Epigenetic changes and disease – what is the connection?

September 20 2017

Previous research has shown that there is a connection between epigenetic changes and some of our common illnesses. But what does this connection mean? A new study shows that external factors, such as lifestyle aspects, often affect both the epigenetic pattern and cause the disease. The results have been published in *PLOS Genetics*.

The risk of developing our most common diseases depends on inheritance, lifestyle and environment. In recent years, epigenetic changes have also been assumed to increase the risk of various diseases. Epigenetic changes are chemical modifications of the DNA that turn genes on or off. In the current study from Uppsala University, researchers show that epigenetic changes mainly occur in response to a disease, rather than being an underlying causal risk factor.

Today, we know that the environment we live in and our lifestyle can lead to epigenetic changes as well as to increased risk for various diseases. Earlier studies have shown that there is a strong link between epigenetic changes and the risk of various diseases such as myocardial infarction, asthma, rheumatism and schizophrenia. However, it has not been known whether such epigenetic changes are the cause of the diseases or if they arise due to the disease, or whether both epigenetic changes and the development of a disease are due to external factors, such as a person's lifestyle.

In the current study, the researchers investigated the relationship between genetic variation, epigenetic changes, and lifestyle factors in

relation to biological markers for cardiovascular diseases, so called biomarkers.

"The results show that the link between epigenetic changes and disease status is largely due to external factors, such as the lifestyle, or due to the genetic variation we inherit from our parents," says Åsa Johansson, researcher at the Department of Immunology, Genetics and Pathology, who led the study.

For example, the researchers showed that smoking affects both the epigenetic pattern and the presence of different disease biomarkers.

"Such cases can easily be interpreted as the epigenetic changes causing the disease, when it's actually smoking that is the causal factor," says Åsa.

Similarly, the researchers could see that genetic variation also affected the epigenetic pattern and many disease biomarkers. However, they found no evidence that epigenetic changes could affect the disease risk. The researchers hope that the study should contribute to a better understanding of what role epigenetic changes play in disease pathogenesis, where epigenetic changes should be recognised as markers of different exposures rather than something causing disease.

More information: Muhammad Ahsan et al. The relative contribution of DNA methylation and genetic variants on protein biomarkers for human diseases, *PLOS Genetics* (2017). DOI: 10.1371/journal.pgen.1007005

Provided by Uppsala University