

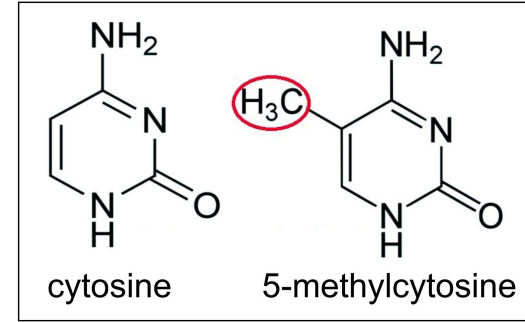
# **Illumina EPIC Array**

(850k Array)

By : Group 3

# DNA methylation: a historical perspective

**1925:** Discovery of 5-methylcytosine in bacteria.



**Subsequent Decades (1940s-1960s):** Slow recognition of its biological importance.

**Expansion of Research (1970s-1980s):** Recognition of widespread presence across life forms.

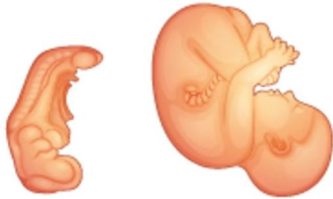
**Modern Era (1990s-2000s):** Detailed understanding of DNA methylation mechanisms and functions.

**Current Research:** Ongoing exploration of DNA methylation patterns and their precise roles.

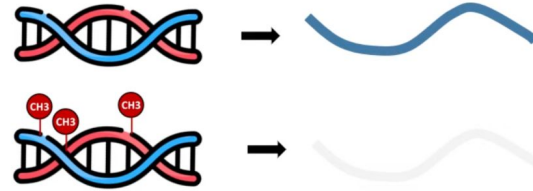
- Cancer

# What is DNA methylation? Importance?

embryonic development



genomic imprinting



X-chromosome inactivation



preservation of chromosome stability.



# EPIC Array

Advanced technology for **DNA methylation** profiling

Interrogation of methylation patterns at the **genome-wide level**

**Extensive Coverage:** Targets more than 935,000 CpG sites, offering comprehensive coverage of the human methylome

**High-Throughput Capability:** Enables large cohort screenings at a lower total cost per sample

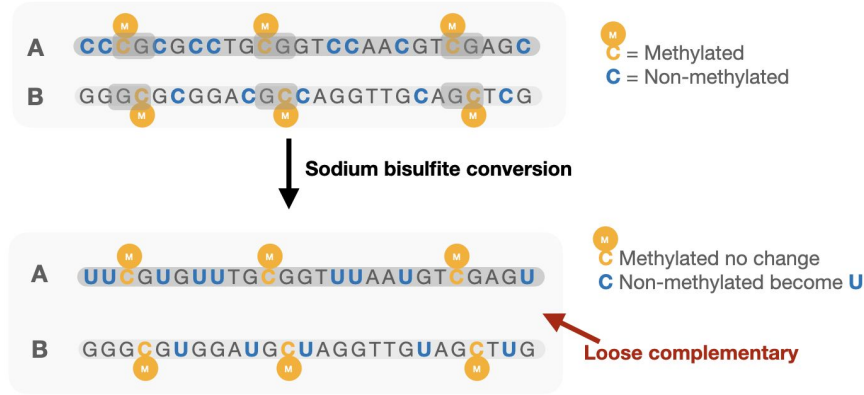
## Comparison with other assays

|                  | <b>27K</b>       | <b>450K</b>               | <b>EPIC</b>   |
|------------------|------------------|---------------------------|---|
| <b>CpG sites</b> | 27,000           | 450,000                   | 850,000   |
| <b>Regions</b>   | Promoter regions | Gene-centric sites        | Enhancer regions,<br>gene bodies, and<br>intergenic regions |
| <b>Assay</b>     | Infinium I       | Infinium I<br>Infinium II | Infinium I<br>Infinium II                                   |

More than 90 % of the sites on the HM450, plus more than 350,000 CpGs at regions identified as potential enhancers

