

A pseudoSTORY

What do we know so far?

What do we want to find out?

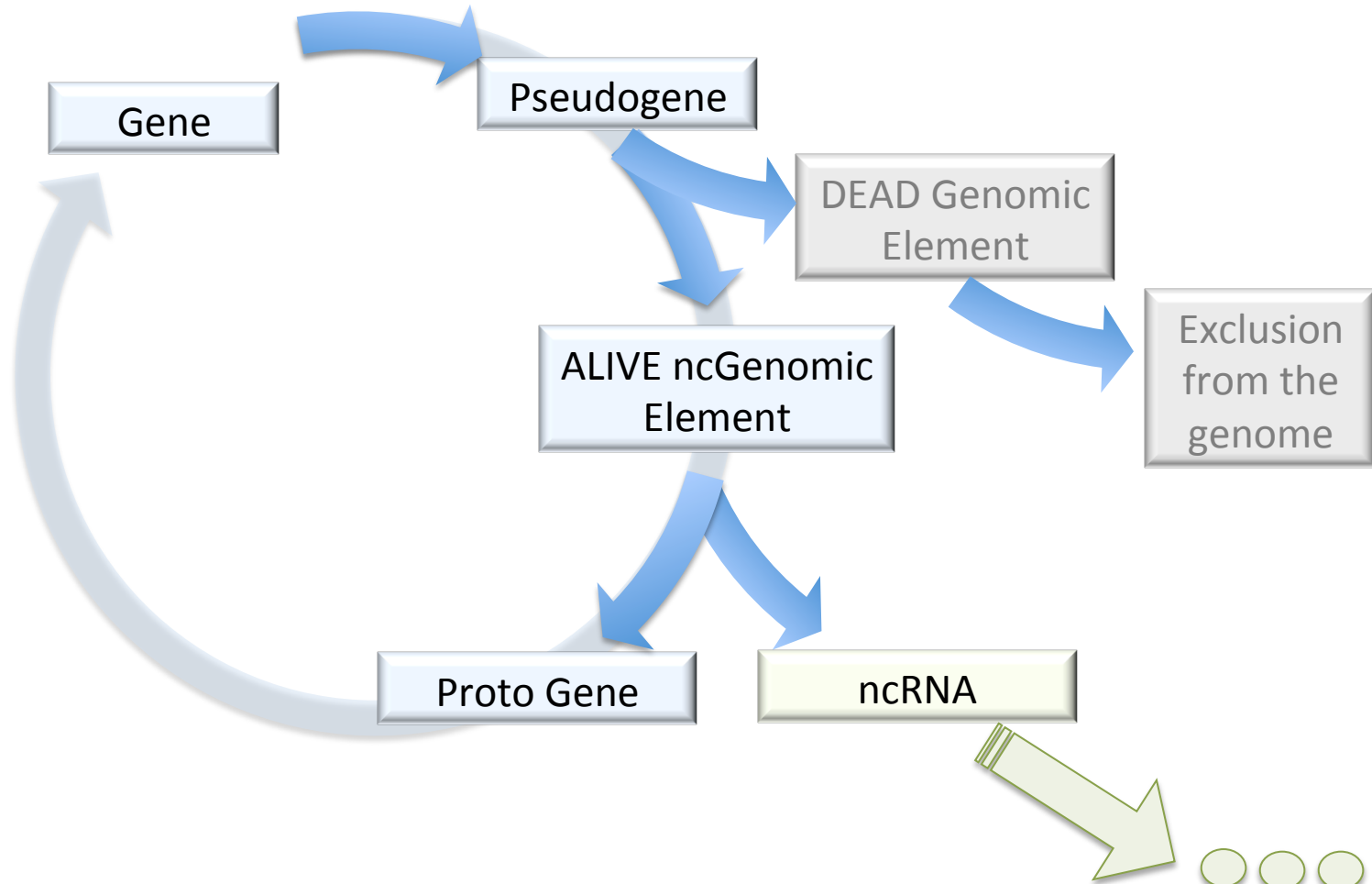
What can we learn from comparing different organisms?

Are pseudogenes related to diseases?

Facts we know!

