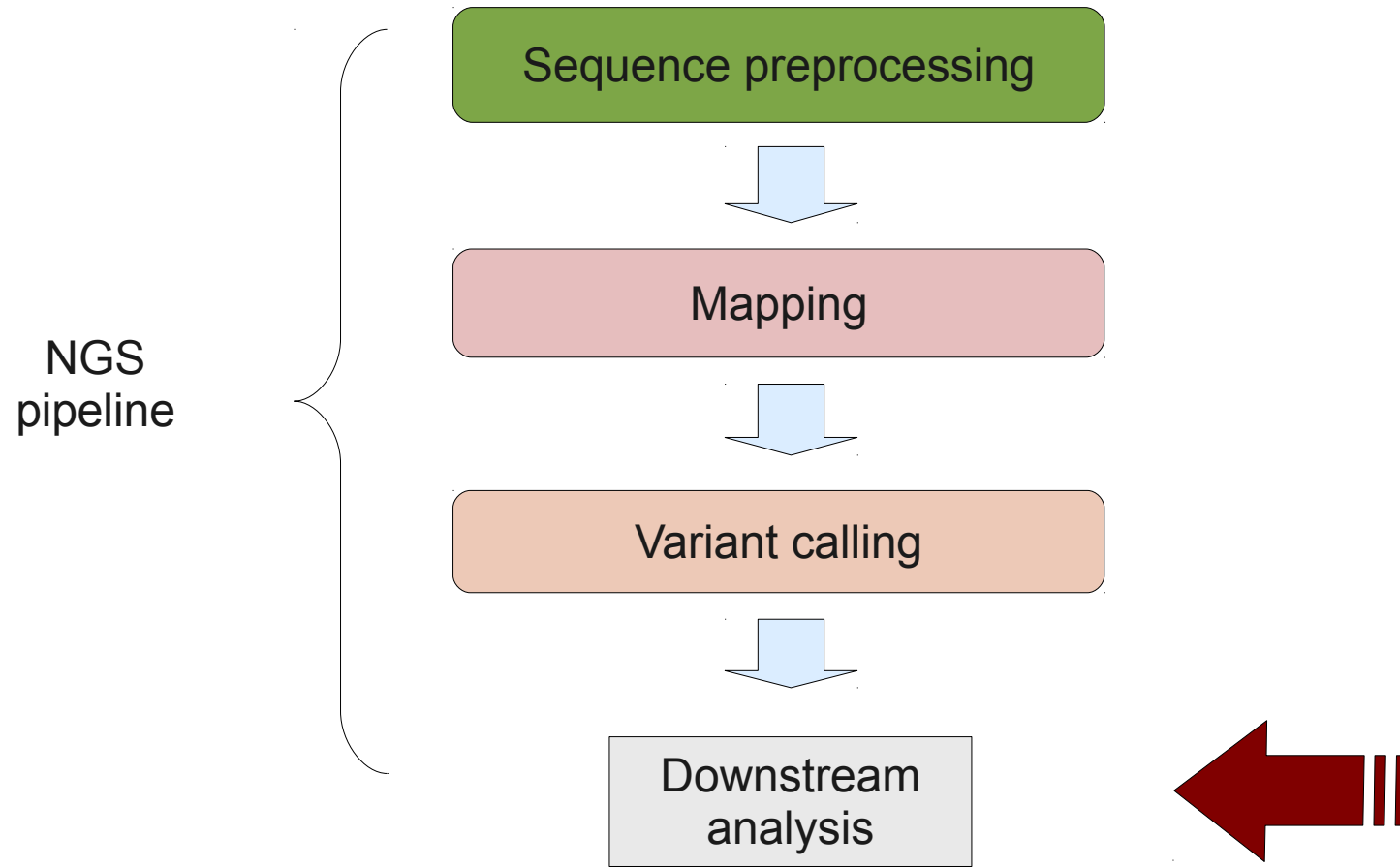


Results interpretation

Jorge Jiménez
jjimeneza@cipf.es
BIER - CIBERER
Genomics Department
Centro de Investigación Príncipe Felipe (CIPF)
(Valencia, Spain)

NGS Sequence preprocessing

Where we are?



<http://bioinfo.cipf.es/bierwiki>

[[group:grinberg:sidebar]]

ciberer **BIER**
PLATAFORMA DE INFORMACIÓN PARA LA INVESTIGACIÓN BÁSICA

Show page Old revisions Recent changes Sitemap Login

Search

Login

You are currently not logged in! Enter your authentication credentials below to log in. You need to have cookies enabled to log in.

