GWAS and GS Are as Easy as Clicking and Dragging with iPat

World Congress On Genetics Applied to Livestock Production (WCGALP, 11th)

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WASHINGTON STATE UNIVERSITY

The Challenges of Biologists

Programming skills
Big data
Formatting
Modeling
…

iPat makes it easier for biologists to analyze data and stay focused on biology

The Challenges of Biologists

Programming skills
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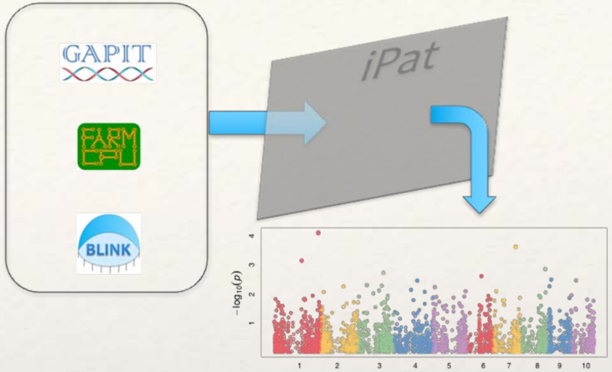


iPat

iPat makes it easier for biologists to analyze data and stay focused on biology

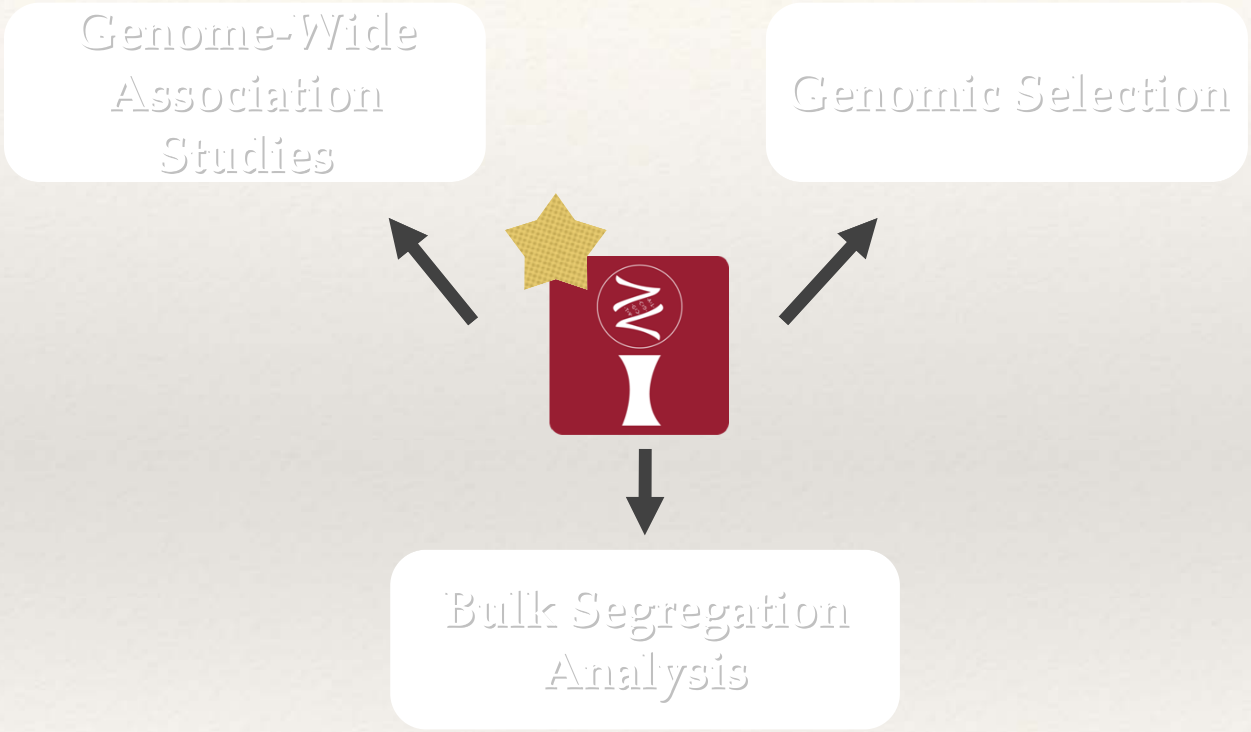
The Focus of iPat

Bio-sample

Sequencing

Functional Genomics Studies

The Focus of iPat



Finding Associated Genes

Genome-Wide Association Studies

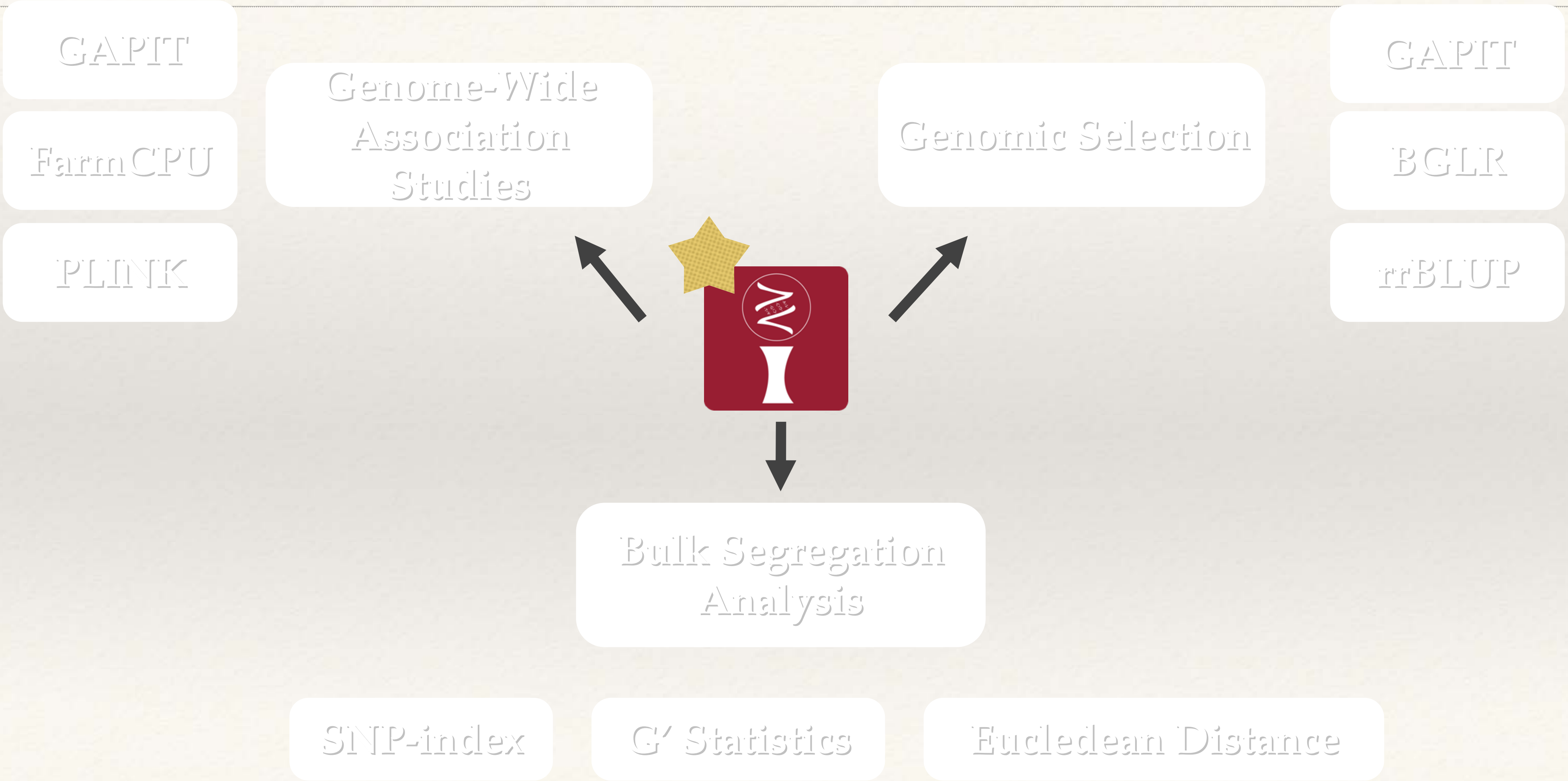
Genomic Selection

Predict Phenotype

Bulk Segregation Analysis

Finding Associated Genes by Pool Sequencing

Embedded Tools in One Place!



