Investigating the relevance of blood DNA methylation signals for disease management in CJD.

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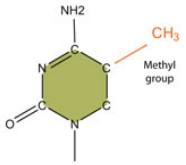
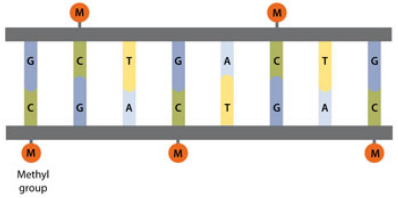
DNA methylation

- What it is — modification of the DNA
  epi: “on top of”
- Where it is — on Cytosine
- Why we care — changes gene expression
  does not modify the code
  differentiation and specificity
  allow response to environment





DNA methylation

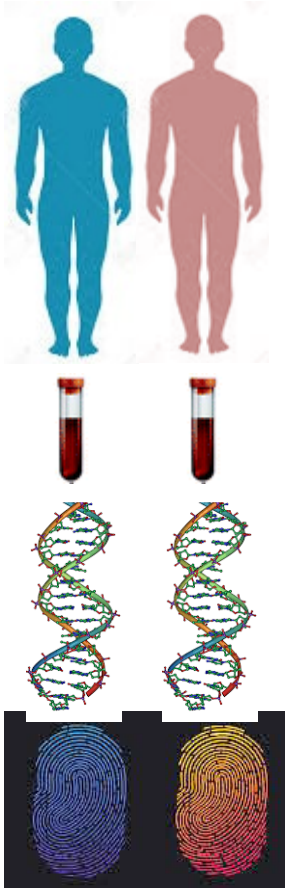
- Role regulate gene expression
- In diseases profiles are deregulated

  drug targets (treat)

  biomarkers (detect)

DNA methylation in CJD: what we have done so far

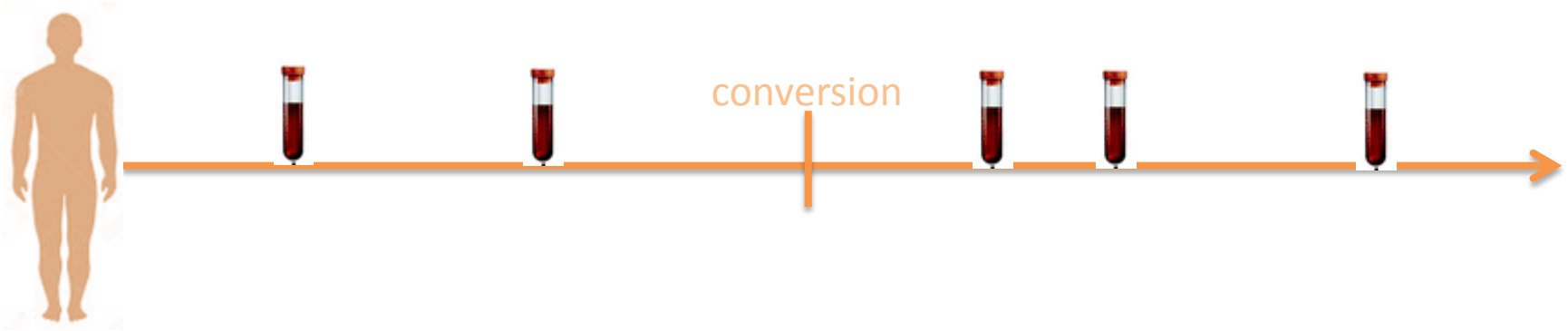
- Sporadic CJD and controls (112; 116)
- Bloods
- Profile DNA methylation
- Sites where changes are
- Fingerprints of changes



We can identify patients with sCJD using blood DNA methylation signatures

DNA methylation in inherited CJD: what we proposed to do

- hundreds of inherited cases
- 8 converters



- Q1: is the fingerprint that we detected in sCJD also present in inherited patients?
- Q2: what are the differences in DNA methylation landscapes before/after conversion;
- Q3: can we predict conversion based on DNA methylation profiles?

DNA methylation in inherited CJD: expected outcomes

- ✓ Improve understanding of mechanisms involved in inherited prion diseases
- ✓ Provide potential basis for new diagnosis tools
- ✓ Potential for predicting conversion age/ factors

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