Login

Username

Password

Remember me

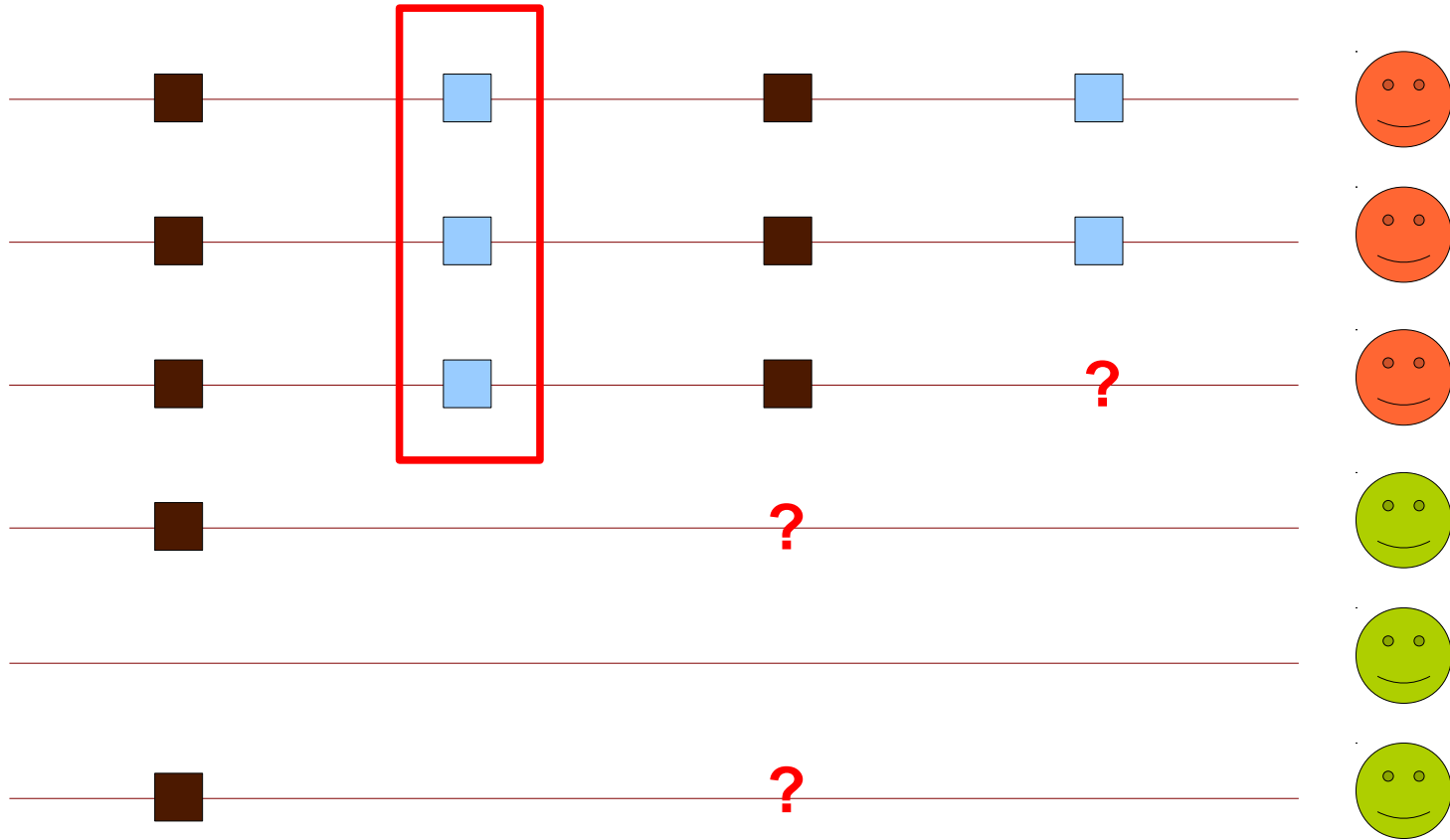
Login

You don't have an account yet? Just get one: [Register](#)

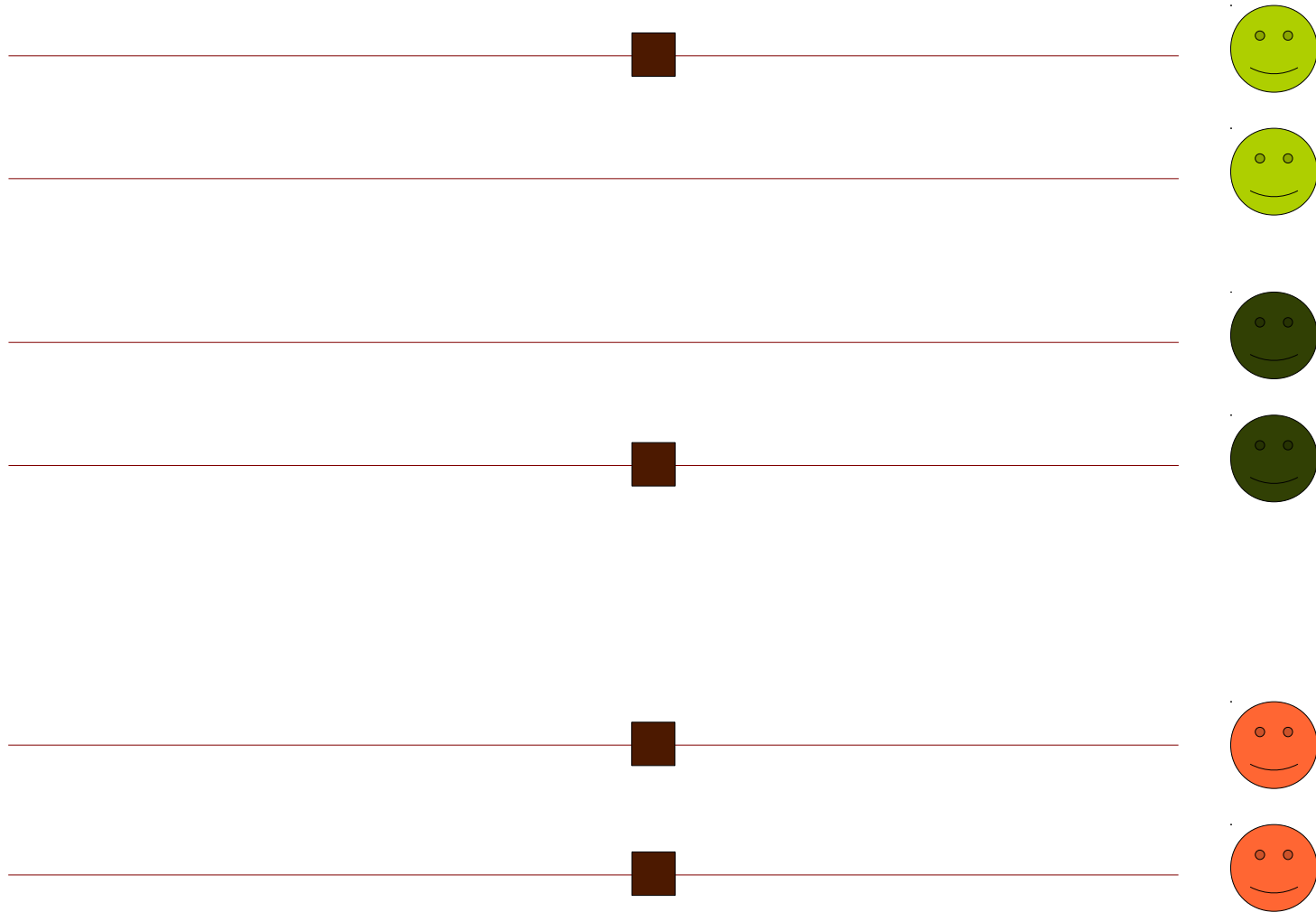
Forgotten your password? Get a new one: [Send new password](#)

Show page Old revisions [Back to top](#) [unknown link type]