Genome-Wide Association Studies

GAPIT

FarmCPU

PLINK

Genomic Selection

GAPIT

BGLR

rrBLUP

Bulk Segregation Analysis

SNP-index

G′ Statistics

Eucledean Distance

Unfortunately… not all formats work on them

Tool	Supported Format	
GAPIT	Numeric	Hapmap
FarmCPU	Numeric	
BGLR	Numeric	
rrBLUP	Numeric	
PLINK	PLINK	Binary

Unfortunately… not all formats work on them

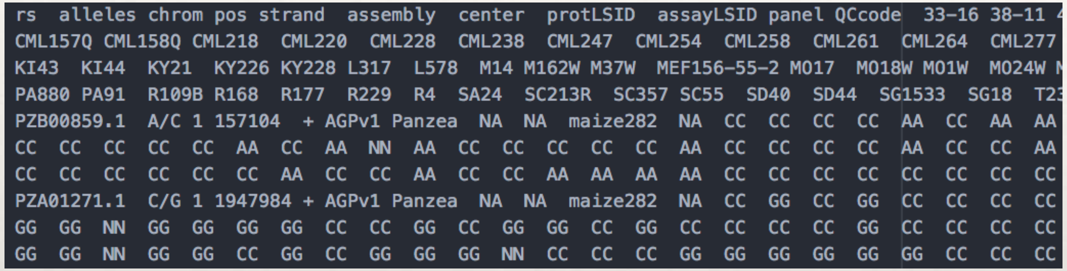
Tool	Supported Format	
GAPIT	Numeric	Hapmap
FarmCPU	Numeric	
BGLR	Numeric	
rrBLUP	Numeric	
PLINK	PLINK	Binary

?

VCF

Inspect the format

Hapmap



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rs  alleles chrom pos strand  assembly  center  protLSID  assayLSID panel QCcode  33-16 38-11 4
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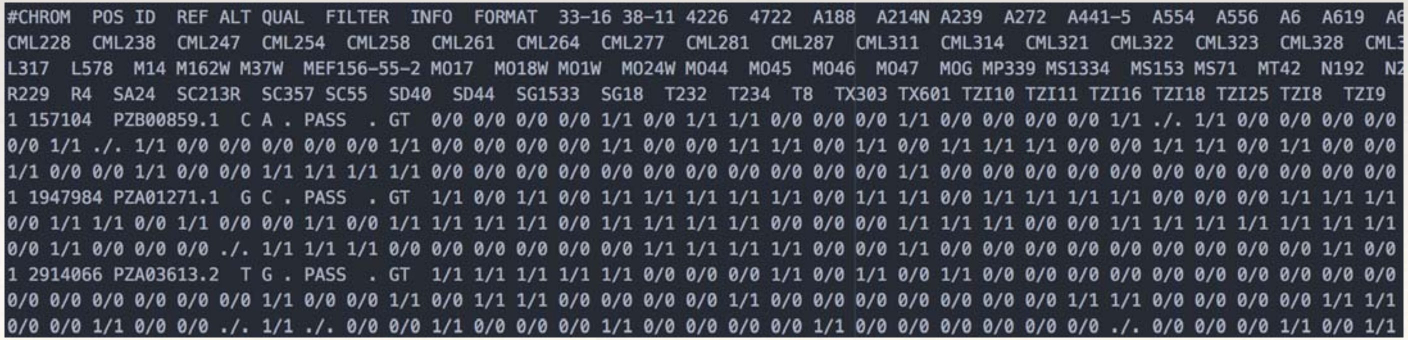
KI43  KI44  KY21  KY226 KY228 L317  L578  M14 M162W M37W  MEF156-55-2 MO17  MO18W MO1W  MO24W M
PA880 PA91  R109B R168  R177  R229  R4  SA24  SC213R  SC357 SC55  SD40  SD44  SG1533  SG18  T23
PZB00859.1  A/C 1 157104  + AGPv1 Panzea  NA  NA  maize282  NA  CC  CC  CC  CC  AA  CC  AA  AA
CC  CC  CC  CC  CC  AA  CC  AA  NN  AA  CC  CC  CC  CC  CC  AA  CC  CC  CC  CC  AA  CC  CC  AA
CC  CC  CC  CC  CC  CC  AA  CC  CC  AA  CC  CC  AA  AA  AA  AA  CC  CC  CC  CC  CC  CC  CC  CC
PZA01271.1  C/G 1 1947984 + AGPv1 Panzea  NA  NA  maize282  NA  CC  GG  CC  GG  CC  CC  CC  CC
GG  GG  NN  GG  GG  GG  GG  CC  CC  GG  CC  GG  GG  CC  GG  CC  CC  CC  CC  GG  CC  CC  CC  CC
GG  GG  NN  GG  GG  CC  GG  CC  GG  GG  GG  NN  CC  CC  CC  GG  GG  GG  GG  GG  GG  CC  CC  CC
```

PLINK

