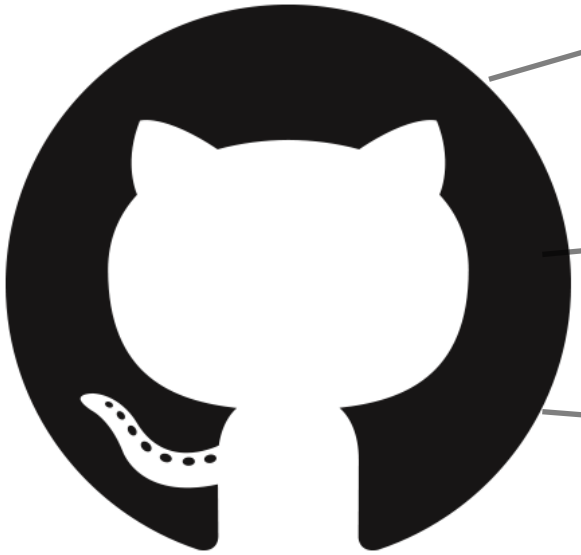
Leverage proxy-cases in your data!

Beta version available!



Identify unaffected relatives of cases in your data

Peform meta-analysis using GWAX and GWAS with shared controls

Create phenotype files for BOLT-LMM for multiple models

https://github.com/bnwolford/proxyPower

Proxy-cases are unaffected first degree relatives of cases

Affected

Unaffected first degree relative of affected

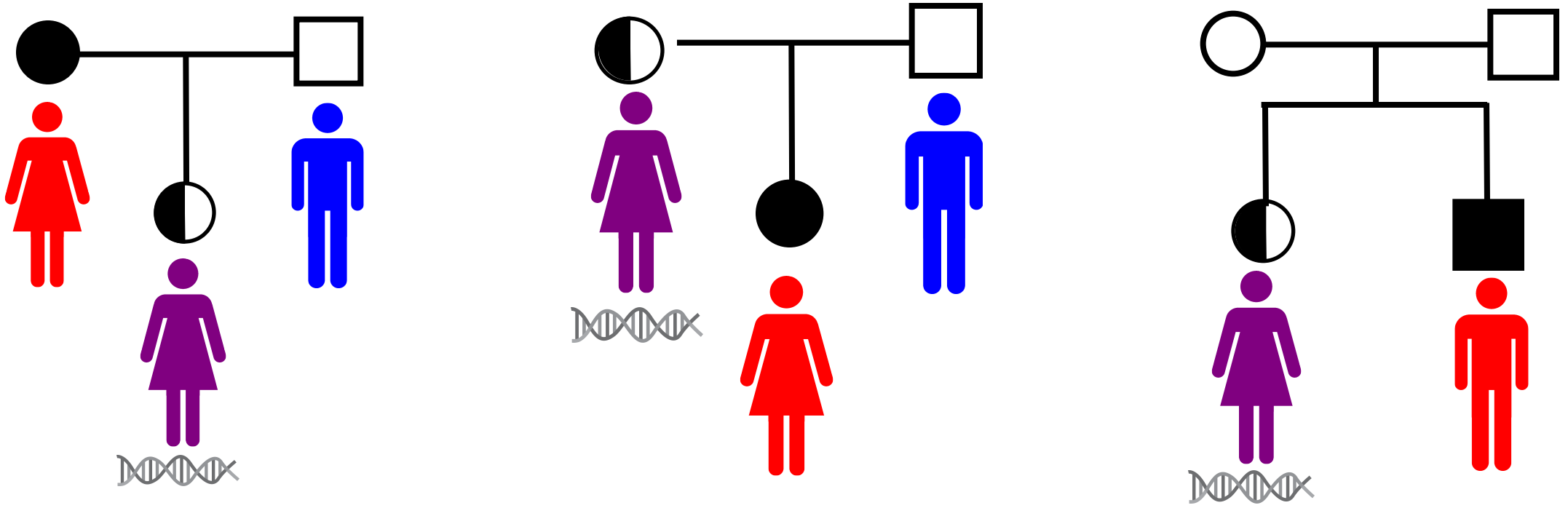