# Sodium Bisulfite Conversion

C non methylated  $\longrightarrow$  T  
C methylated  $\longrightarrow$  C



PCR amplification STRAND A

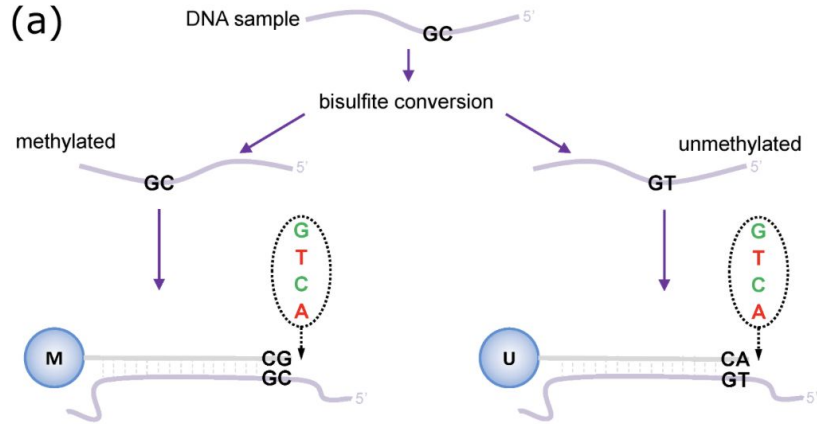
PCR amplification STRAND B



# Infinium Chemistry

C non methylated  $\longrightarrow$  T  
C methylated  $\longrightarrow$  C

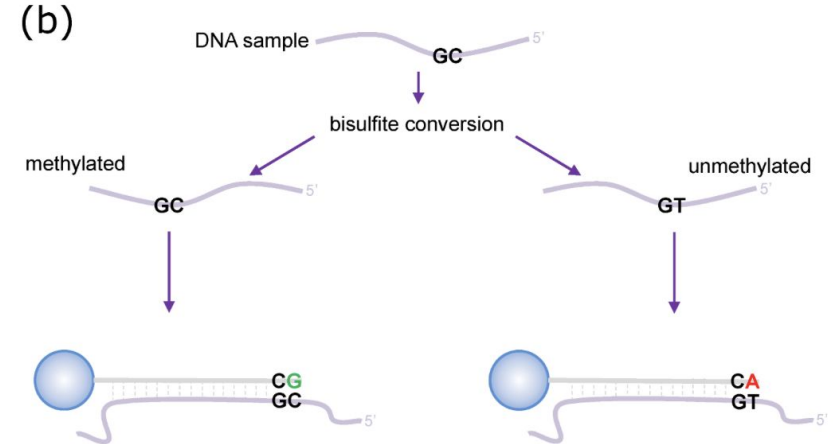
## Infinium I



Two beads:

- Methylated (M) bead: CG in probe binds to GC in DNA
- Unmethylated (U) bead: CA in probe binds to GT in DNA

## Infinium II



One bead:

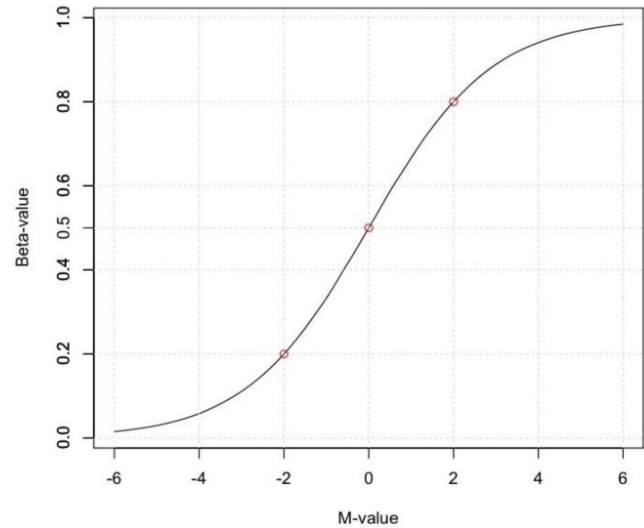
- Both signals are measured by the same bead
- Probe designed with R bases that hybridises both T and C bases.

# Level of methylation at a locus

$$\beta = M / (M + U)$$

Every copy of CpG is completely **methy**lated

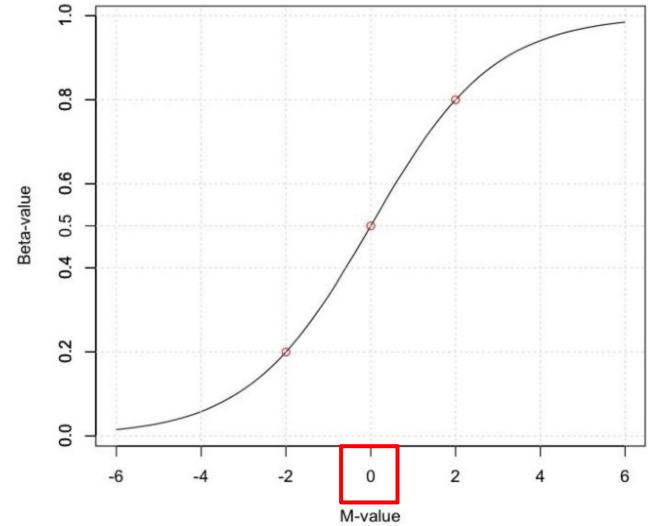
All CpG site copies are completely **unmethy**lated





# Level of methylation at a locus

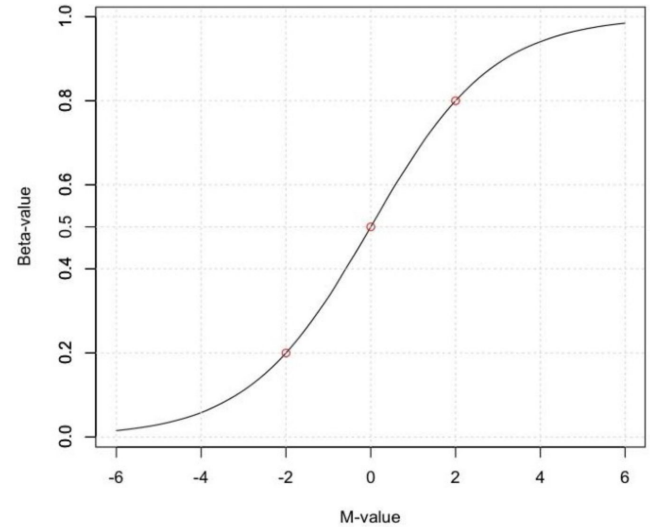
$$Mvalue = \log_2(M/U)$$



Similar intensity between  
methylated and  
unmethylated probes  
(CpG site is  
half-methylated)

# Level of methylation at a locus

$$Mvalue = \log_2(M/U)$$



More molecules are  
unmethylated

More molecules are  
methylated