'Junk DNA' could spotlight breast and bowel cancer

5 January 2010

Scientists at The University of Nottingham have found that a group of genetic rogue elements, produced by DNA sequences commonly known as 'junk DNA', could help diagnose breast and bowel cancer. Their research, funded by Cancer Research UK, is published in this month's *Genomics* journal.

The researchers, led by Dr Cristina Tufarelli, in the School of Graduate Entry Medicine and Health Sciences, discovered that seven of these faulty genetic elements — known as chimeric transcripts — are more common in breast cancer cells. Five were only present in breast cancer cells while two were found in both normal and breast cancer cells.

These rogue elements are produced by DNA sequences called LINE-1 (L1). Despite being labelled as 'junk DNA' it is clear that some of these sequences have important roles in the genome, such as influencing when genes are switched on.

L1s carry a switch that is able to randomly turn on nearby genes. When genes are inappropriately switched on in this way they make the genetic rogue elements that can sabotage the normal functioning of cells. To prevent the potentially damaging effects of these rogue elements, normal cells silence L1s with a chemical 'off switch'. In cancerous cells this 'off switch' is often missing, leading to the production of these genetic rogue elements.

Dr Tufarelli, a lecturer at the University who has also received funding from a Royal Society Dorothy Hodgkin Research Fellowship, said: "This study has generated new research tools to investigate the role of 'junk DNA' in cancer development. The next step is to find out if the switching on of these genes is driving cancer or if they are a result of the cancer. Even if they are innocent bystanders of cancer they could be useful biomarkers helping us to diagnose or monitor the disease."

The researchers extended their studies to look at two bowel cancer cell lines. Two of the genetic rogue elements were found in invasive bowel cancer cell lines, but not in the pre-invasive cells, suggesting that these sequences could play a role in cancer progression.

Dr Tufarelli said: "If this 'junk DNA' does turn out to play a role in cancer then we could be at the tip of the iceberg in understanding a completely new mechanism behind the disease. If we do find out that they are playing a role then they could be useful targets for new treatments."

Dr Lesley Walker, Cancer Research UK's director of cancer information, said: "These really interesting findings are the most comprehensive study of these transcripts that have ever been carried out. We are learning more about the genes involved in cancer but these so-called 'junk' regions receive relatively little attention. We are beginning to see that they could play a really important role."

Provided by University of Nottingham