Unaffected

Case

Proxy-case

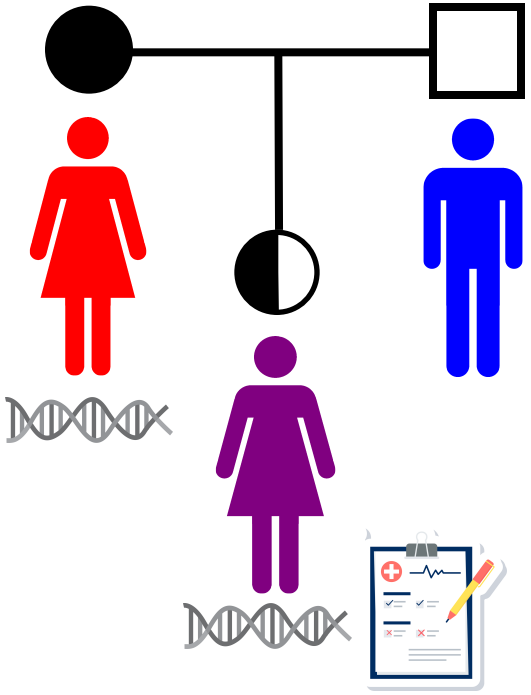
Control

Proxy-case has some combination of affected mother, father, child, sibling depending on epidemiological survey wording

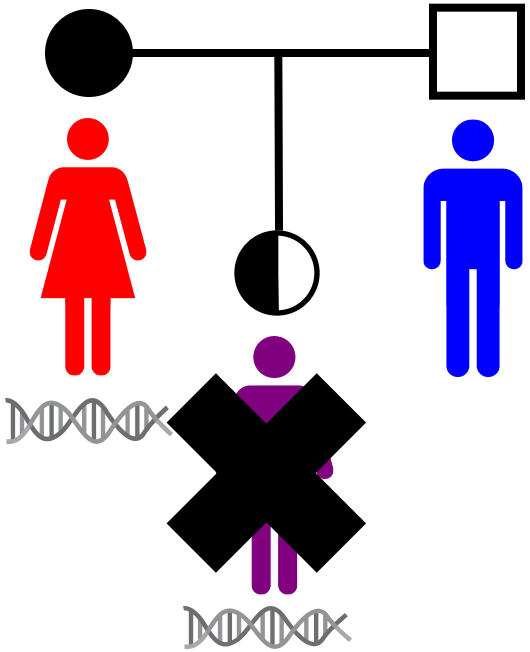


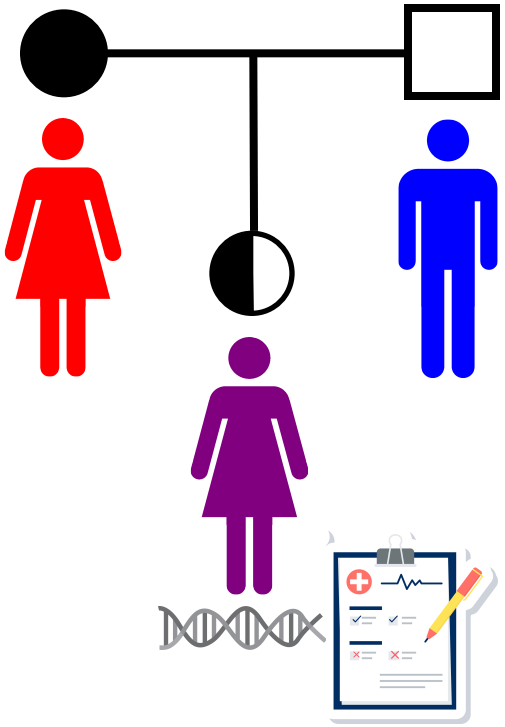
Proxy-case definition

*double counting genetic info of cases



Self report only:
Identify proxy-cases who report an affected relative on a epidemiological survey*