```
abc1  abc1  0 0 1 1 T T T T T T A A A A C C T T G G C C T T G G A
A A G G T T G G C C A A C C G G A A T T T T C C A A C C G A T T G
A A T T C C C C A A A A T T C C C C T T C C A A G T A A C C C C C
A A T T C C G T C C G G A A C C A A C C C C G G A A T C C A C C C
A A C C G G T T G G G A A A G G C A A A G G A A C C C T C C T T C
T T A A G G T T T T G G T T T T C T C C C C T T G G C C C C G G G
C C G G T T A A C C C C C C C C A A G G C C C C G G G G G G T T A
```

VCF



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CML228  CML238  CML247  CML254  CML258  CML261  CML264  CML277  CML281  CML287  CML311  CML314  CML321  CML322  CML323  CML328  CML3
L317  L578  M14 M162W M37W  MEF156-55-2 MO17  MO18W MO1W  MO24W MO44  MO45  MO46  MO47  MOG MP339 MS1334  MS153 MS71  MT42  N192  N2
R229  R4  SA24  SC213R  SC357 SC55  SD40  SD44  SG1533  SG18  T232  T234  T8  TX303 TX601 TZI10 TZI11 TZI16 TZI18 TZI25 TZI8  TZI9
1 157104  PZB00859.1  C A . PASS  . GT  0/0 0/0 0/0 0/0 1/1 0/0 1/1 1/1 0/0 0/0 0/0 1/1 0/0 0/0 0/0 0/0 1/1 ./. 1/1 0/0 0/0 0/0 0/0
0/0 1/1 ./. 1/1 0/0 0/0 0/0 0/0 0/0 1/1 0/0 0/0 0/0 0/0 1/1 0/0 0/0 1/1 1/1 0/0 1/1 0/0 1/1 1/1 1/1 0/0 0/0 1/1 1/1 0/0 1/1 0/0 0/0
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1 1947984 PZA01271.1  G C . PASS  . GT  1/1 0/0 1/1 0/0 1/1 1/1 1/1 1/1 1/1 0/0 1/1 1/1 0/0 1/1 1/1 1/1 1/1 0/0 0/0 0/0 1/1 1/1 1/1
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0/0 1/1 0/0 0/0 0/0 ./. 1/1 1/1 1/1 0/0 0/0 0/0 0/0 0/0 0/0 1/1 1/1 1/1 1/1 0/0 0/0 1/1 0/0 0/0 0/0 0/0 0/0 0/0 0/0 0/0 0/0 1/1 0/0
1 2914066 PZA03613.2  T G . PASS  . GT  1/1 1/1 1/1 1/1 1/1 0/0 0/0 0/0 1/1 0/0 1/1 0/0 1/1 0/0 0/0 0/0 0/0 0/0 0/0 0/0 0/0 0/0 0/0
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0/0 0/0 1/1 0/0 0/0 ./. 1/1 ./. 0/0 0/0 1/1 0/0 0/0 0/0 1/1 0/0 0/0 0/0 0/0 1/1 0/0 0/0 0/0 0/0 0/0 0/0 ./. 0/0 0/0 0/0 1/1 0/0 1/1
```
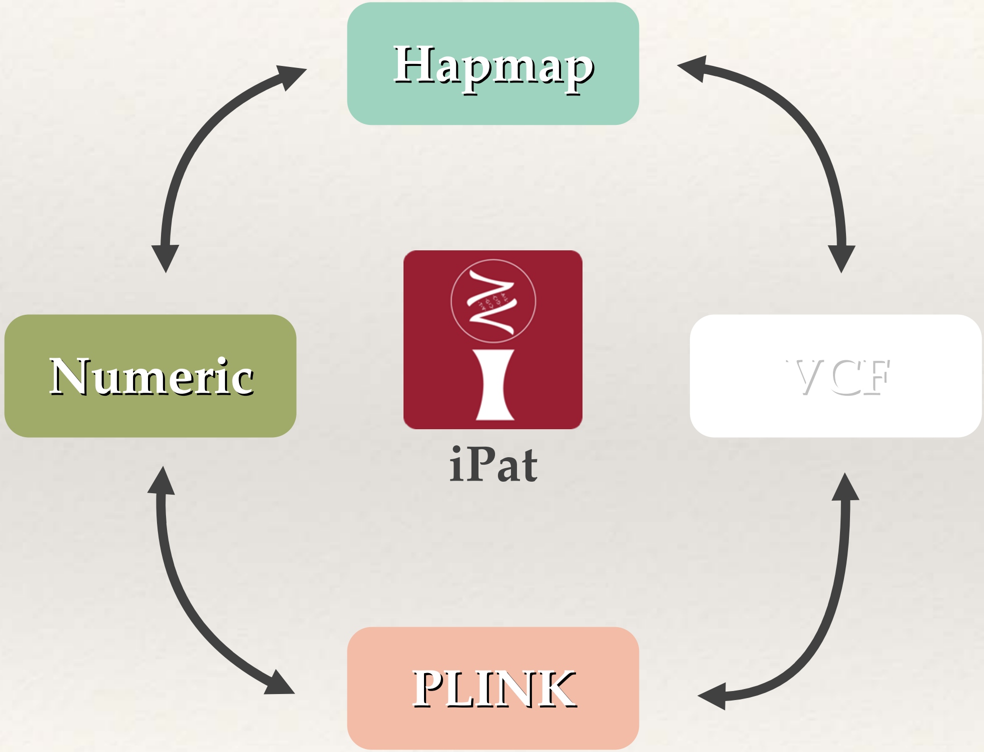

Format Free



Hapmap

Numeric

iPat

VCF

PLINK

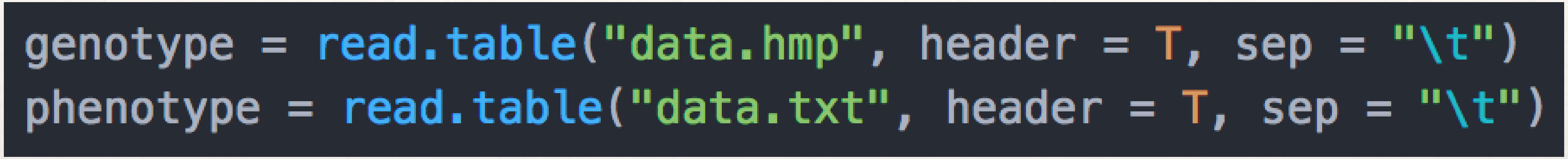
No need to spend effort on format conversion

Let’s work with iPat step by step!

Task : GWAS

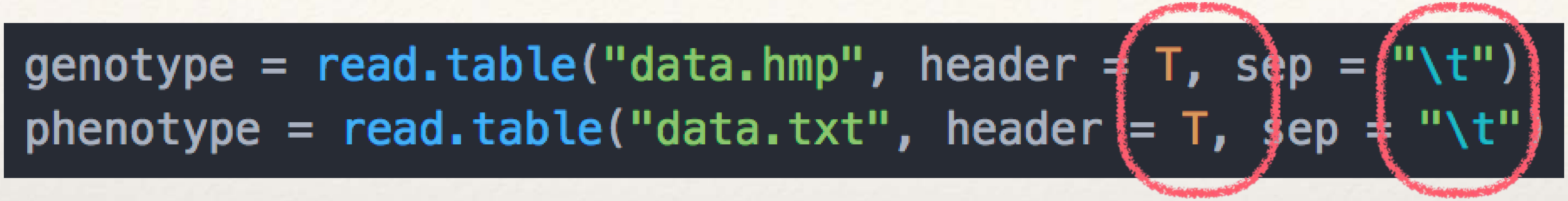
Import Data → **Quality Control** → **Define a Method** → **Inspect Results**

Import Data



```
genotype = read.table("data.hmp", header = T, sep = "\t")
phenotype = read.table("data.txt", header = T, sep = "\t")
```

