

- checkpoint #3  
methods
- DESeq2
- groups

Week 8



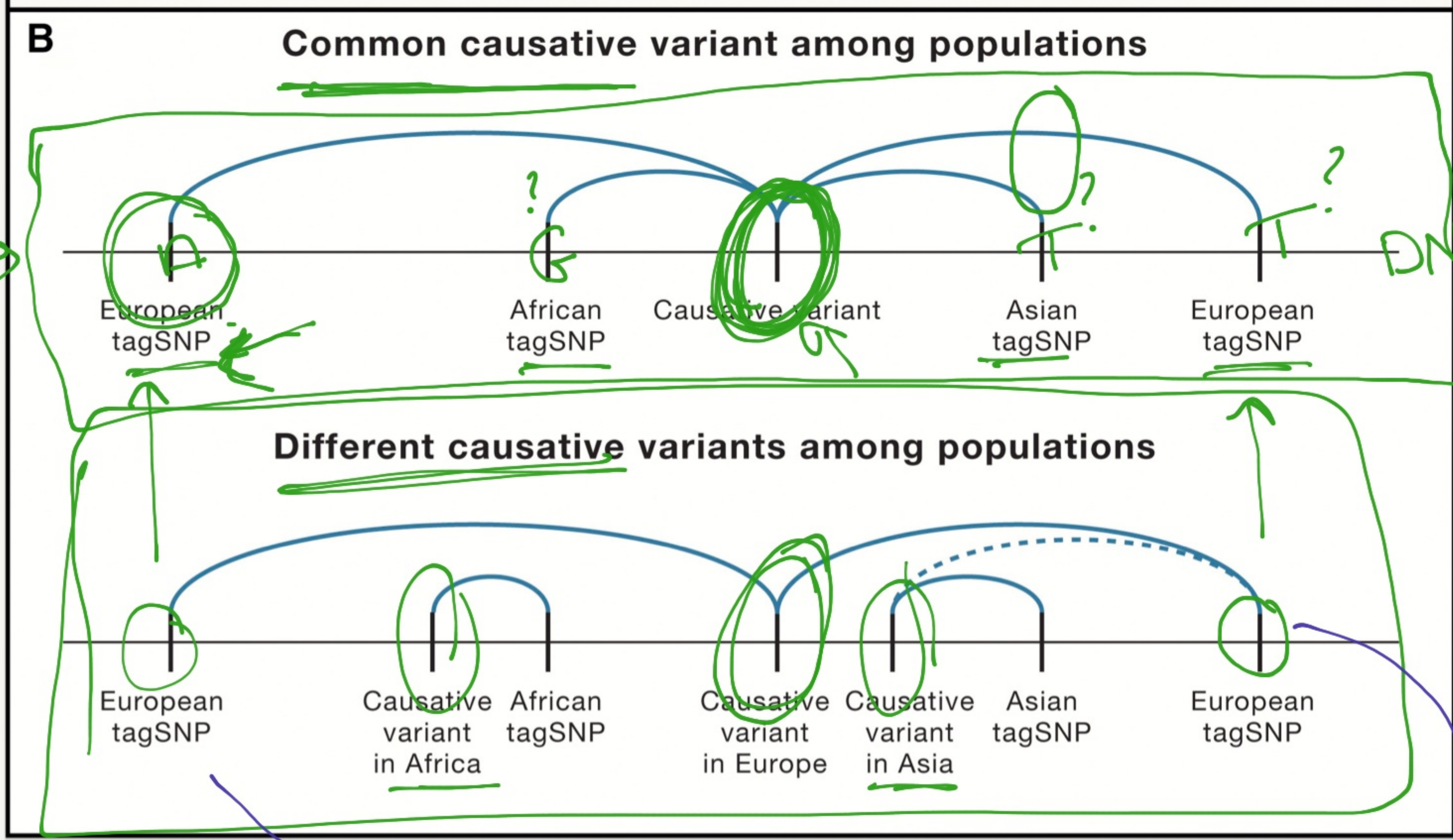
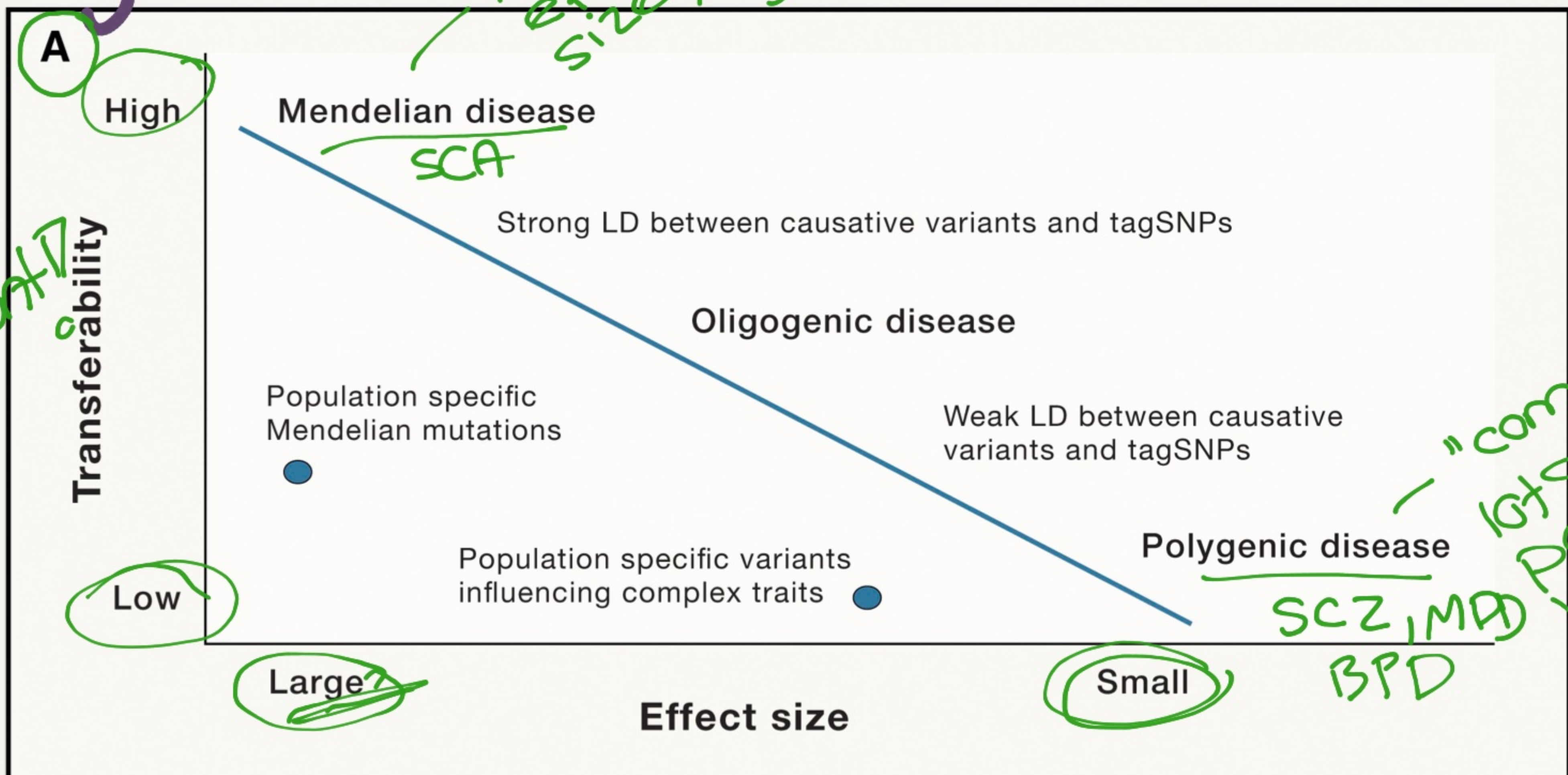
Fig1