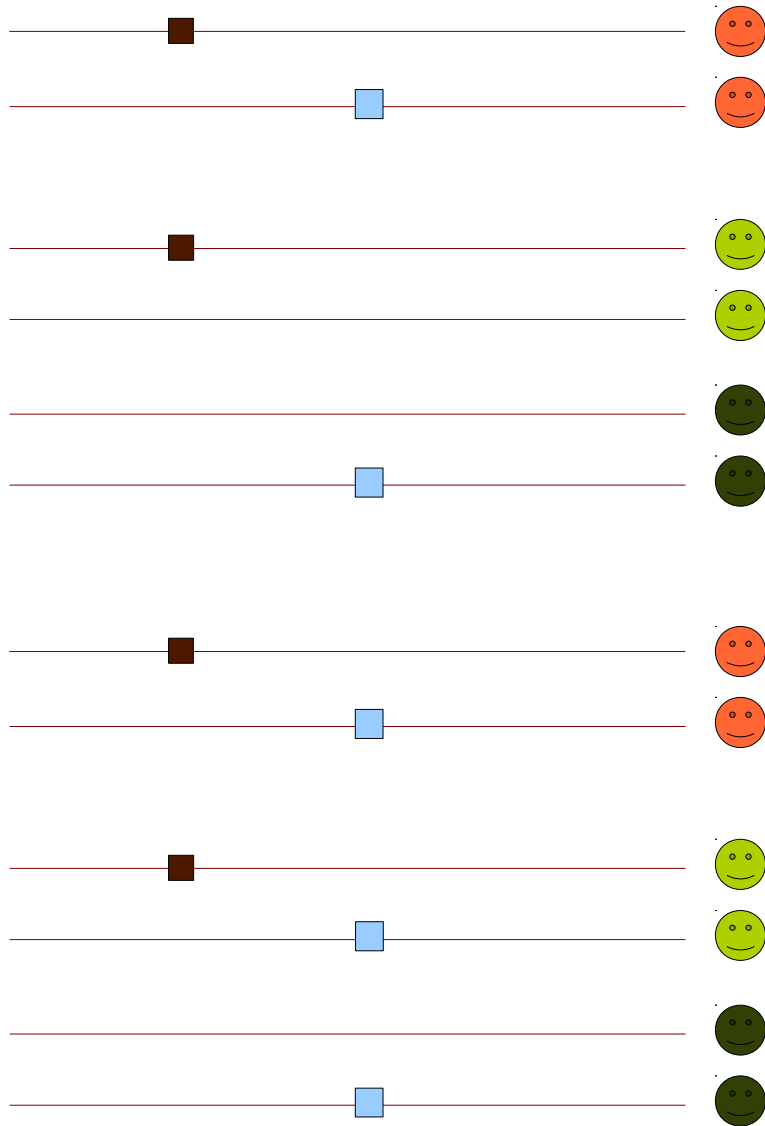
Dominant



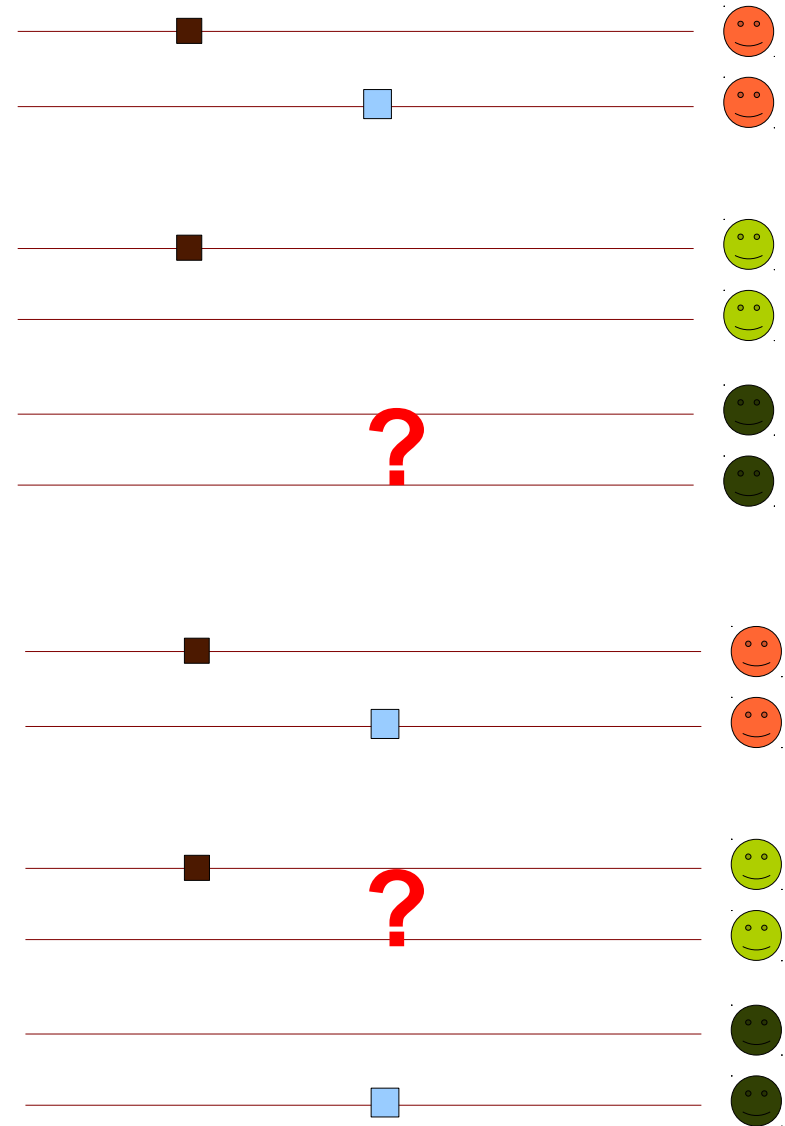
Recessive - homozygous