Import Data



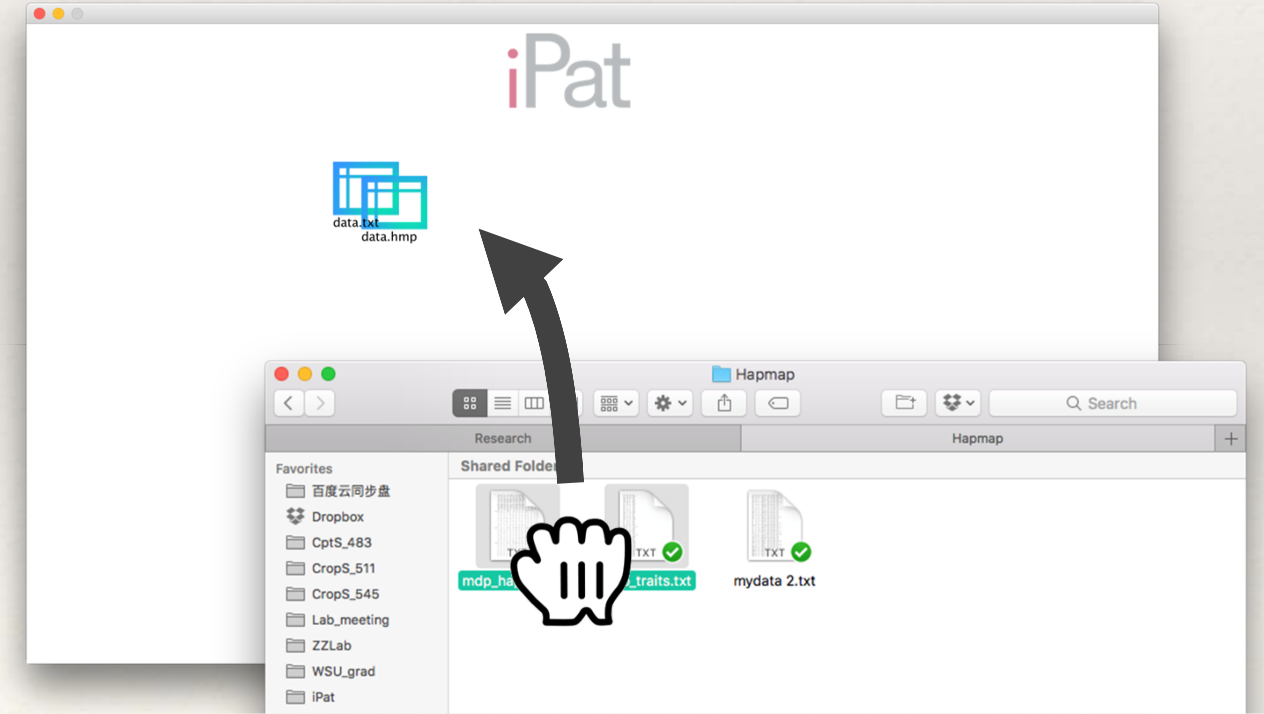
```
genotype = read.table("data.hmp", header = T, sep = "\t")
phenotype = read.table("data.txt", header = T, sep = "\t")
```

Does data contain a header?

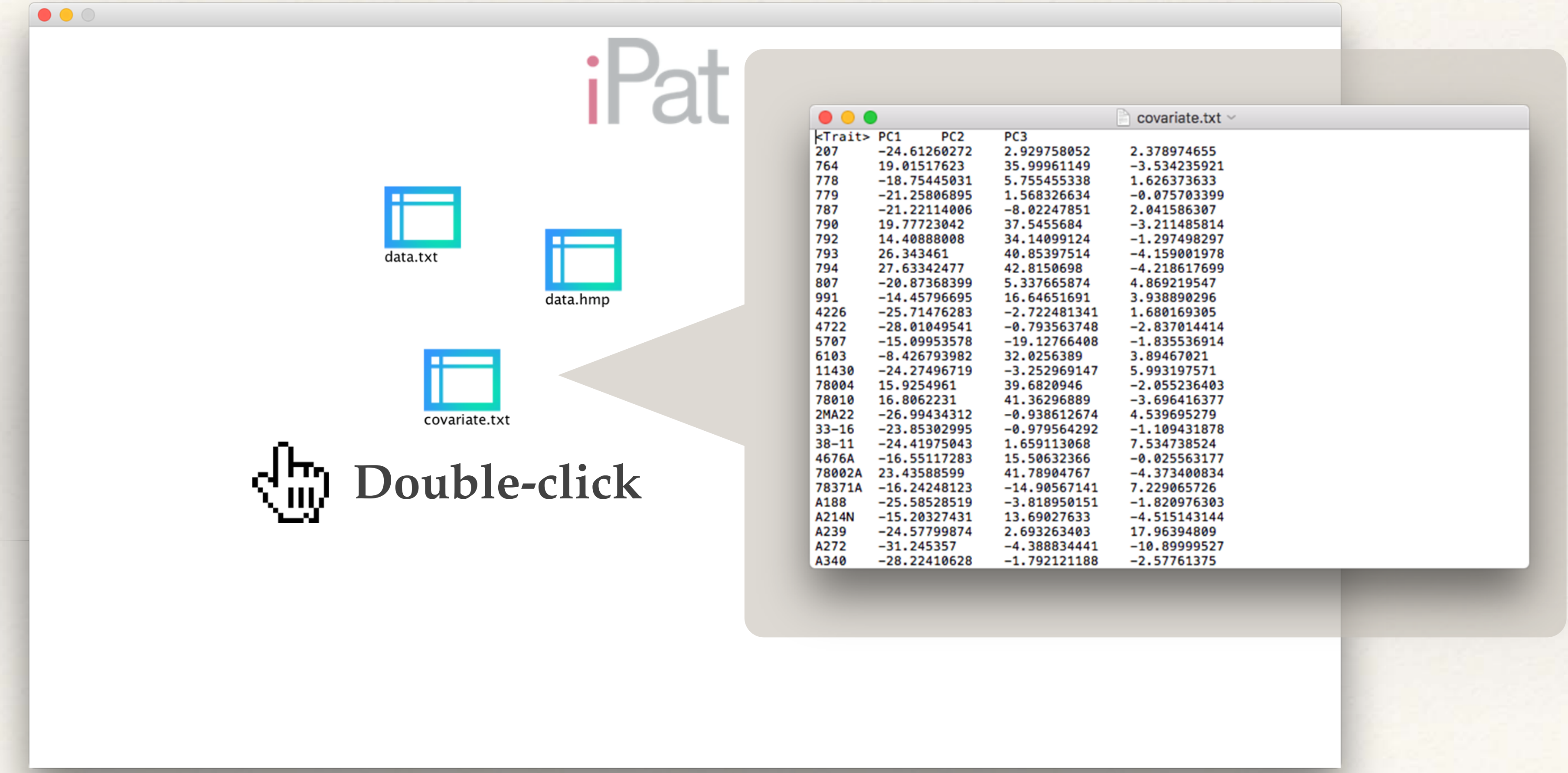
What's the delimiter? Tab? Comma? Space?