large effect size  
single genetic mutation  
→ disease

(Punnett squares)

	A	a
A	AA	Aa
a	Aa	aa

Environment

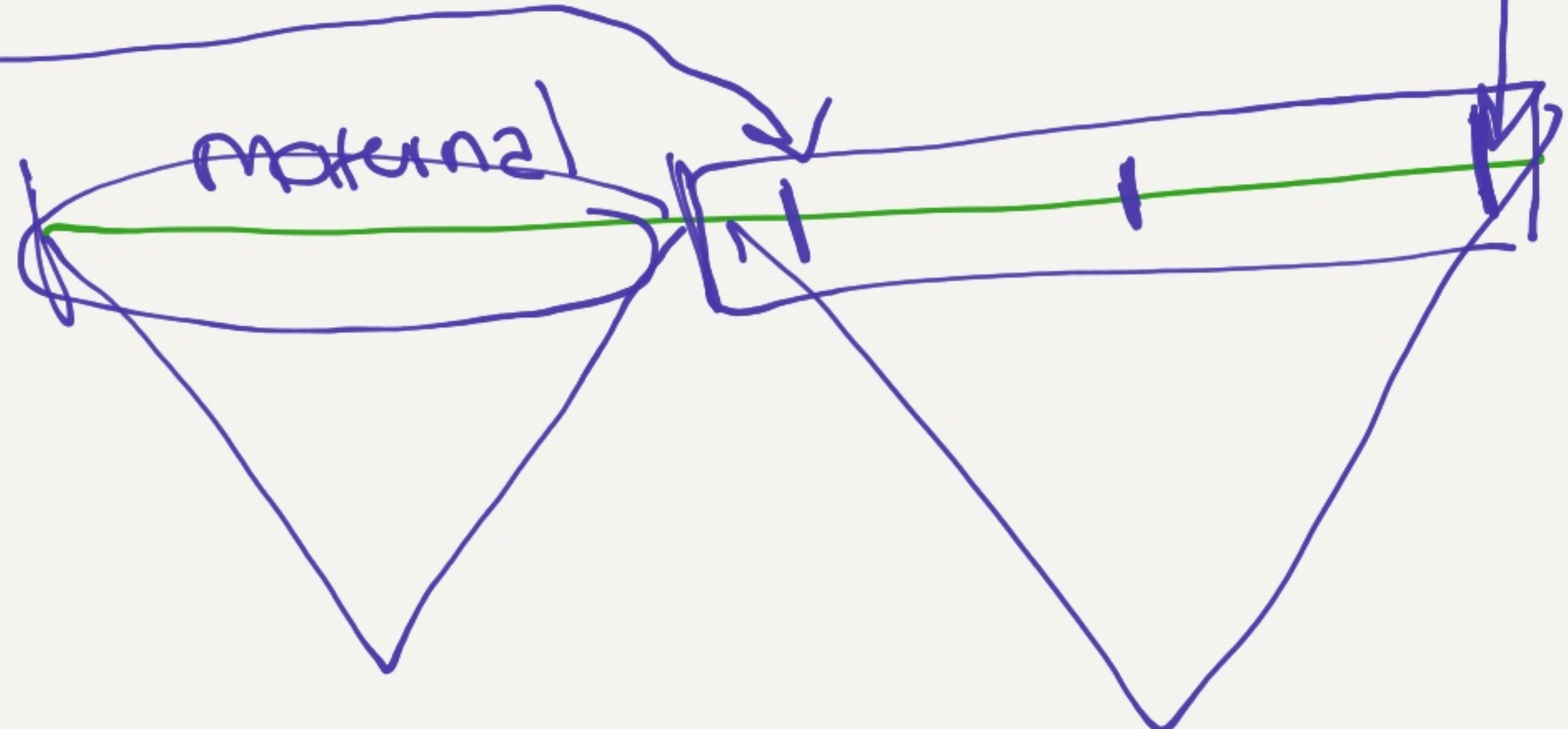


targeted seq.