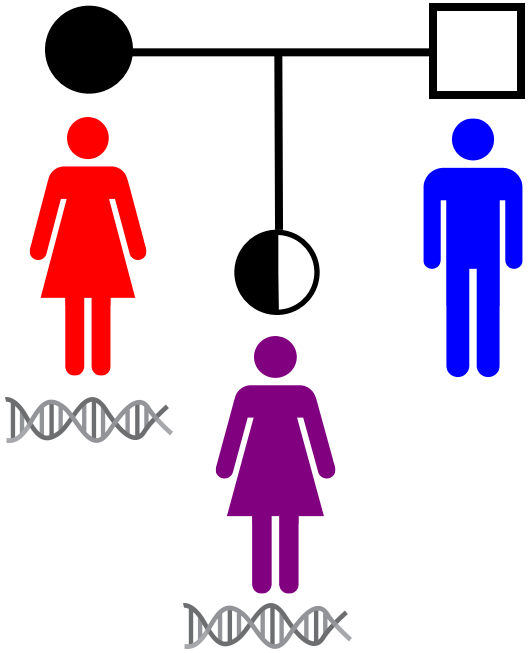




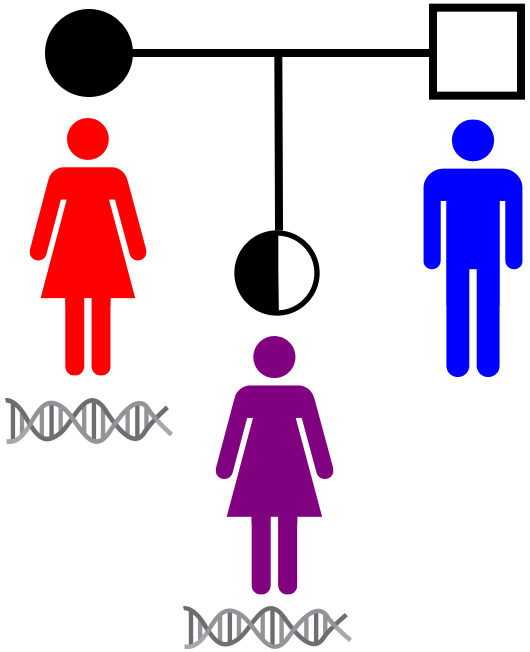
Self report minus kinship:
Exclude proxy-cases whose affected relative is in dataset, use only proxy-cases whose affected relative is not in dataset

Proxy-case definition

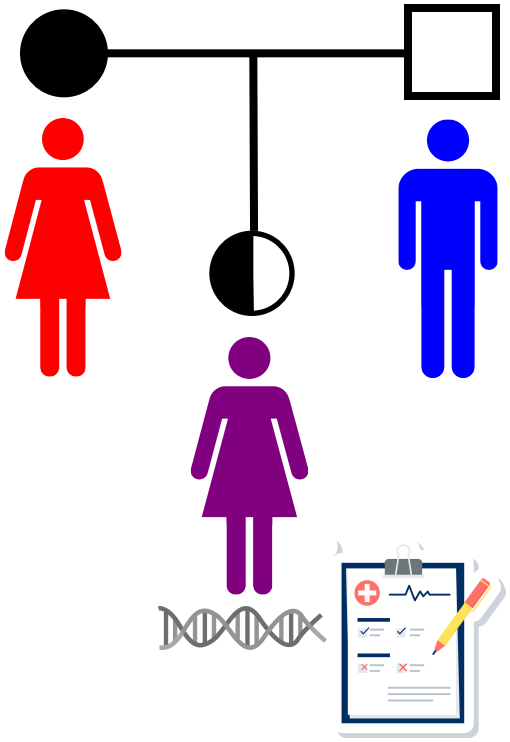
*double counting genetic info of cases



Kinship only:
Identify proxy-cases whose affected relative is in the dataset using kinship matrix

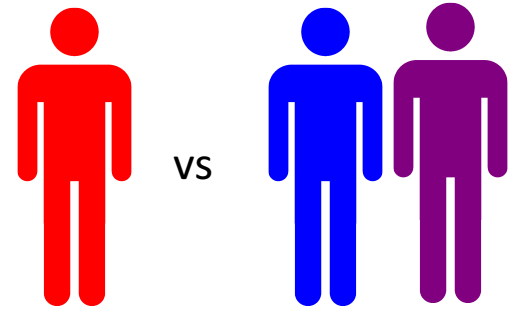


and



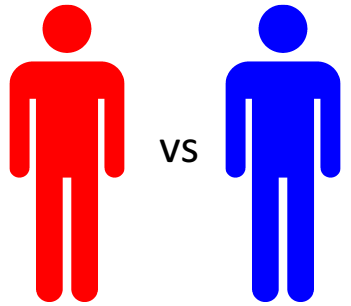
Self report plus kinship:
Identify proxy-cases whose affected relative is in dataset even if the sample did not report having an affected relative in the epidemiological survey*

