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Polymorphisms of DNA repair genes and disease risk

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Large scale population-based cohort prospective studies offer the most comprehensive approach to understand what is contributing to disease. Data on general lifestyle, physical activity, diet, reproductive factors, occupational exposure and other factors linked to human activity are necessary for the evaluation in the cohort study. These data were mostly obtained through direct interview or self-administered questionnaire. Anthropometric measurements and clinical laboratory findings are also collected for data entry. In a complex interaction, risk from the environmental exposures is modulated by genetic susceptibility factors. With the development of technological tools in molecular epidemiology, we have the potential to clarify the contribution of environmental factors to disease causation and to identify high-risk groups and individuals for purposes of prevention. In general, conventional epidemiological studies have not been regarded to collect biological samples. Now, there is a new move to start to collect biological materials for epidemiologic analysis because of limited analysis of risk factors without consideration of genes and gene-environmental interaction.

Since DNA repair gene is particularly important for repairing DNA damage by extrinsic factors such as smoking or ionizing radiation exposure, the genetic polymorphism associated with DNA repair may affect disease risk such as cancer. So, we hypothesized that XRCC1 or ATM genetic polymorphisms contribute to increased risk of colorectal or lung cancer. We found significant associations between XRCC1 or ATM polymorphisms and colorectal or lung cancer risk. We also found that XRCC1 polymorphisms affect survival after treatment with radiotherapy for patients with lung cancer. Interestingly, these genetic polymorphisms are also known to influence risk of subcutaneous fibrosis and telangiectasia after radiotherapy.