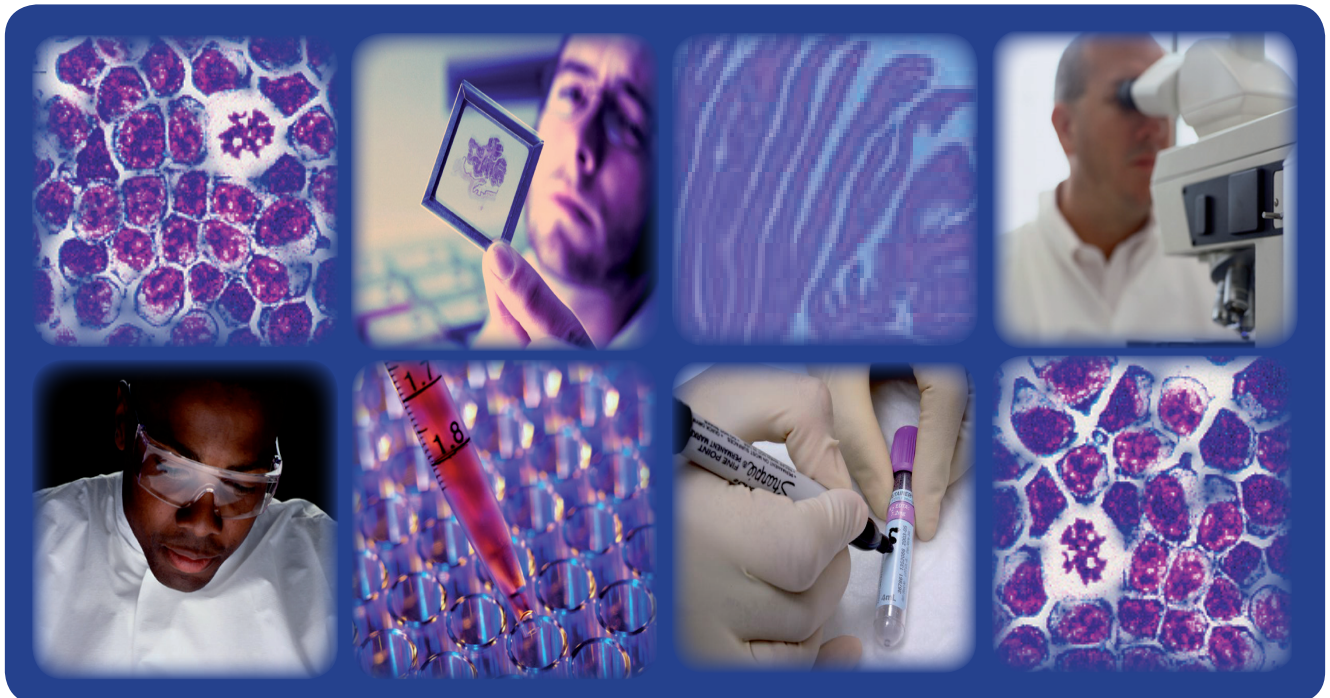


# Biomarkers in familial colorectal cancer screening

Expert workshop, 14th February 2006



## Executive summary



**Public Health Genetics Unit**  
Cambridge, UK

Dr Philippa Brice  
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## **Biomarkers in familial colorectal cancer screening - background**

Hereditary non-polyposis colorectal cancer (HNPCC) screening is performed where colorectal cancer patients are identified as having a high risk of HNPCC or Lynch Syndrome based on their family history and/or clinical features. The main aim of screening is the identification of at-risk, asymptomatic relatives in order to prevent advanced disease and mortality in these individuals, and to exclude relatives who are not at risk from unnecessary clinical surveillance. Germline mutation testing is expensive and is not appropriate for first-line screening. However, there are two techniques for pre-screening tumours by the analysis of biomarkers: microsatellite instability (MSI) and immunohistochemical (IHC) analysis of colorectal tumour samples. Most experts agree that one or both of these approaches should ideally be used prior to germline mutation testing, but UK clinical practice varies between different centres, and no guidelines exist to the use of these techniques as part of HNPCC screening.

The main objective of the expert workshop held in London on 14th February 2006 was to bring together professionals with an interest in the diagnosis of HNPCC (including molecular and clinical geneticists, genetic counsellors, pathologists, colorectal surgeons and policy experts), in order to evaluate current evidence for and against different approaches, and move towards development of a consensus optimal strategy for national implementation. Delegates heard presentations on HNPCC screening including the selection of families for screening, the use of MSI, IHC and mutation analysis in the identification of HNPCC, and economic aspects, along with a summary of current practice in the UK. The workshop then held discussions on whether the techniques should be adopted and in what form, and considered key issues and barriers for the creation of a UK strategy for best practice in the diagnosis of HNPCC. The workshop steering group then developed recommendations based on these proceedings.

## **Recommendations**

- 1. Both MSI and IHC testing for colorectal tumours should be available across the UK.**
- 2. Current evidence should be used to agree on a national testing strategy, to include clear referral guidelines for colorectal cancer patients and protocols for investigation using MSI, IHC and mutation screening as appropriate.**
- 3. Results of MSI and/or IHC testing, wherever that testing is done, should be reported back to the hospital where the tumour was diagnosed and should be included in the patient's tumour pathology reports, including electronically wherever possible.**
- 4. An appropriate auditing system for the national testing programme should be put in place; this will generate the necessary data to determine the most cost-efficient use of the two techniques for the identification of HNPCC (and potentially refine the national strategy).**
- 5. A quality assurance process should form part of the national strategy.**
- 6. Assessment of MSI and IHC test results should involve a multidisciplinary team including clinical and laboratory geneticists and histopathologists, irrespective of which laboratory performs the tumour block preparation, DNA extraction and testing.**
- 7. IHC and MSI testing of tumours should be performed in CPA accredited laboratories that have staff with the appropriate skills and expertise; implementation will require additional investment of time and resources.**
- 8. A co-operative approach and excellent communication between different testing locations is essential to optimise the identification of familial colorectal cancer cases.**
- 9. Access to tumour tissue blocks and related issues, such as consent, must also be addressed as part of the national strategy.**
- 10. The UKGTN should, as with other novel genetics laboratory services, provide oversight of national strategy implementation via selected laboratories, and through the system of gene dossiers incorporate future technological developments.**