Drag and Drop To Import

Detect the header and delimiter automatically



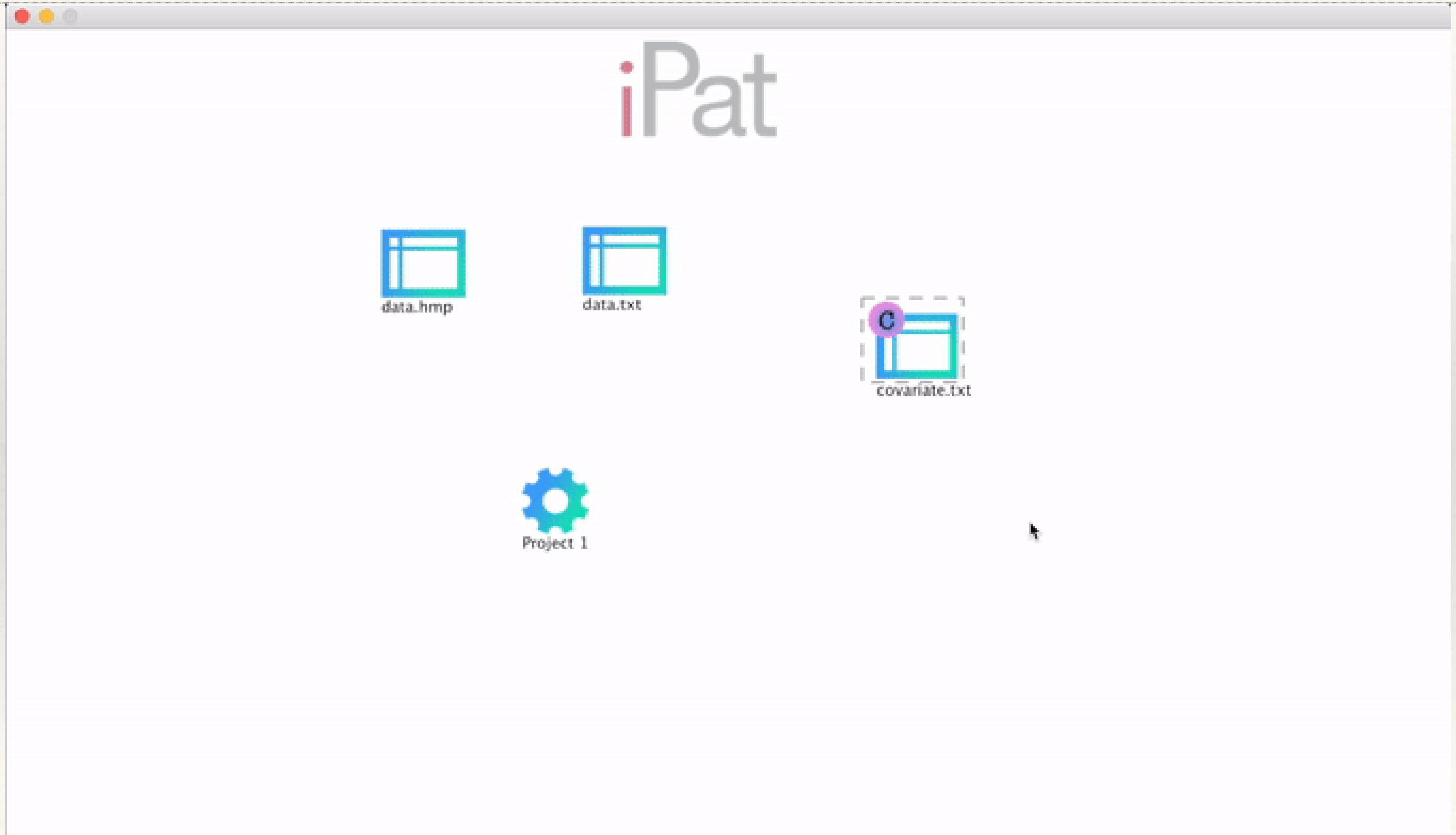
Inspect



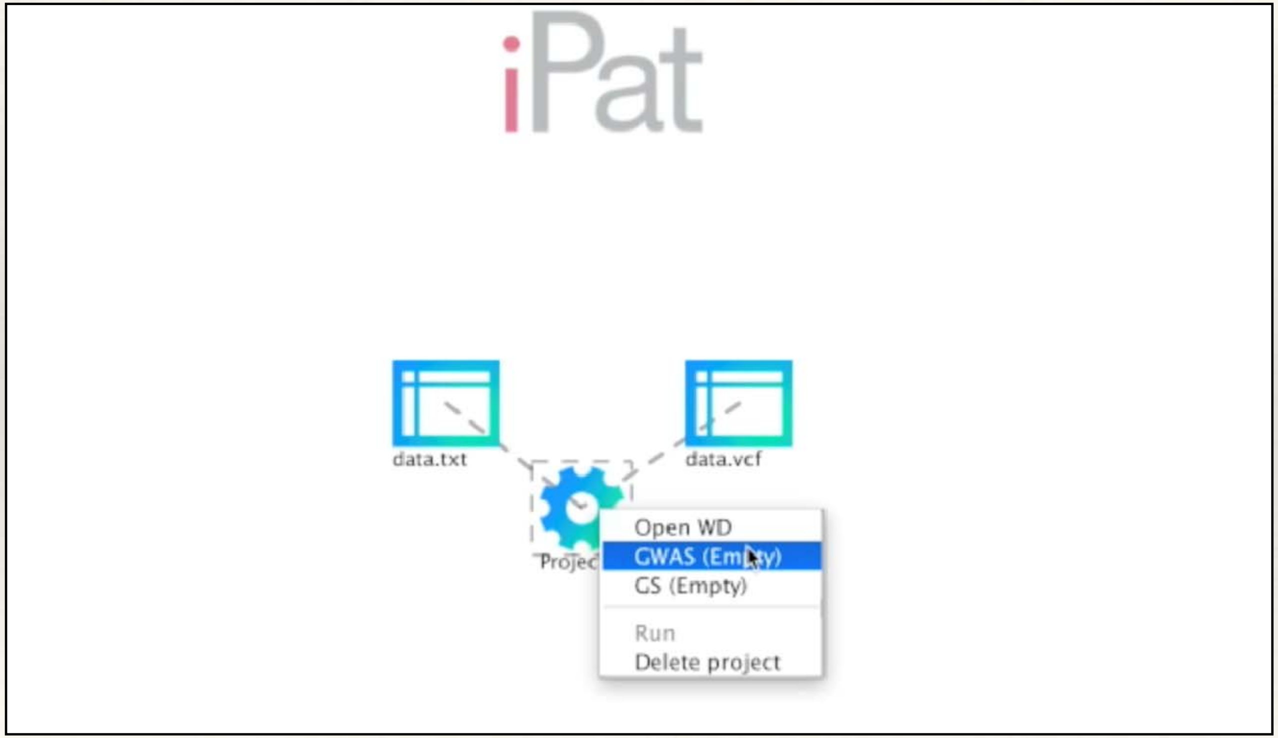
Double-click

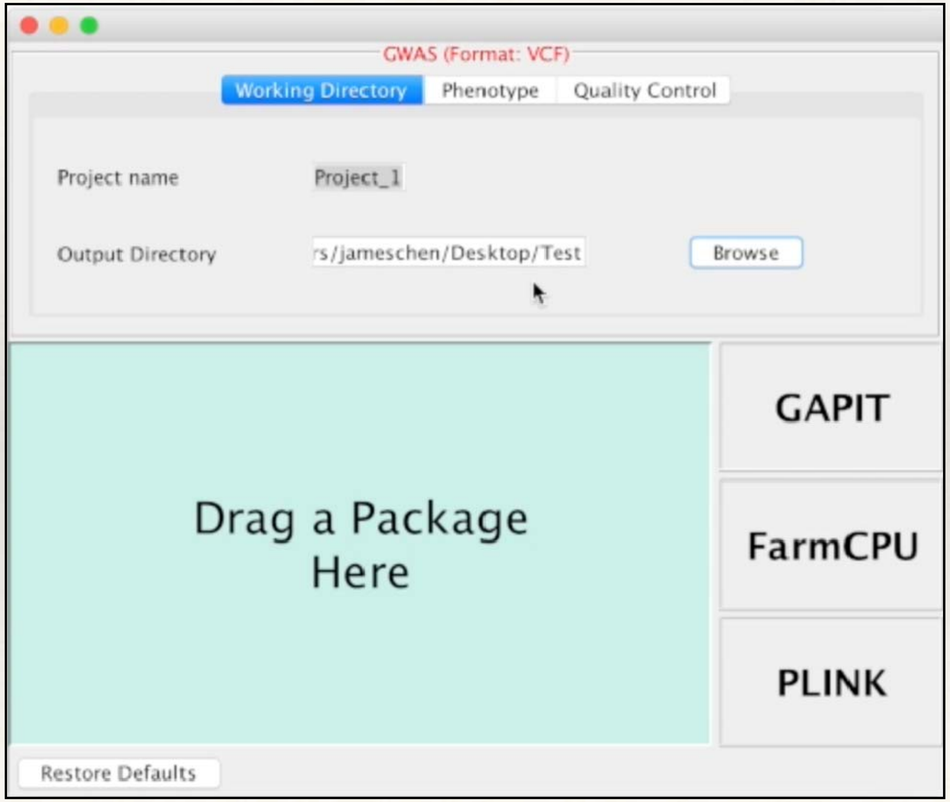
```
<Trait> PC1     PC2     PC3
207     -24.61260272    2.929758052     2.378974655
764     19.01517623     35.99961149     -3.534235921
778     -18.75445031    5.755455338     1.626373633
779     -21.25806895    1.568326634     -0.075703399
787     -21.22114006    -8.02247851     2.041586307
790     19.77723042     37.5455684      -3.211485814
792     14.40888008     34.14099124     -1.297498297
793     26.343461       40.85397514     -4.159001978
794     27.63342477     42.8150698      -4.218617699
807     -20.87368399    5.337665874     4.869219547
991     -14.45796695    16.64651691     3.938890296
4226    -25.71476283    -2.722481341    1.680169305
4722    -28.01049541    -0.793563748    -2.837014414
5707    -15.09953578    -19.12766408    -1.835536914
6103    -8.426793982    32.0256389      3.89467021
11430   -24.27496719    -3.252969147    5.993197571
78004   15.9254961      39.6820946      -2.055236403
78010   16.8062231      41.36296889     -3.696416377
2MA22   -26.99434312    -0.938612674    4.539695279
33-16   -23.85302995    -0.979564292    -1.109431878
38-11   -24.41975043    1.659113068     7.534738524
4676A   -16.55117283    15.50632366     -0.025563177
78002A  23.43588599     41.78904767     -4.373400834
78371A  -16.24248123    -14.90567141    7.229065726
A188    -25.58528519    -3.818950151    -1.820976303
A214N   -15.20327431    13.69027633     -4.515143144
A239    -24.57799874    2.693263403     17.96394809
A272    -31.245357      -4.388834441    -10.89999527
A340    -28.22410628    -1.792121188    -2.57761375
```

