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Epigenetics: New Tool for Precision Medicine

Four new papers, co-published by an international consortium of biomedical researchers, mark the feasibility of epigenetic analysis for clinical diagnostics and precision medicine. Epigenetic analysis addresses key limitations of genetic testing, helping to ensure that patients are accurately diagnosed and treated with the right drug at the right time.

(Vienna, 23 June 2016) Epigenetic changes occur in all cancers, and in various other diseases. Measuring these changes provides unprecedented insights into the disease mechanisms at work in individual patients, which is important for better diagnosis and patient-specific treatment decisions.

In a series of four papers led by Christoph Bock (CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, Vienna) and Stephan Beck (University College London, UCL), an international group of scientists have validated the feasibility of epigenetic analysis for clinical applications.

Building upon years of technology development in laboratories around the world, this series of papers shows the accuracy and robustness of epigenetic tests. Going forward, clinical researchers will optimize and apply these methods for specific diseases, and it is expected that epigenetic tests will become widely used for selecting personalized treatments in cancer and other diseases.

Epigenetics refers to chemical modifications of the DNA and its associated proteins that control gene activity independent of the genetic code. These epigenetic modifications define how two meters of DNA in each human cell are folded into tiny cell nuclei.

Epigenetic modifications can be inherited during cell division, which helps maintain the hundreds of cell types of the human body carrying the same genes. Moreover, epigenetic mechanisms provide an interface by which the environment influences gene activity.

In many diseases, including all cancers, the epigenetic control of the genome is heavily distorted. Measuring these alterations provides a detailed picture of the disease-specific changes, which is often informative for distinguishing disease subtypes or identifying suitable treatments. Therefore, epigenetics has much to offer for improving disease diagnosis and treatment choice.

The now published studies, which have been performed in the context of the European BLUEPRINT project and the International Human Epigenome Consortium, constitute a milestone for utilizing epigenetic information in clinical diagnostics and precision medicine.

The four papers in more detail

1. In a study published in *Nature Biotechnology* and coordinated by CeMM, 18 research groups from three continents compared all promising methods for analyzing DNA methylation in the clinic. This multicenter benchmarking study identifies the most accurate methods and shows that epigenetic tests based on DNA methylation are a mature technology ready for broad clinical use.
<http://doi.org/10.1038/nbt.3605>
 2. Also in *Nature Biotechnology*, the UCL team presents a computational validation of genome-wide DNA methylation sequencing technology, confirming its practical use for identifying DNA methylation differences between samples and disease subtypes.
<http://doi.org/10.1038/nbt.3524>
 3. The UCL team further extend their analysis in a paper published in *Nature Communications*, where they present new bioinformatic methods for discovering disease-specific DNA methylation patterns from cost-effective low-coverage DNA methylation sequencing data.
<http://doi.org/10.1038/ncomms11306>
 4. Finally, a *Nature Communications* paper by CeMM researchers – in collaboration with the clinicians at the University of Southampton and the Royal Bournemouth Hospital – demonstrates the utility of chromatin mapping for identifying disease subtypes and predicting prognosis in chronic lymphocytic leukemia. This study highlights the clinical utility of epigenetic biomarkers especially for diseases with widespread heterogeneity between individual patients.
<http://doi.org/10.1038/ncomms11938>
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Quotes

Christoph Bock (Principal Investigator at CeMM): “Epigenetic tests have a key role to play for making precision medicine a clinical reality. Epigenetics captures part of each cell’s individual history, and it can predict how cancer cells will react to drug treatment. This can be very useful for personalized therapy.”

Stephan Beck (Professor at UCL): “This exciting new technology will advance our ability to understand phenotypic plasticity in health and disease, an important aspect of cancer research.”

Giulio Superti-Furga (Scientific Director of CeMM): “It is part of CeMM’s mission to provide innovative platforms for precision medicine and to make them ready for biomedically important applications. We are proud of the work that Christoph Bock has done together with Stephan Beck and members of the BLUEPRINT consortium. It shows that epigenetics bridges genomics and disease.”

CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences is an interdisciplinary research institute committed to advancing the understanding of human diseases through basic and biomedical research. Located at the center of the Medical University of Vienna’s campus, CeMM and fosters a highly collaborative and interactive research mindset. Focusing on medically relevant questions, CeMM researchers

concentrate on human biology and diseases like cancer and inflammation/immune disorders. In support of scientific pursuits and medical needs, CeMM provides access to cutting-edge technologies and has established a strategic interest in personalized medicine. Since 2005, Giulio Superti-Furga is the Scientific Director of CeMM.

<http://www.cemm.oeaw.ac.at/>

Christoph Bock is a Principal Investigator at CeMM. Trained as a bioinformatician, he leads a team that integrates biology, medicine, and computer science – working on a vision of precision medicine that is driven by large datasets and a deep understanding of disease mechanisms. He is also a guest professor at the Medical University of Vienna's Department for Laboratory Medicine, and he coordinates the genome sequencing activities of CeMM and the Medical University of Vienna. At CeMM, he co-initiated and leads Genom Austria, the Austrian contribution to the International Network of Personal Genome Projects, and he is a principal investigator in BLUEPRINT / International Human Epigenome Consortium).

<http://epigenomics.cemm.oeaw.ac.at/>

BLUEPRINT is a large-scale research project receiving 30 million Euro funding from the European Union. 39 leading universities, research institutes and industry entrepreneurs participate in what is one of the two first so-called high impact research initiatives to receive funding from the European Union. The BLUEPRINT consortium has been formed with the aim to further the understanding of how genes are activated or repressed in both healthy and diseased human cells. BLUEPRINT focuses on hematopoietic cells from healthy individuals and on their malignant leukaemic counterparts. It aims to generate at least 100 reference epigenomes and study them to advance and exploit knowledge of the underlying biological processes and mechanisms in health and disease.

<http://www.blueprint-epigenome.eu/>

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