

# PROPOSAL FOR INTERNATIONAL RESEARCH PROJECT

Dr Bente Talseth-Palmer

## ROLE OF GENETIC MODIFIERS IN DISEASE RISK - LYNCH SYNDROME

Over the last 10 years I have been leading a research project from Australia, together with Prof. Rodney J. Scott, looking at modifying genes/alleles in patients with a molecular diagnosis of Lynch syndrome. We aim to identify additional genetic factors contributing to disease risk in Lynch syndrome patients that will be instrumental in personal screening programs for these patients. Since the identification of the genetic basis of HNPCC in early 1990's many studies have been undertaken to identify modifier genes that may explain, at least in part, the variation observed in disease expression in these patients (tumour site and age of onset).

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One of the challenges of conducting genetic work on an inherited disease is getting enough patient samples to produce statistically reliable results.

As a result of increasing sample size by expanding international collaborations (from ~200 samples to today's ~1350 samples), we have demonstrated that even though both MLH1 and MSH2 mutation carrier's starts off with the same risk of CRC, other genetic factors are also associated with a differential risk of developing CRC, specifically for MLH1 mutation carriers. But still controversial results are reported.

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We therefor aim to collect as many Lynch syndrome patient samples (DNA from blood samples) as we can. We have DNA samples from New South Wales, Australia and Poland. Currently I am in the process of collecting all Norwegian Lynch syndrome samples and we have genotype data from the Netherland when needed.

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The results from this study will lead to routine use of molecular tests to identify patients with further increased risk of colon cancer

### Current collaborators:

- Bente Talseth-Palmer, Australia/Norway
- Rodney Scott, Australia
- Jan Lubinski, Poland
- Hans Vasen, Netherlands
- Juul Wijnen, Netherlands
- Wenche Sjursen, Norway
- Pål Møller, Norway
- Hildegunn Vetti, Norway

If you are interested in participating in this study, please contact me during the meeting or send an email to:

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