

Researchers sequence human methylome at single base-pair resolution

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DNA methylation plays an important role in many processes such as animal development, X-chromosome inactivation, and carcinogenesis. Understanding the mechanisms and functions of DNA methylation and how it varies from tissue to tissue and between individuals will have profound implications for human health and disease.

A team of Chinese researchers decoded the essentially complete methylome (an inventory of all the bases that are methylated) of the <u>human genome</u> using peripheral blood mononuclear cells (PBMCs). The results will be published in the online, open access journal <u>PLoS Biology</u> next week.

The research is part of YanHuang (YH) Project, which has been launched by BGI (previous known as Beijing Genomics Institute) at Shenzhen, which aims to sequence 100 Chinese individuals in 3 years to accelerate the discovery of <u>disease genes</u> and mutations in an Asian population. The methylome was generated from the same donor whose genome was deciphered in the YH project. The methylome was examined at 20 distinct features including regulatory, protein-coding, non-coding, and repeat sequences.

The integration of the data with the previously determined genome sequence of the same Asian individual allowed the identification of allele-specific methylation (ASM) differences between the methylomes of the genomes inherited from either parent. This revealed that ASM was highly correlated with allele-specific gene expression (ASE) which



indicated that parental gene imprinting (that is the favored expression of the genes inherited from one parent) may be more common than previously thought.

The research not only provides a comprehensive resource for future epigenomic research but also demonstrates a paradigm for epigenetic studies through new sequencing technology. The PBMC methylome data has been deposited to NCBI (<u>http://www.ncbi.nlm.nih.gov/Traces/sra/</u>, accession number: SRA008544). It is expected to form a lasting resource as part of the International Human Epigenome Project.

More information: Li Y, Zhu J, Tian G, Li N, Li Q, et al. (2010) The DNA Methylome of Human Peripheral Blood Mononuclear Cells. PLoS Biol 8(11): e1000533. <u>doi:10.1371/journal.pbio.1000533</u>

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