DNA

LD  
Linkage  
Disequilibrium

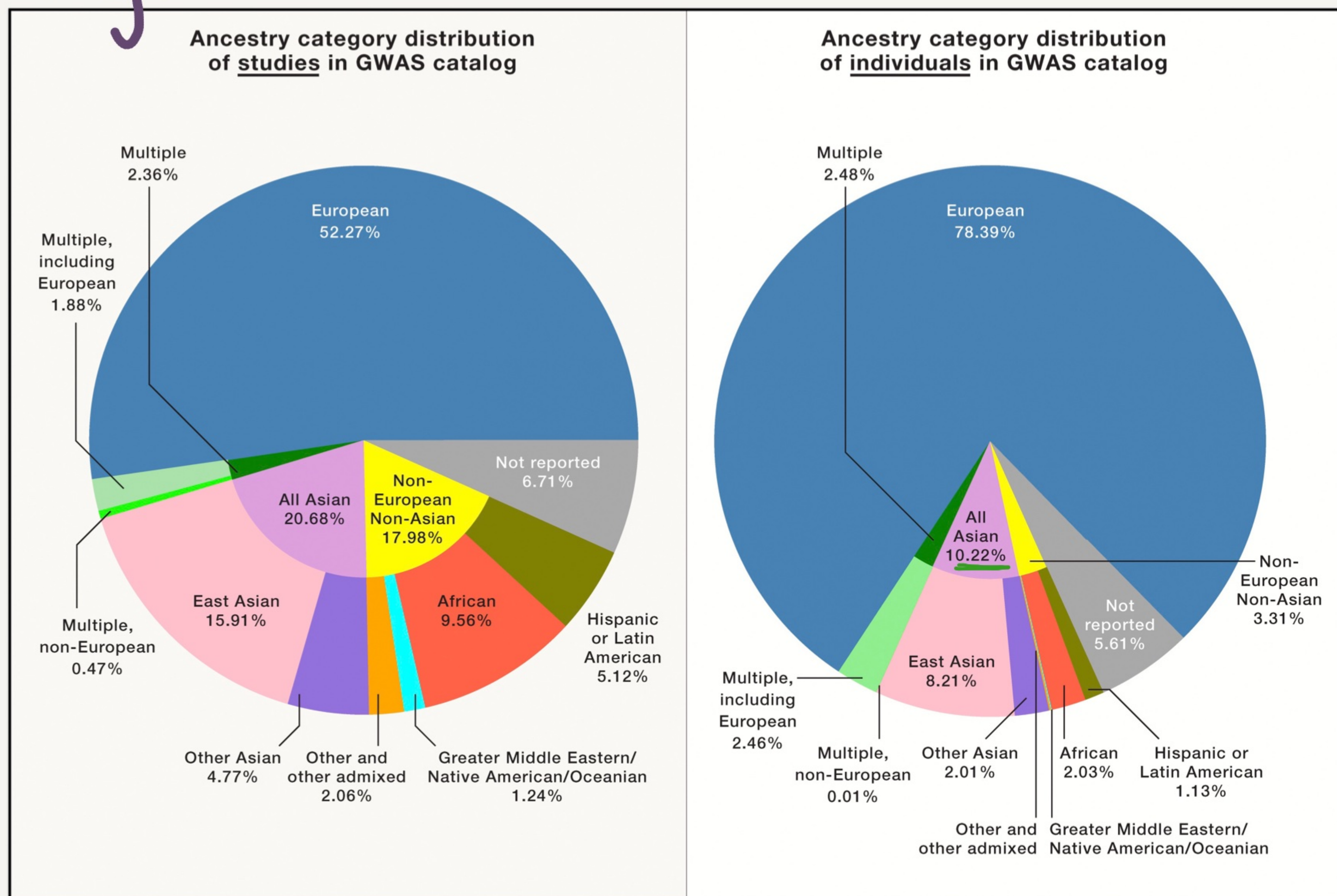
EA



chr1



Fig 2



Worldwide:  
 Asia : 59%  
 Africa : 17.2%  
 Europe : 9.6%  
 NA : 7.6%  
 SA : 5.53%