- ① Annotation:
 - Human, Worm, Fly, Mouse, Chimp, Gorilla
- ② Activity:
 - Transcription
 - Activity features (Chromatin, TFs, Pol2) -> pseudogenes with various levels of activity
 - Translation – some pseudogene peptides are found in Worm
- ③ Biological Function:
 - Sporadic examples of functional pseudogenes: eg. PTENP1
- ④ Conservation:
 - SNPs – pseudogenes are enriched in rare alleles, however there is no clear distinction between different pseudogenes clusters (biotypes/activity groups)
 - Defining selection on individual pseudogenes is statistically challenging
- ⑤ Repeats:
 - Pseudogenes are depleted in repeats compared to random, however so are the CDS and UTR regions.
 - Processed pgenes are enriched in tRNA (repeat class) but so are other nc regions (lincRNA, miRNA)
- ⑥ Chromosomal localisation:
 - Chromosomal distribution of pgenes is organism dependent
 - Pseudogene biotype influences chromosomal localisation:

The circle of pseudoLife



What do we want to know?

① Evolution:

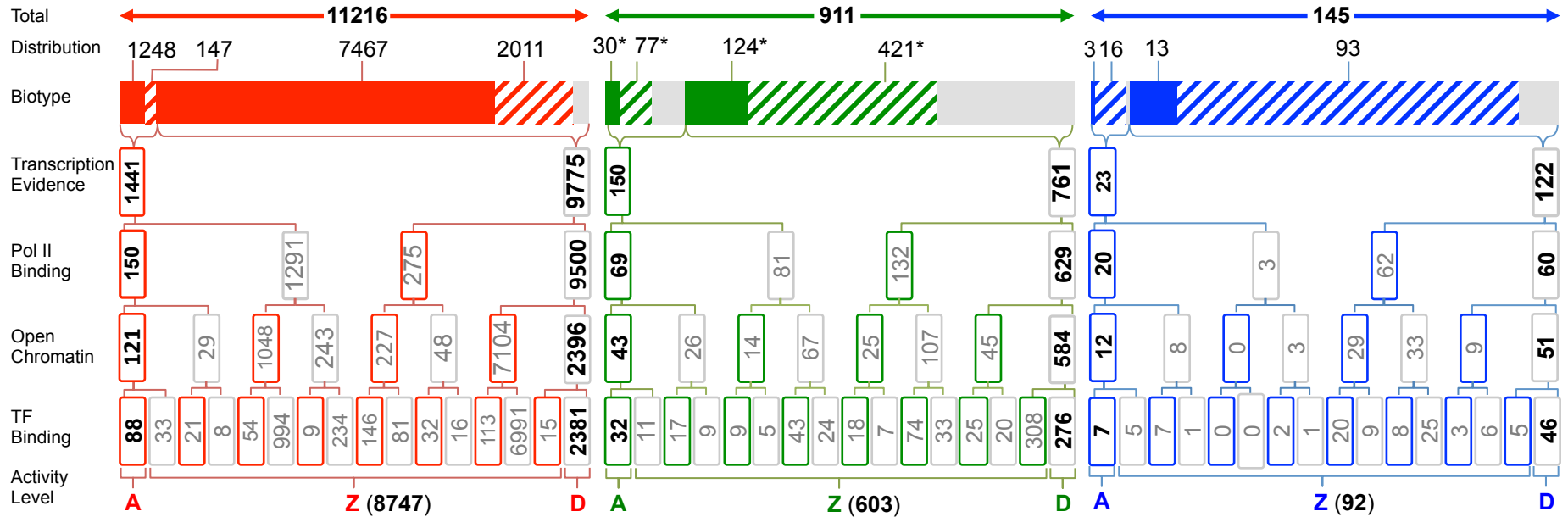
- Is the pseudogenization process organism specific?
 - Looking at gene/pseudogene ortholog pairs in different organism we can rank genes based on their organisms specificity ~ functionality ~ importance
 - ncRNA/pseudogene -> MRS
- are different organisms deprecating functional elements in different ways?
 - Which functional features are most conserved depending on age/type (eg: OC in human, TF/pol2 in worm, TF fly)
 - How fast do the upstream regions change depending of the central genomic element : gene vs pgene
- which are the shared features of the mammalian pseudogenes?

What else do we want to know?