Recessive – compound heterozygosity



X

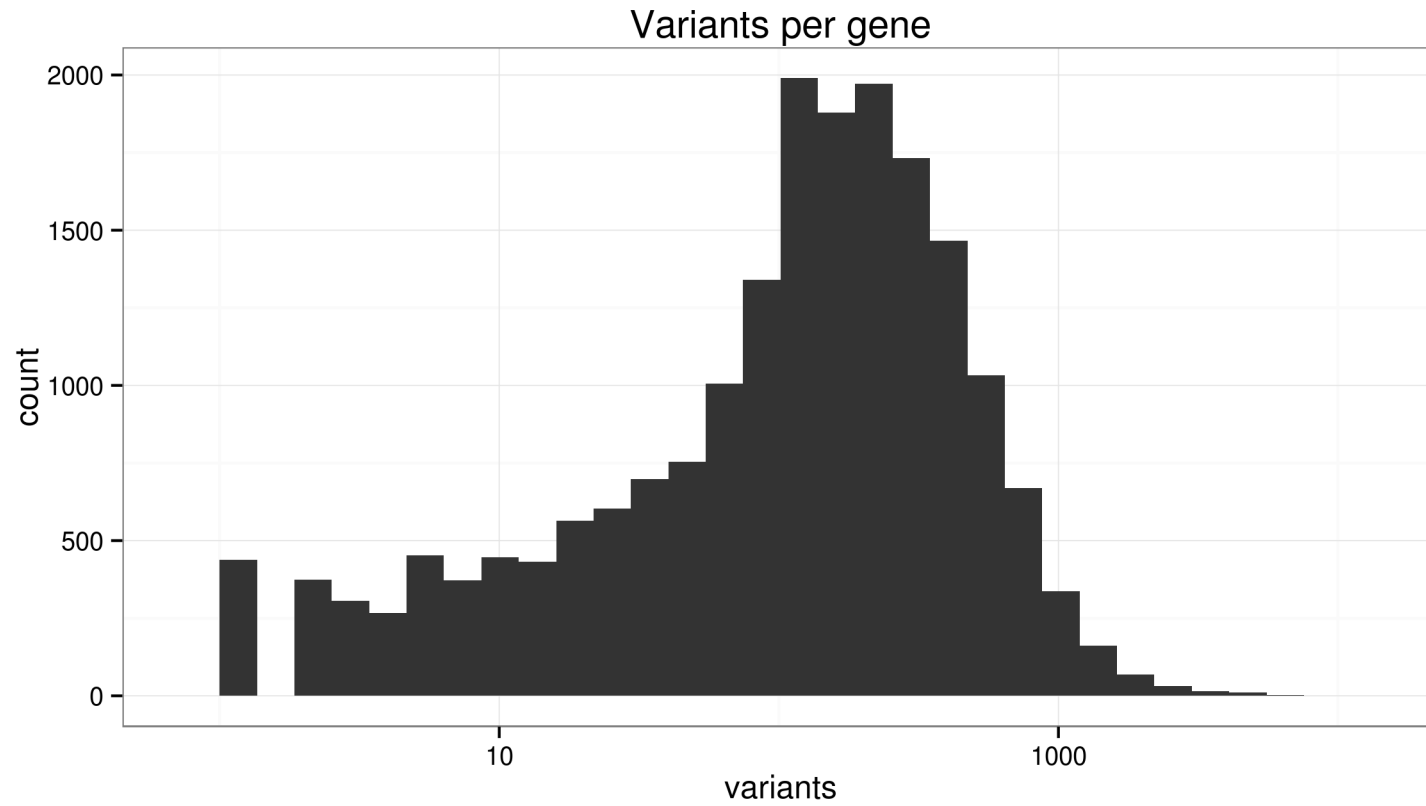


FILTERS

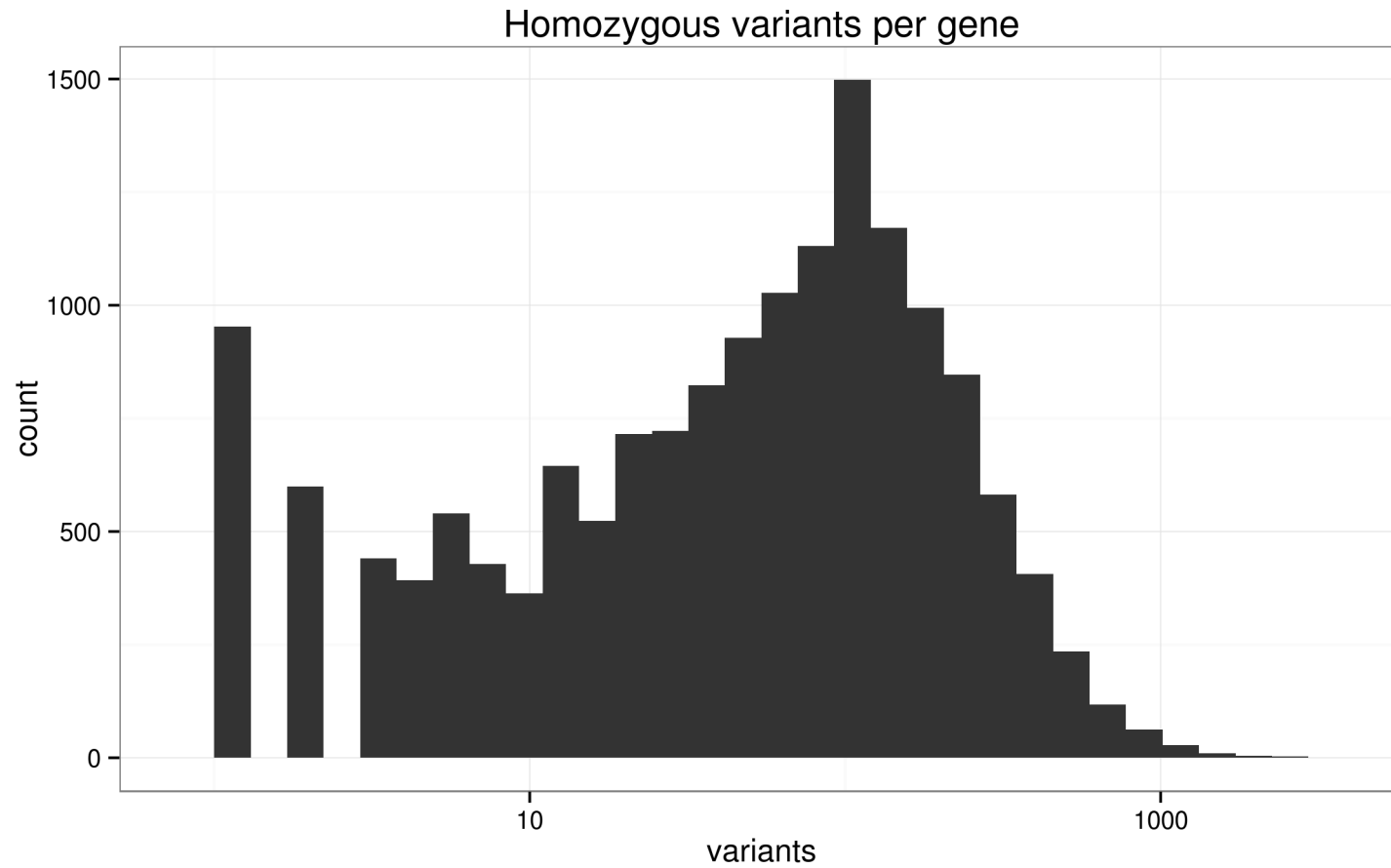
Available:

- Present in BIER samples (allele frequency)
- Consequence type
- % cases and controls with variant
- SNVs and indels
- 1000 genomes allele frequency
- Recessive: genes with deleterious variants
- De novo mutations: mark or delete
- present in dbSNP
- Regions

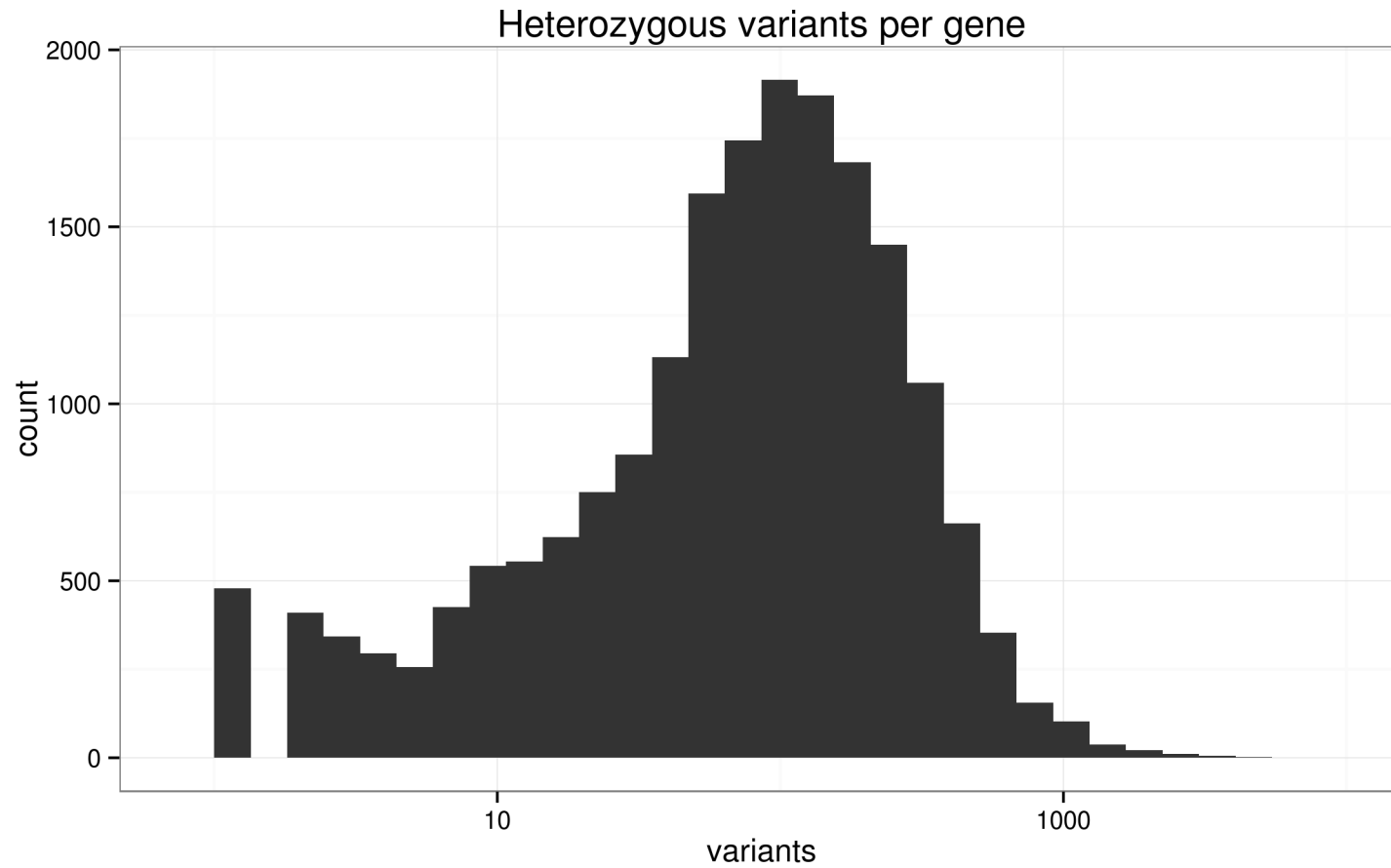
Some statistics: Variants per gene



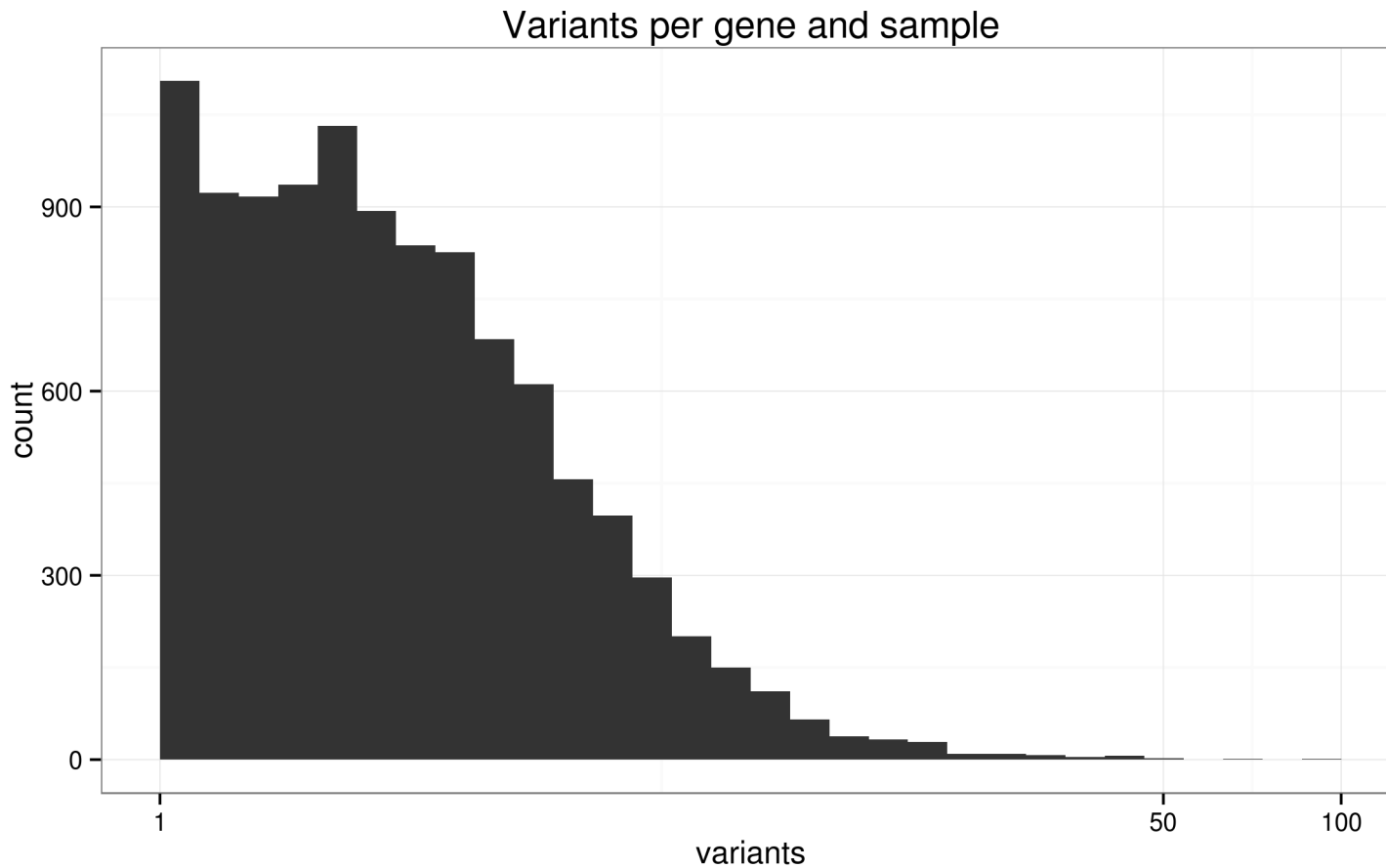
Some statistics: Homozygous variants per gene