Study designs in cohort-based GWAS



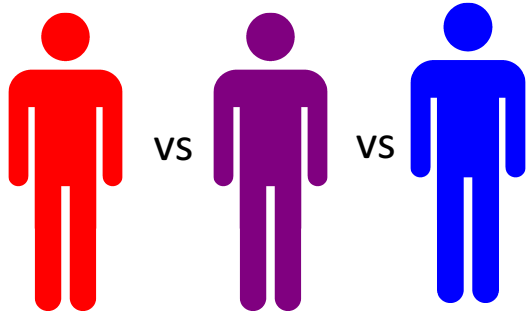
GWAS

1



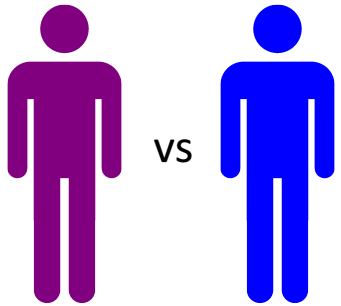
Exclude proxy-cases

2



Model proxy-cases

4



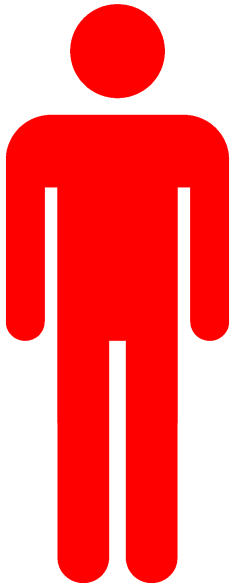
GWAX

Liu et al, 2017

3

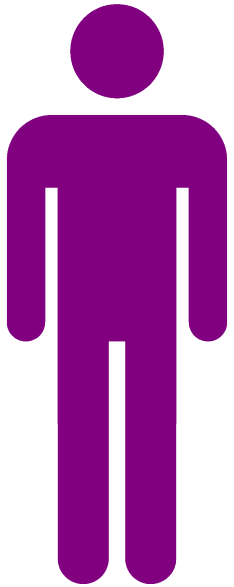
Modeling coefficient of relationship to a case

