate risk assessment of at-risk FDR and avoid unnecessary psychological burden in normal gene carriers. It can also help to improve compliance in subsequent screening and treatment of mutated gene carriers.

### 11 Evidence-based medicine

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Evidence-based medicine (EBM) has been defined as "the conscientious, explicit, and judicious use of current best evidence in making decisions about the care of individual patients". It involves formulating answerable questions about specific clinical problems, searching the relevant medical literature, assessing the validity and usefulness of the evidence that has been identified, and then applying that evidence in the care of individual patients.

Claimed by its proponents to represent a new paradigm in medical practice, EBM demands new skills from physicians in evaluating the clinical literature at the same time as it de-emphasises the unsystematic application of clinical experience. This new approach is not without its critics, however, who argue that such critical appraisal of the literature is unrealistic for busy clinicians, that it is often hard to apply information from population-based studies to the care of individual patients, and that the value of hard-won clinical expertise should not be discounted. This presentation will provide an overview of EBM, and look at both sides of the debate.