Some statistics: Heterozygous variants per gene

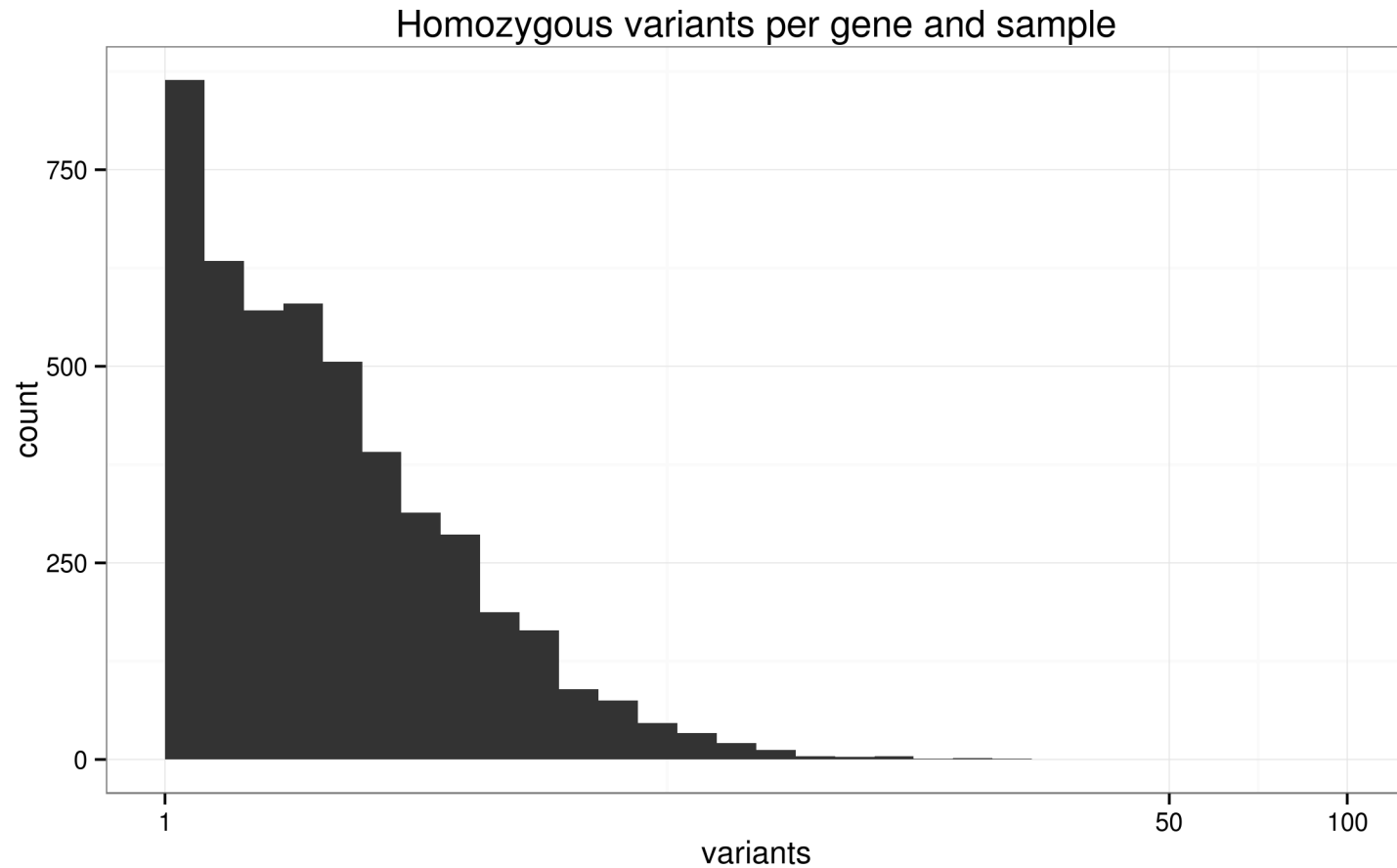


Some statistics: Variants per gene and sample

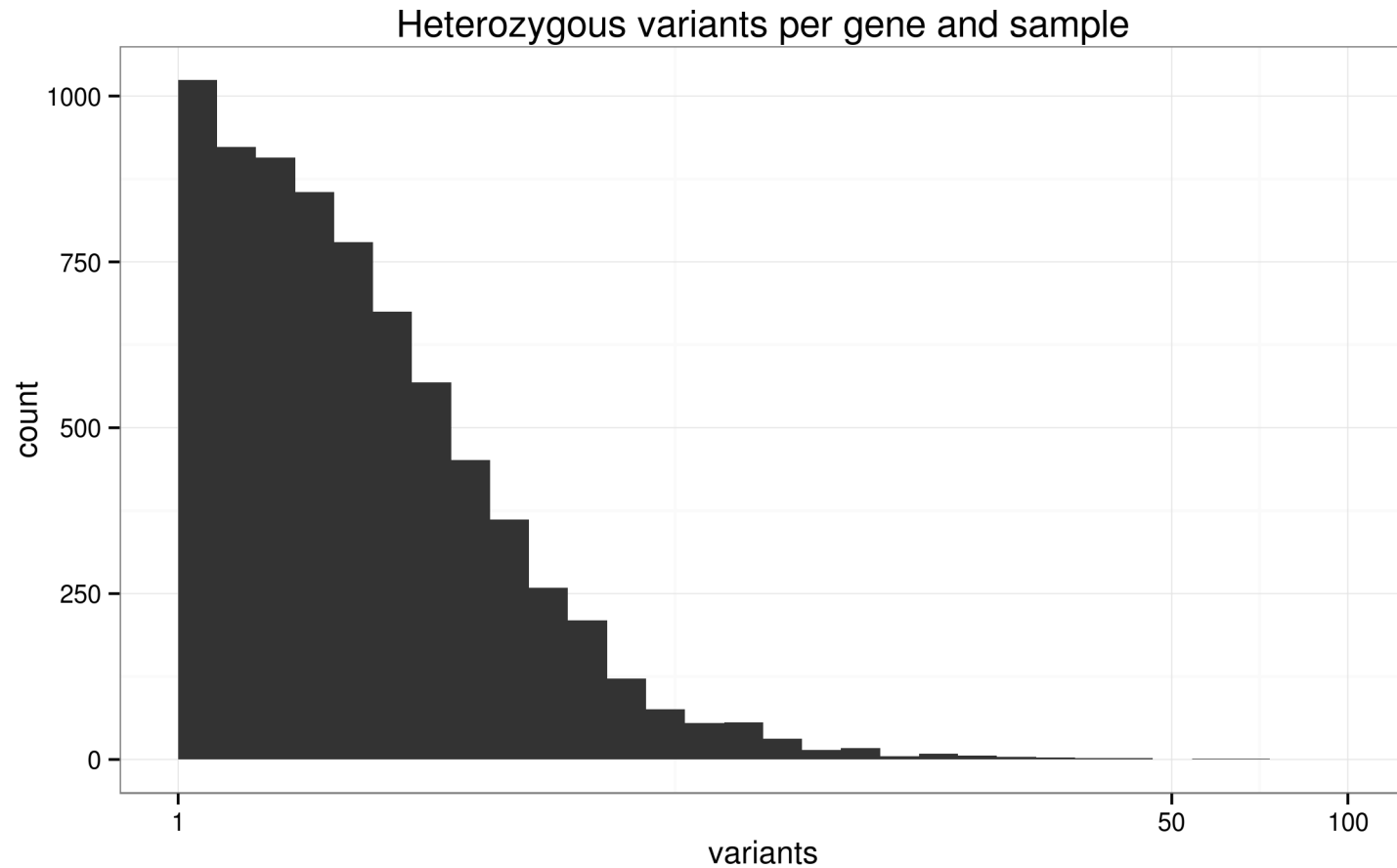


MUC16	88.59
PDE4DIP	64.61
MUC4	52.27
PKD1L2	47.99
AHNAK2	46.06
OR4C45	42.18
MAP2K3	41.96
TTN	40.95
HLA-A	39.93
HLA-C	39.89
OBSCN	39.79
DNAH17	39.26
OR4C3	38.50
LAMA5	36.59
KCNJ18	32.37
KCNJ12	32.37
HSPG2	31.37
SYNE1	31.03

