Motivation: epigenetics & CpG islands

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Picking up signals

So far, we’ve focused on how to stitch fragments of evidence into longer units, i.e. genomes.

Equipped with genomes, we can ask more questions:

Where are the genes?
Where/what is the *functional* DNA?
What’s different about the DNA in different tissues?
In what abundance do we find various molecules?
What differences exist between individuals?
Through many experiments, we know much more about the genome than just its DNA sequence:

- Experimentally observed products, e.g. messenger RNAs
- Epigenetic marks
- Sequence conservation among related species
- Sites that vary across individuals

40 K nt region of chromosome 17

http://genome.ucsc.edu/cgi-bin/hgTracks

[Image: A graph showing various tracks and analyses, such as RNA expression levels and DNA conservation patterns, across different locations in the genome.]

[Logo: Johns Hopkins Whiting School of Engineering]
CpG Islands

A signal we can discern from genome sequence alone: CpG islands

Dinucleotide “CG” (AKA “CpG”) is special because the C can possibly have a methyl group attached

Proteins involved in gene expression can be repelled or attracted by the methyl group

Unmethylated or Methylated
CpG Islands

*CpG island:* part of the genome where CG occurs particularly frequently

CpG islands usually regulate expression of nearby genes

Cells from different tissues have different patterns of CpG methylation, in turn giving them different gene expression profiles

Key *epigenetic* phenomenon
Background: Epigenetics

http://en.wikipedia.org/wiki/File:Stem_cells_diagram.png
Background: Epigenetics
Background: Epigenetics

Study of how characteristics are inherited across generations without changes to the DNA sequence itself

How does a heart cell know it’s a heart cell?

How does a calico cat get its splotches?

Epigenetic changes are important in various diseases: Fragile X, Rett, and Angelman syndromes, cancer

http://en.wikipedia.org/wiki/Calico_cat