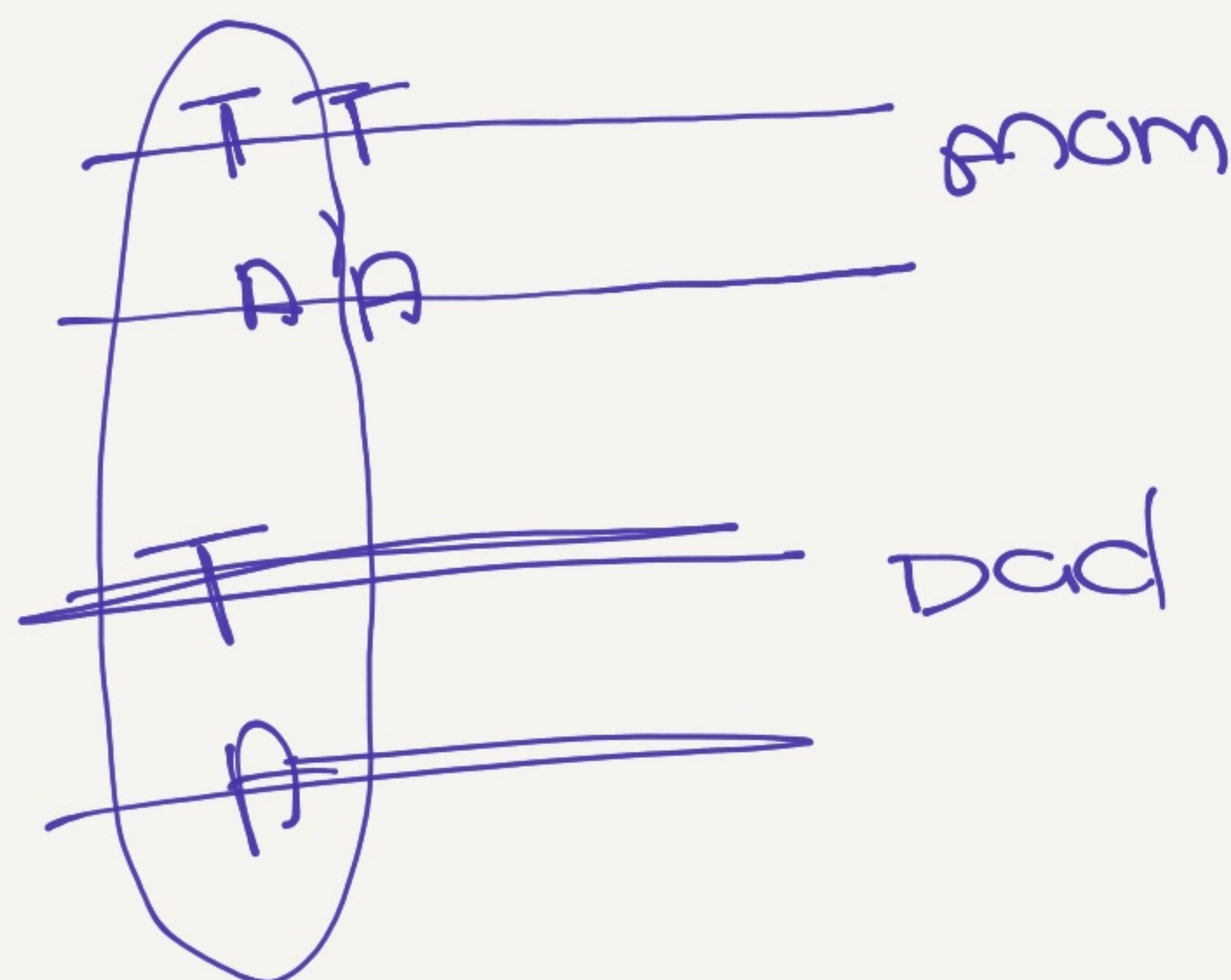


# Vocab

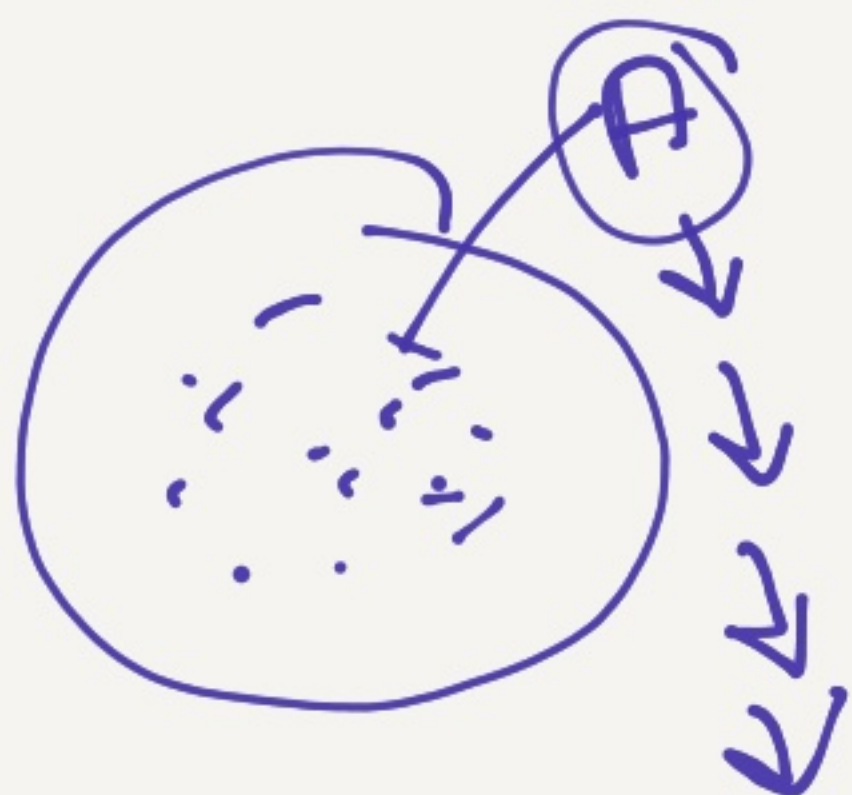
## homozygosity

AT (heterozygous)