Build a Module



Define Your Analysis

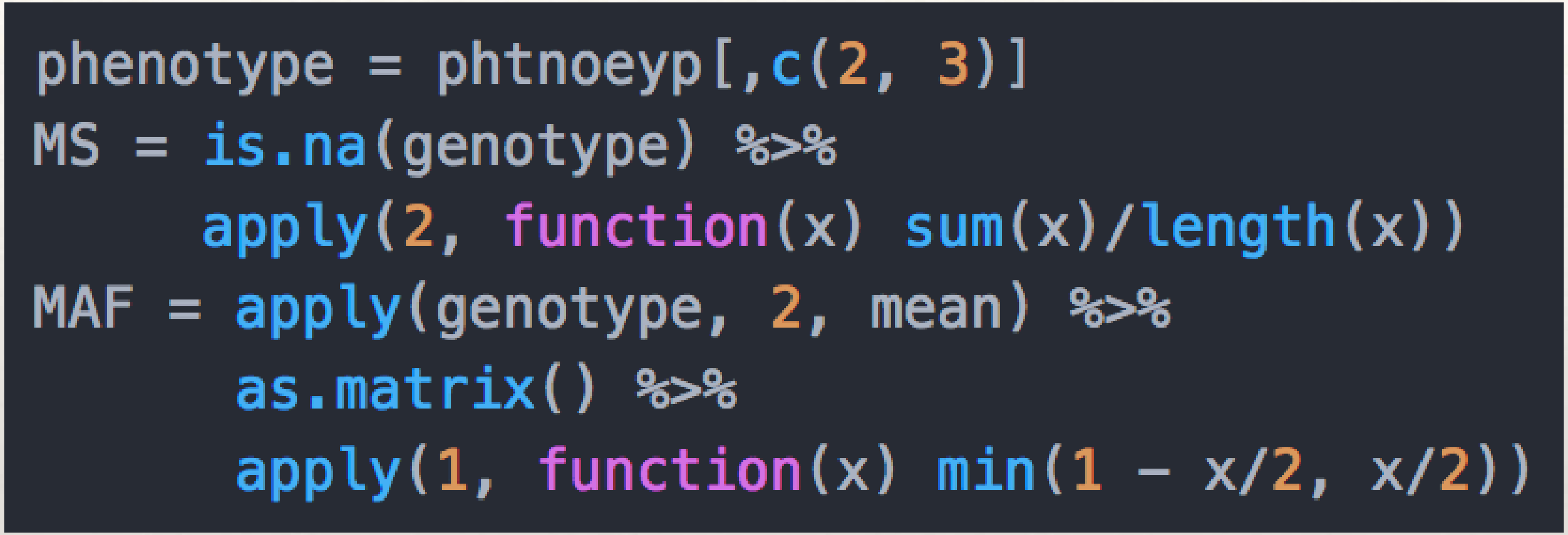




Project name Project_1

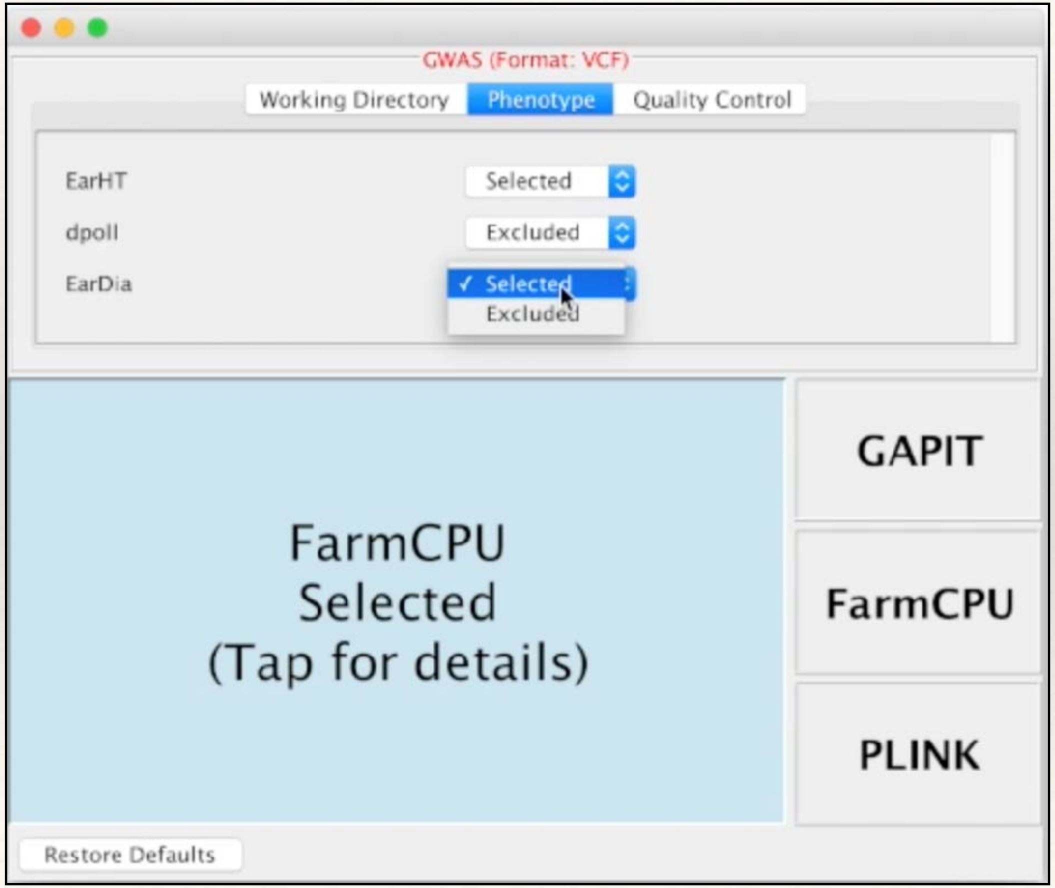
Output Directory rs/jameschen/Desktop/Test

Missing Rate? Minor Allele Frequency?



```
phenotype = phtnoeyp[,c(2, 3)]
MS = is.na(genotype) %>%
     apply(2, function(x) sum(x)/length(x))
MAF = apply(genotype, 2, mean) %>%
      as.matrix() %>%
      apply(1, function(x) min(1 - x/2, x/2))
```

iPat Got You Back!