$F = 1$



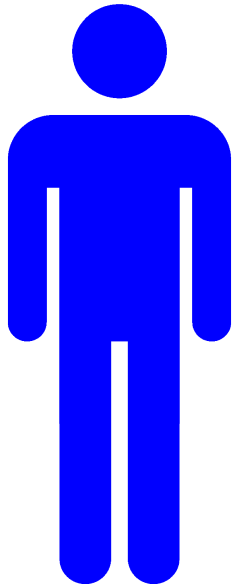
vs

$F = 0.5$

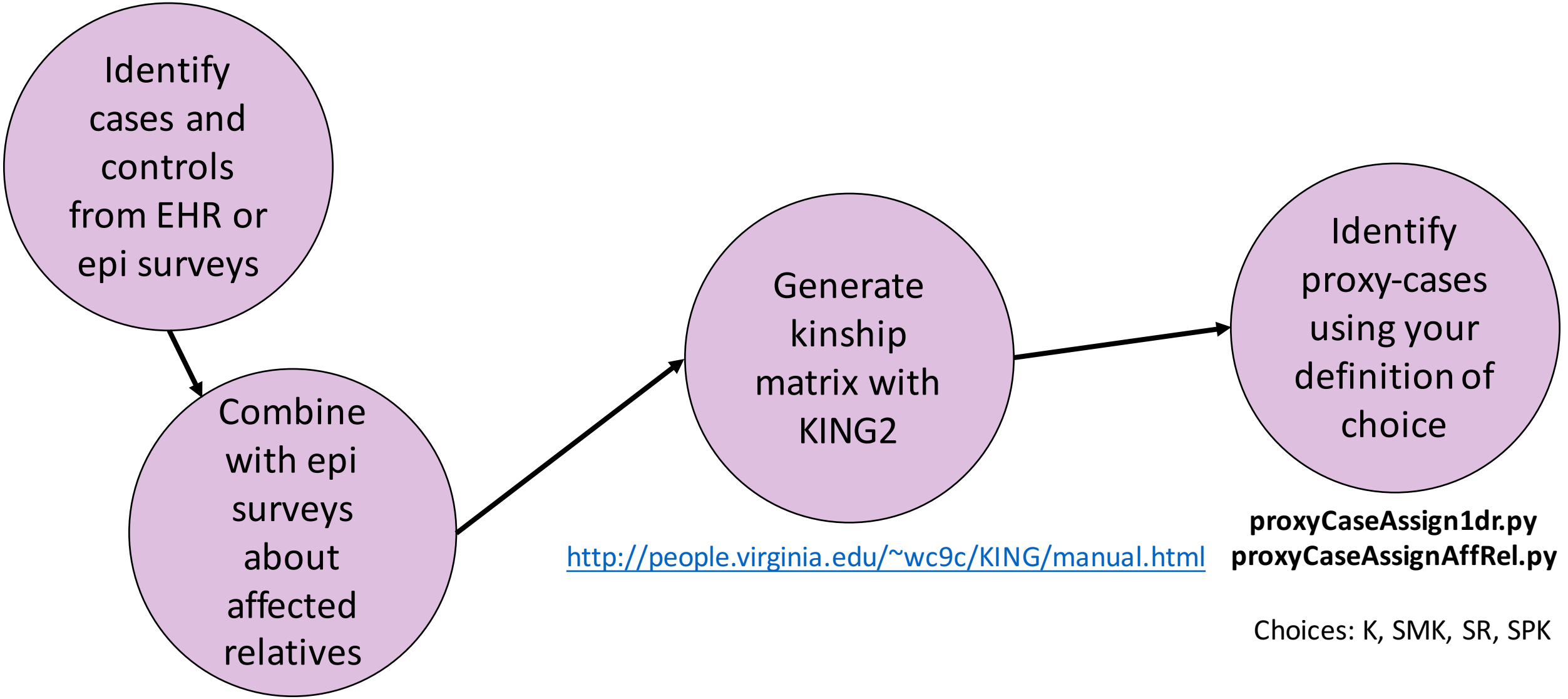


vs

$F = 0$



proxyPower workflow

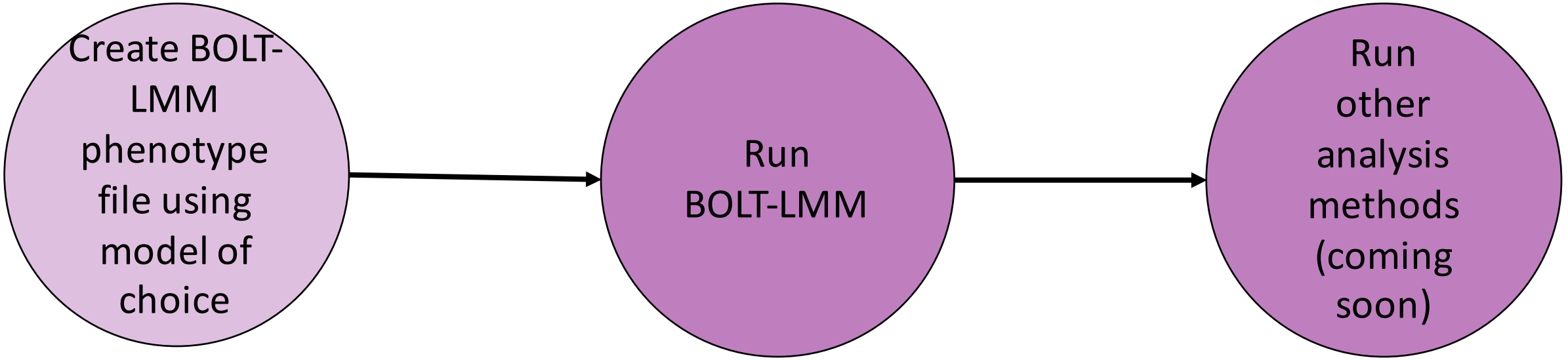


http://people.virginia.edu/~wc9c/KING/manual.html

proxyCaseAssign1dr.py
proxyCaseAssignAffRel.py

Choices: K, SMK, SR, SPK

proxyPower workflow



proxyModel.py

https://data.broadinstitute.org/alkesgroup/BOLT-LMM/

Choices: 1, 2, 3, 4, 5