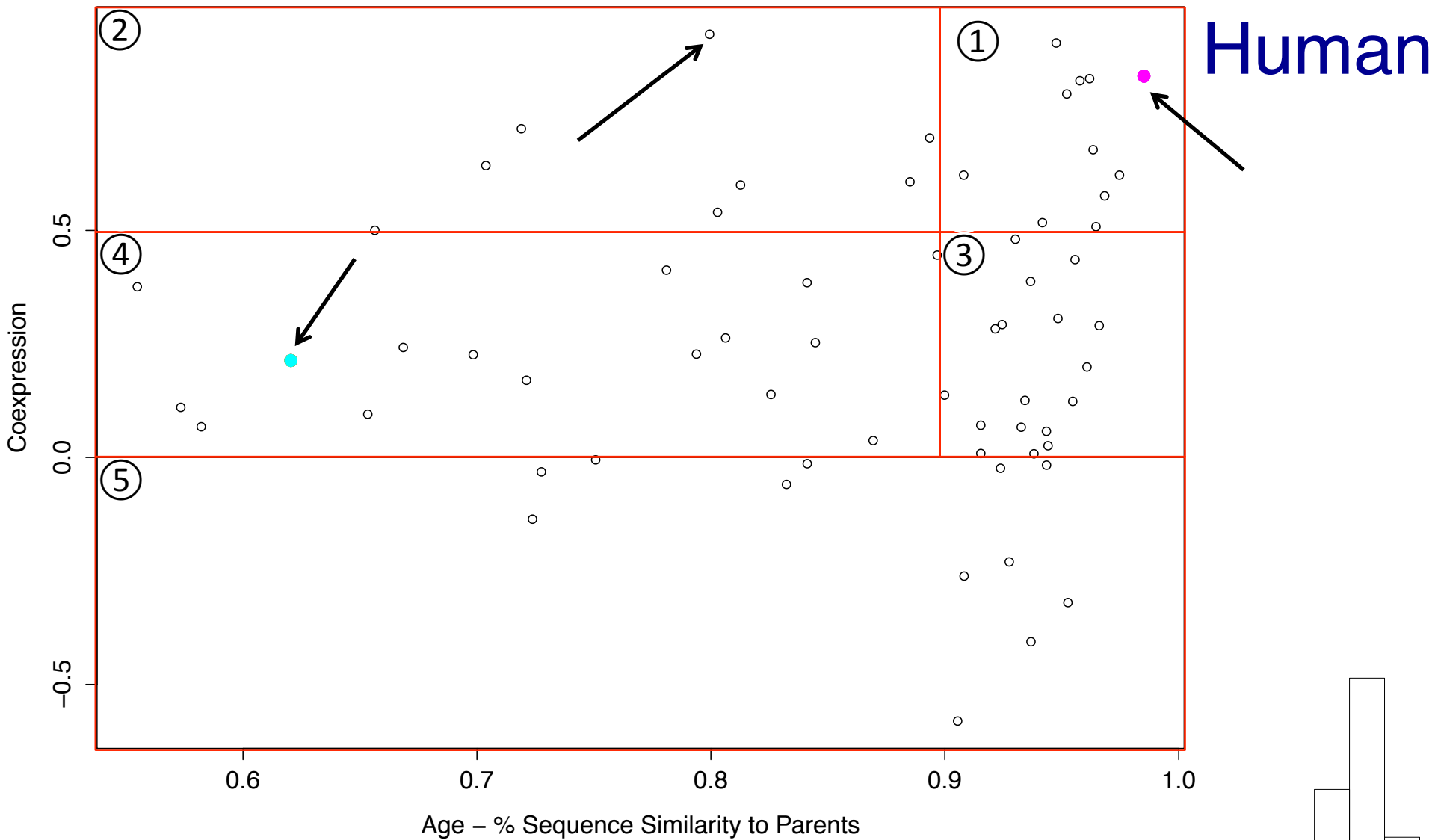
- ② Biological significance:
 - Provide a set of potentially functional pseudogenes -> set validated by PTEN-P1

- ③ Are pseudogene related to cancer /other diseases?
 - Are any of the disease genes pseudogene parents?
 - Are the cancer pseudogenes transcribed?
 - Do disease pseudogenes fulfill any regulatory roles?