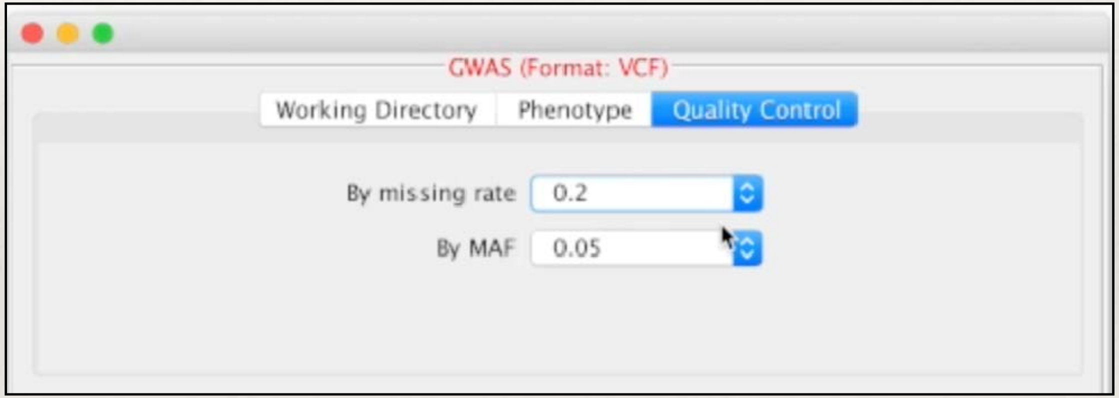

GWAS (Format: VCF)

EarHT Selected

dpoll Excluded

EarDia ✓ Selected

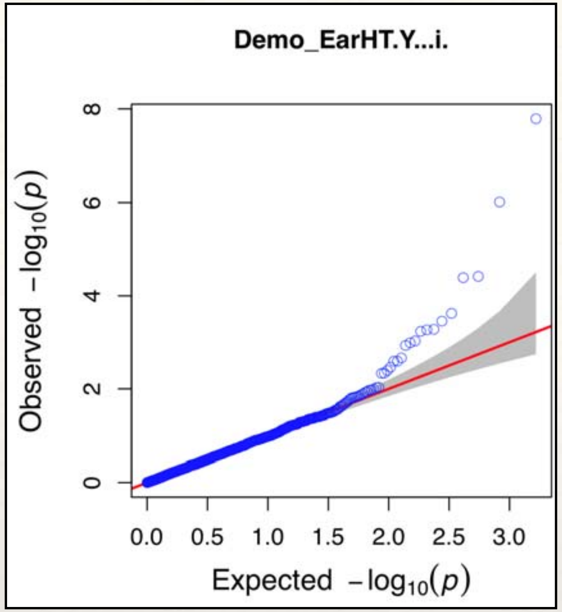
FarmCPU
Selected
(Tap for details)

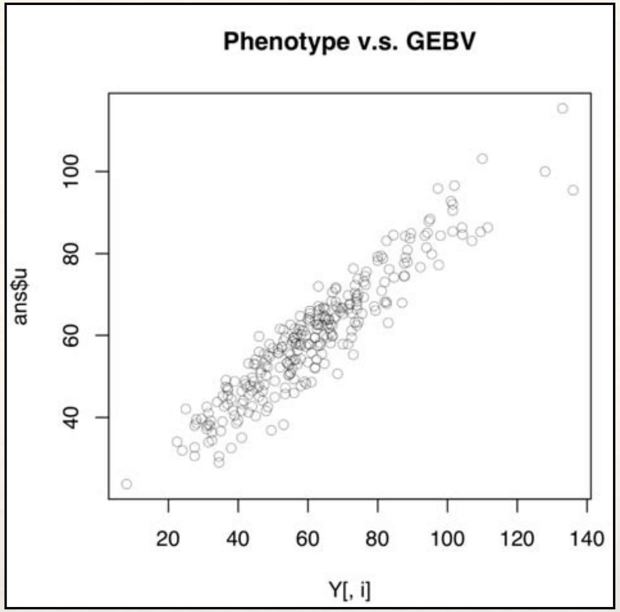

GWAS (Format: VCF)

