

## **BLUEPRINT: A BLUEPRINT of haematopoietic epigenomes**

### **Description**

In response to the call for a high impact initiative on the human epigenome, the BLUEPRINT Consortium was formed with the aim of generating at least 100 reference epigenomes and studying them to advance and exploit knowledge of the underlying biological processes and mechanisms in health and disease. BLUEPRINT focused on distinct types of haematopoietic cells from healthy individuals and on their malignant leukaemic counterparts.

Reference epigenomes were generated by state-of-the-art technologies from highly purified cells for a comprehensive set of epigenetic marks in accordance with quality standards set by IHEC. This resource-generating activity was conducted at dedicated centres, complemented by confederated hypothesis-driven research into blood-based diseases, including common leukaemias and autoimmune disease (T1D), by epigenetic targets and compound identification, and by discovery and validation of epigenetic markers for **diagnostic use**. By focusing on 100 samples of known genetic variation BLUEPRINT completed an epigenome-wide association study, maximizing the biomedical relevance of the reference epigenomes.

Key to the success of BLUEPRINT has been the integration with other data sources (i.e. ICGC, 1000 genomes and ENCODE), comprehensive bioinformatic analysis, and user-friendly dissemination to the wider scientific community. The involvement of innovative companies also assisted in energizing epigenomic research in the private sector by creating new targets for compounds and the development of smart technologies for better diagnostic tests.

BLUEPRINT outreached through a network of associated members and formed critical alliances with leading networks in genomics and epigenomics within Europe and worldwide. Through its interdisciplinarity and scientific excellence combined with its strong commitment to networking, training and communication BLUEPRINT strived to become the cornerstone of the EU contribution to IHEC.

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