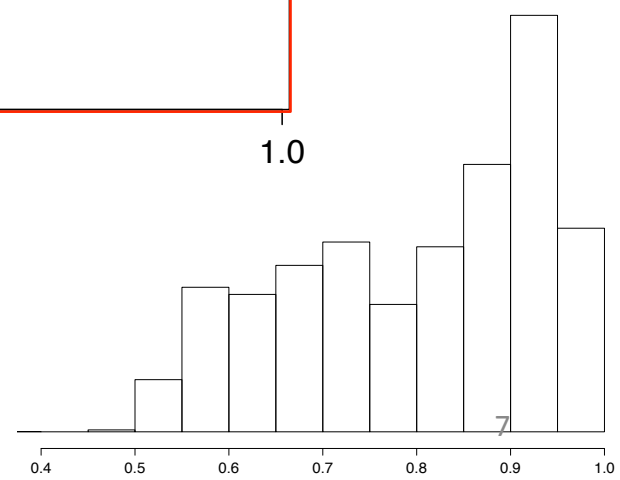
Pseudogene Activity



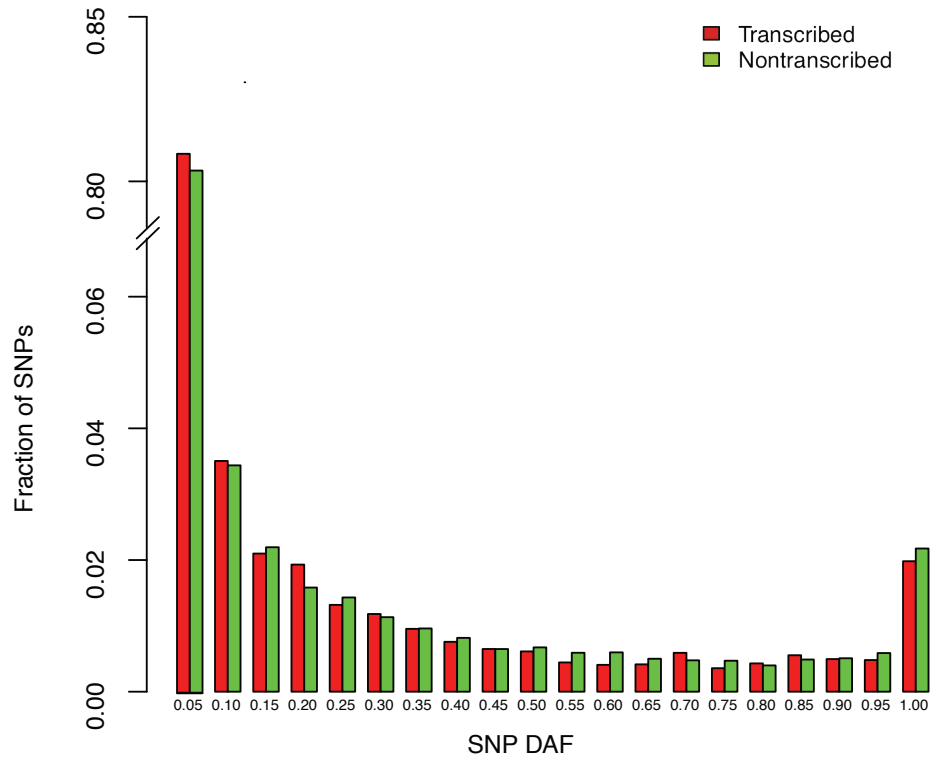
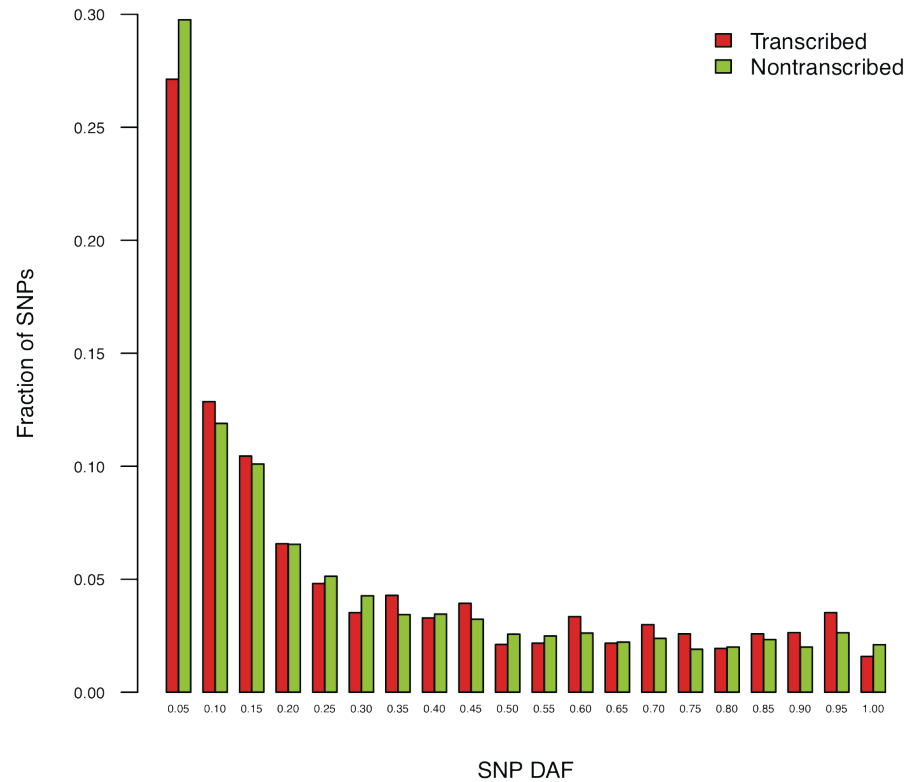
- Human
- Worm
- Fly
- Processed
- Duplicated
- Other
- Yes
- No
- A** Active
- Z** Zombie
- D** Dead