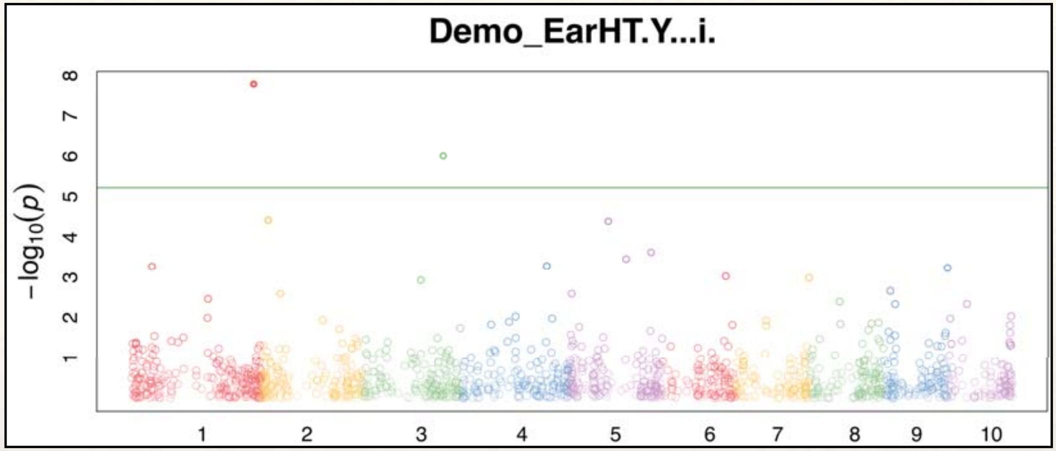
By missing rate 0.2

By MAF 0.05

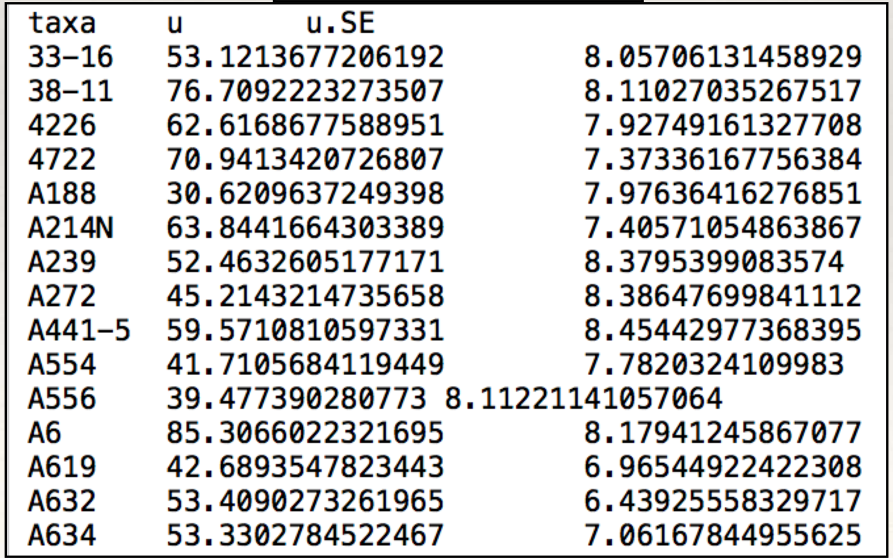

iPat





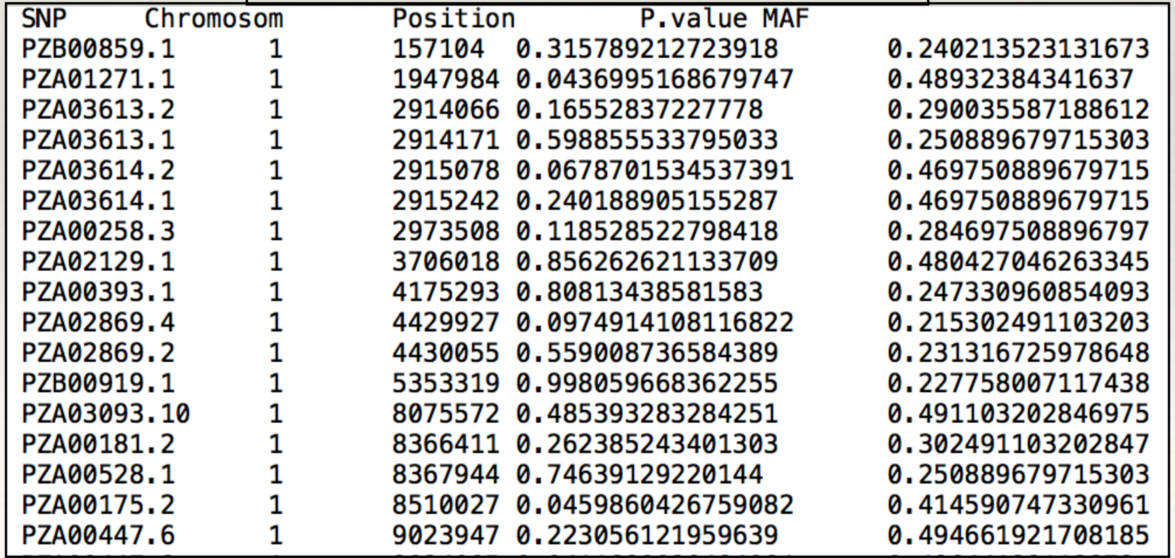


Breeding value



```
taxa    u         u.SE
33-16   53.1213677206192        8.05706131458929
38-11   76.7092223273507        8.11027035267517
4226    62.6168677588951        7.92749161327708
4722    70.941342072680 7        7.37336167756384
A188    30.6209637249398        7.97636416276851
A214N   63.8441664303389        7.40571054863867
A239    52.4632605177171        8.3795399083574
A272    45.2143214735658        8.38647699841112
A441-5  59.5710810597331        8.45442977368395
A554    41.7105684119449        7.7820324109983
A556    39.477390280773 8.11221141057064
A6      85.3066022321695        8.17941245867077
A619    42.6893547823443        6.96544922422308
A632    53.4090273261965        6.43925558329717
A634    53.3302784522467        7.06167844955625
```

P value of association test



```
SNP      Chromosom        Position         P.value MAF
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PZA01271.1      1         1947984 0.0436995168679747      0.48932384341637
PZA03613.2      1         2914066 0.16552837227778        0.290035587188612
PZA03613.1      1         2914171 0.598855533795033       0.250889679715303
PZA03614.2      1         2915078 0.0678701534537391      0.469750889679715
PZA03614.1      1         2915242 0.240188905155287       0.469750889679715
PZA00258.3      1         2973508 0.118528522798418       0.284697508896797
PZA02129.1      1         3706018 0.856262621133709       0.480427046263345
PZA00393.1      1         4175293 0.80813438581583        0.247330960854093
PZA02869.4      1         4429927 0.0974914108116822      0.215302491103203
PZA02869.2      1         4430055 0.559008736584389       0.231316725978648
PZB00919.1      1         5353319 0.998059668362255       0.227758007117438
PZA03093.10     1         8075572 0.485393283284251       0.491103202846975
PZA00181.2      1         8366411 0.262385243401303       0.302491103202847
PZA00528.1      1         8367944 0.74639129220144        0.250889679715303
PZA00175.2      1         8510027 0.0459860426759082      0.414590747330961
PZA00447.6      1         9023947 0.22305612195963 9      0.494661921708185
```
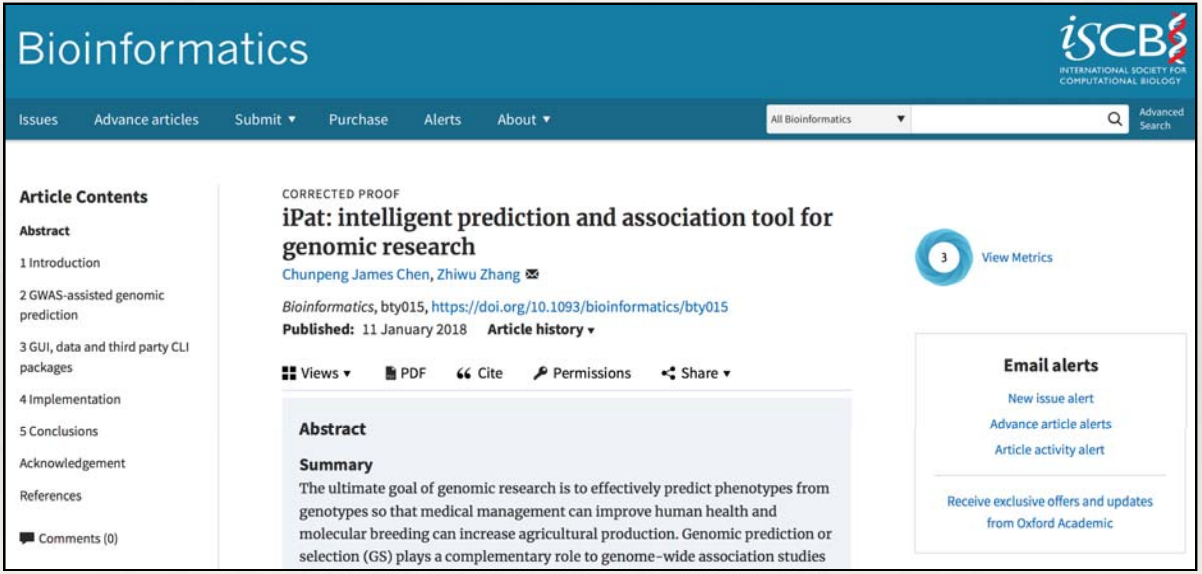

Availability



Article Contents

Abstract
1 Introduction
2 GWAS-assisted genomic prediction
3 GUI, data and third party CLI packages
4 Implementation
5 Conclusions
Acknowledgement
References
Comments (0)

CORRECTED PROOF

iPat: intelligent prediction and association tool for genomic research

Chunpeng James Chen, Zhiwu Zhang

Bioinformatics, bty015, https://doi.org/10.1093/bioinformatics/bty015
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Abstract

Summary

The ultimate goal of genomic research is to effectively predict phenotypes from genotypes so that medical management can improve human health and molecular breeding can increase agricultural production. Genomic prediction or selection (GS) plays a complementary role to genome-wide association studies

Published on Bioinformatics

http://zzlab.net/iPat/

Acknowledgement : Z.Z. Lab

PI:
Dr. Zhiwu Zhang

It's me
James Chen

Zhiwu Zhang Laboratory
for Statistical Genomics

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COMMISSION