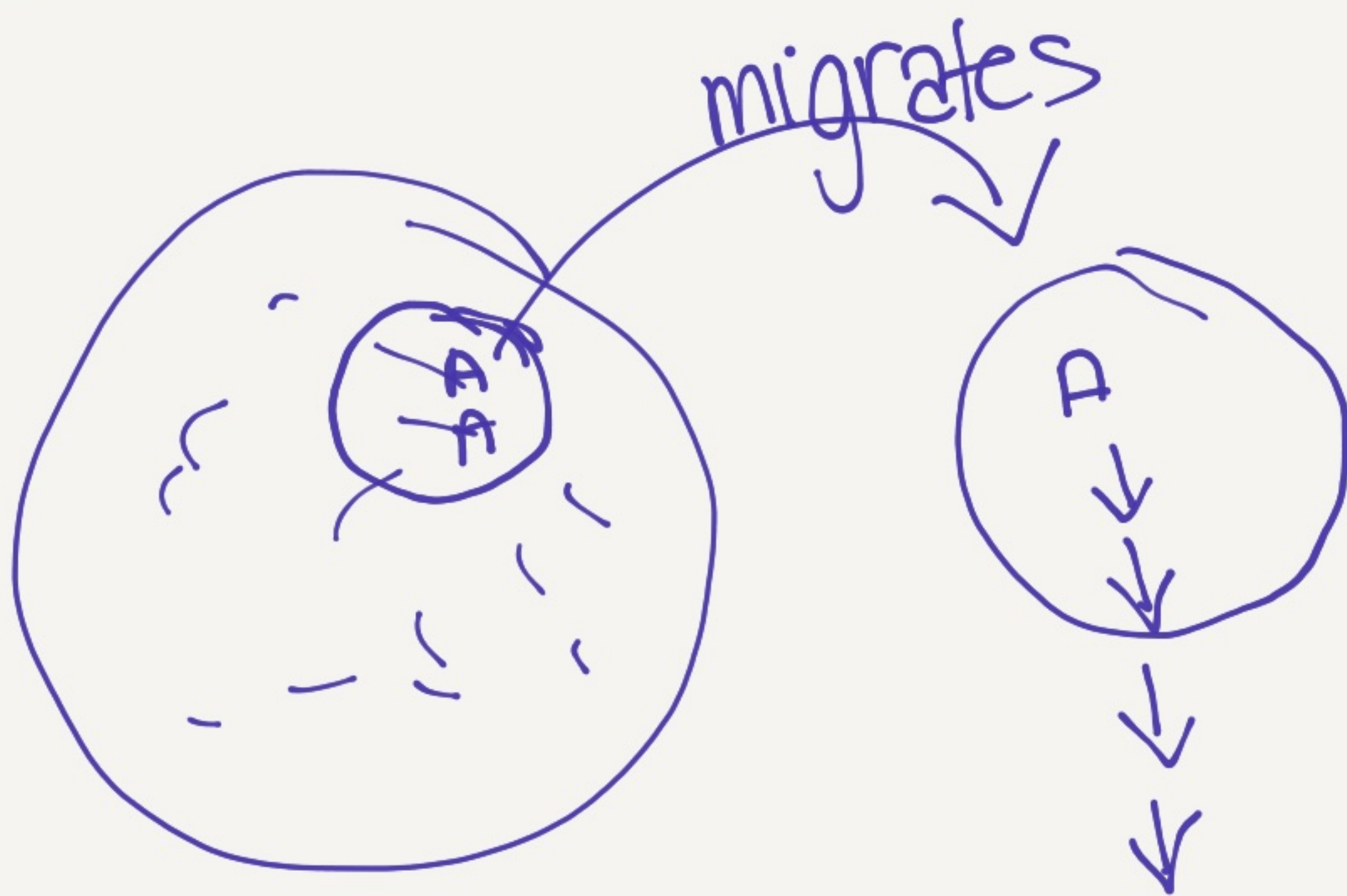
AA (homozygous)  
TT



## founder effects



## population bottlenecks



ivacaftor - medication to treat CF



# Issues in genetic research?

1. Lack of diversity in who gets studied

- ethnicity  
- gender  
sex

2. Trust / outsiders (Past diffic.)

3. Sample Access

- brain, in particular ✓
- Random mating ✓

4. Who does the studying

- who gets funding (large uni; people who know ppl @ univ.)
- what ?s they ask

5. Students, patients

(have access)

Other studies

know about it

23 and me

6. Samples are identifiable

## Solutions?

- \* revamp the system! (Identifying) planning
- \* market w/ diversity in mind  
↳ recruitment != convenience
- \* incentive? (careful of coercion)
- \* blind funding

target  
marketing