Phase 2
Short summary

DNA Methylation and its basic function

Methylation and demethylation occur on CpG sites. It is a defined as adding a methyl group to a cytosine(C) nucleotide. The methyl group has its function.

CpG site is a dinucleotide part of the DNA strand where the C nucleotide is followed by the G nucleotide in the 5'->3' direction.

When grand amounts of CpG sites are located near one another, that region is called CpG island.

One of the main roles of methylation is to repress the expression of some genetic elemetnts.

CpG island shores are more related to methylation than CpG islands.

CpG islands are more related to being promoters for transcription.

Methylation enzymes classes are the following: writers, erasers, readers.

Writers: the DNMT

DNMT1

DNMT3a, DNMT3b

DNMT3L

Transcription factors guard CpG islands from binding methylation.

Alternations in methylation can lead to mental diseases and disorders.

An introduction to WGBS

DNA that has been treated with bisulfite retains only methylated cytosines. Therefore we can observe where are all the locations of the C nucleotides with 5-methylcytosine on the DNA strand.

Methylated-specific primers are used to detect a methylated region (Methylation-specific PCR).

There are limitations to bisulfite treatment like the following:

- 5-methylcytosine and 5-hydroxymethylcytosine are both converted to C after the treatment
- incomplete conversion
- degradation of DNA.

Oxidative bisulfite sequencing can be used as a method to discriminate betwwen 5-methylcytosine and 5-hydroxymethylcytosine.

Aberrant methylation patterns are well characterized in many cancers.

DNA methylation-calling tools for Oxford Nanopore sequencing: a survey and human epigenome-wide evaluation

Electric current patterns are called squiggles.

Some new DNA methylation calling tools (12) for nanopore sequencing are presented.

Step 1: Basecalling and quality control
-translating raw signal dana into nucleotide sequences (Guppy)
-data visualization and processing (NanoPack)

Step2: Genome assembly and polishing
-aligning basecalled reads (minimap2)

Step3: Methylation calling and evaluation
-using methylation calling tools
-detecting methylation patterns
-evaluation of results by various criteria

Questions:

I have just some basic questions regarding chemistry included in this topic. It is my first time to hear about many of the mentioned molecules so it was a bit challenging to understand how this whole process works.