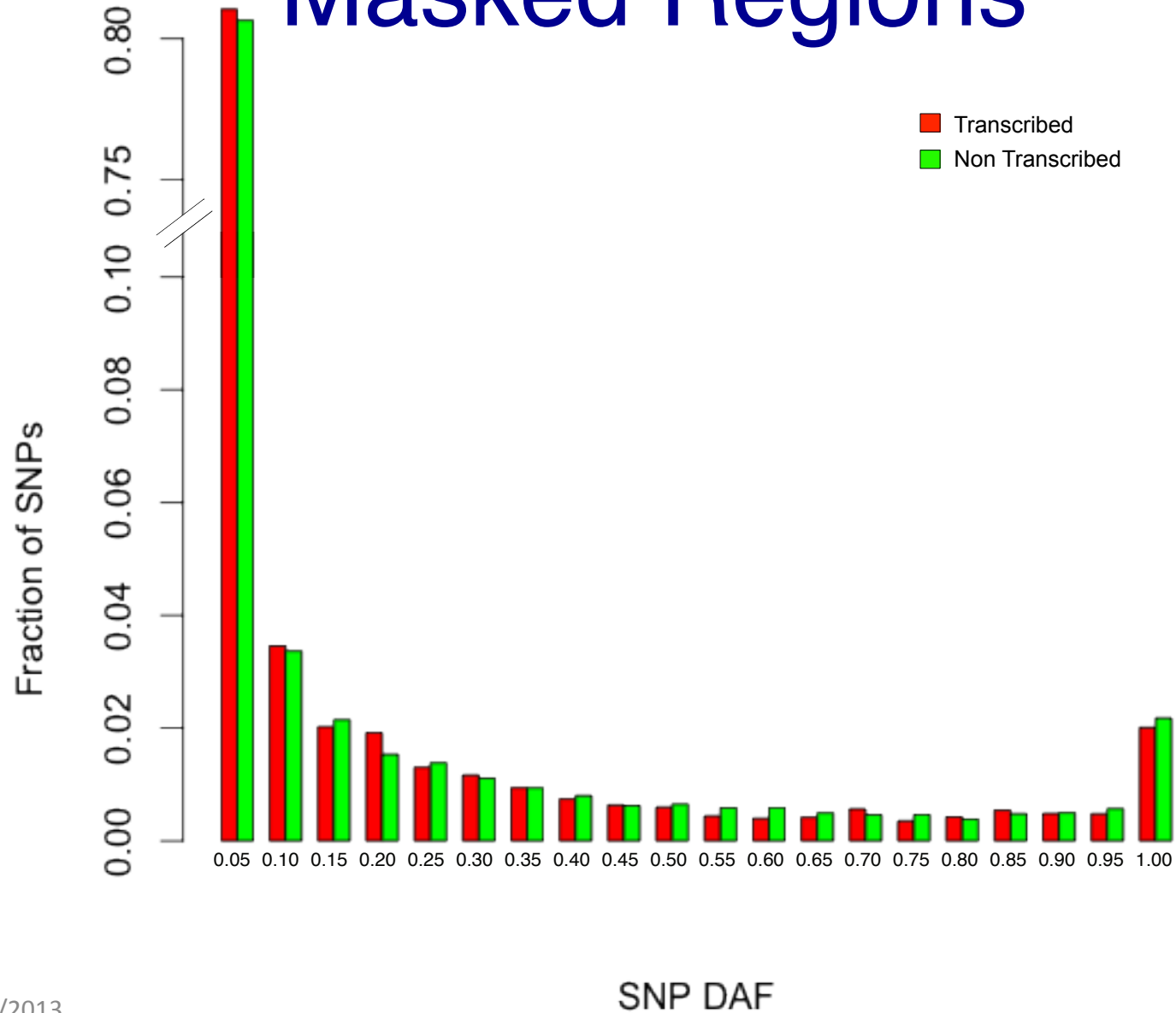
10/07/2013



Pilot vs Phase I data



Masked Regions



per Activity