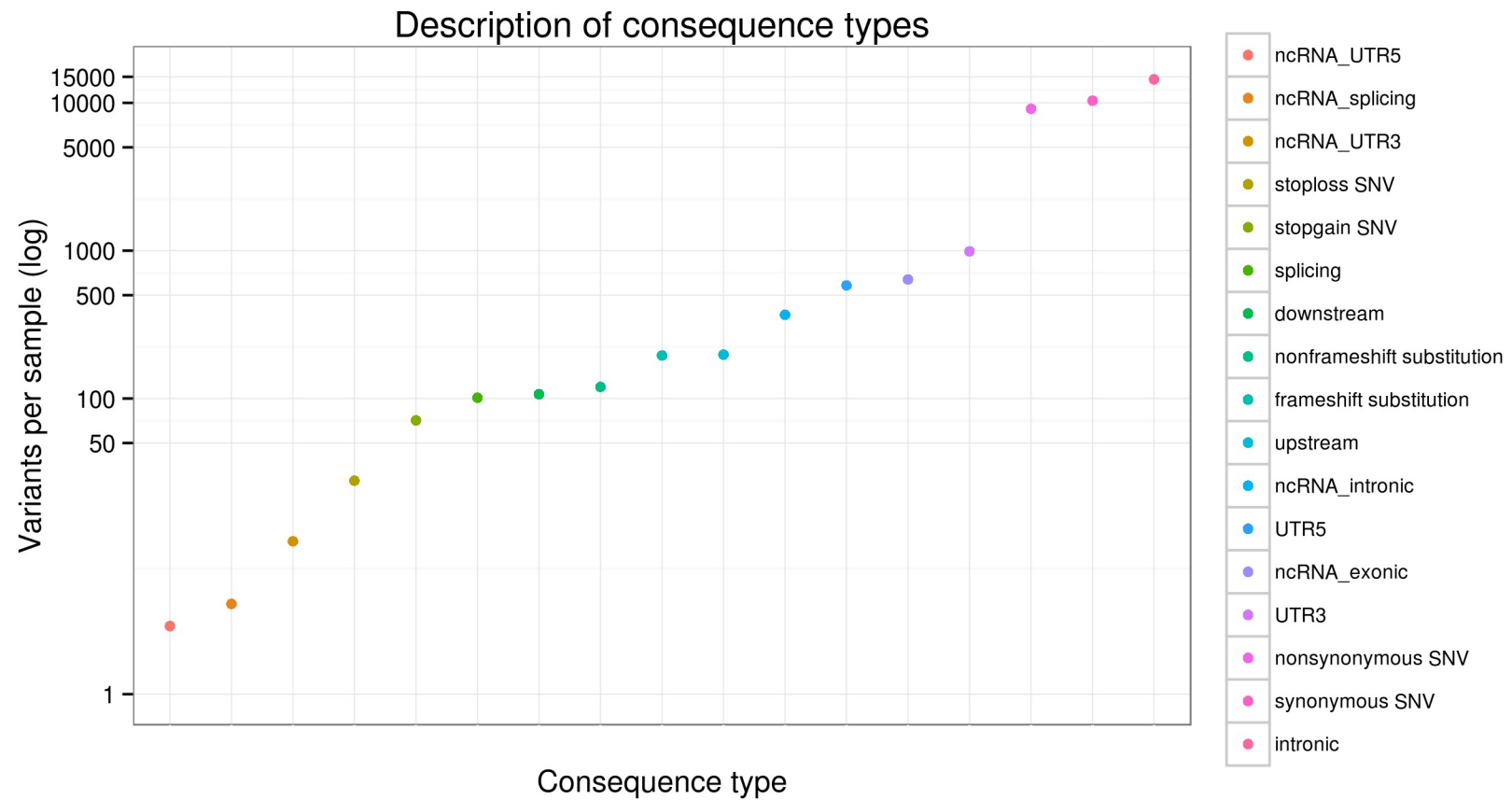
Some statistics: Homozygous variants per gene and sample



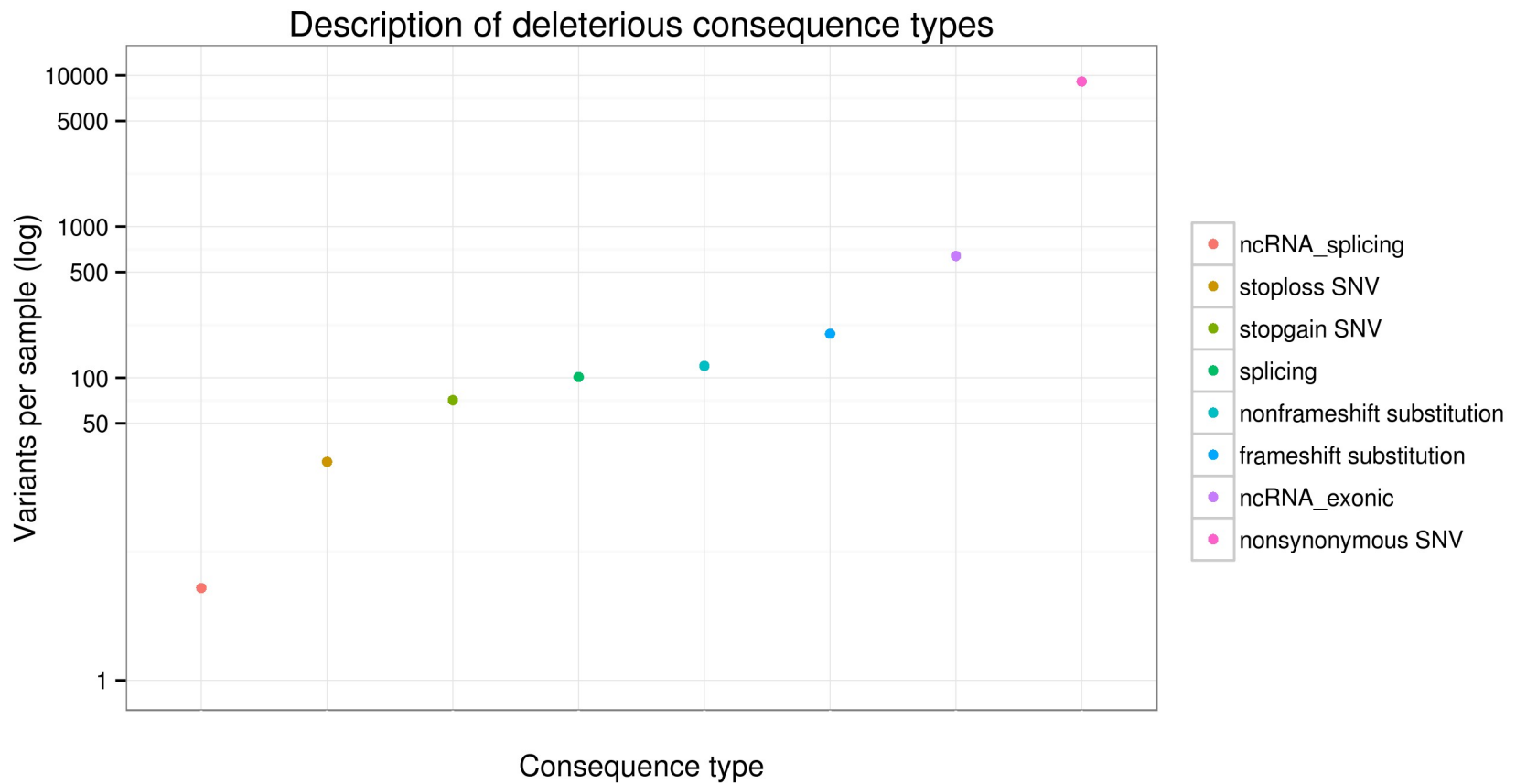
Some statistics: Heterozygous variants per gene and sample



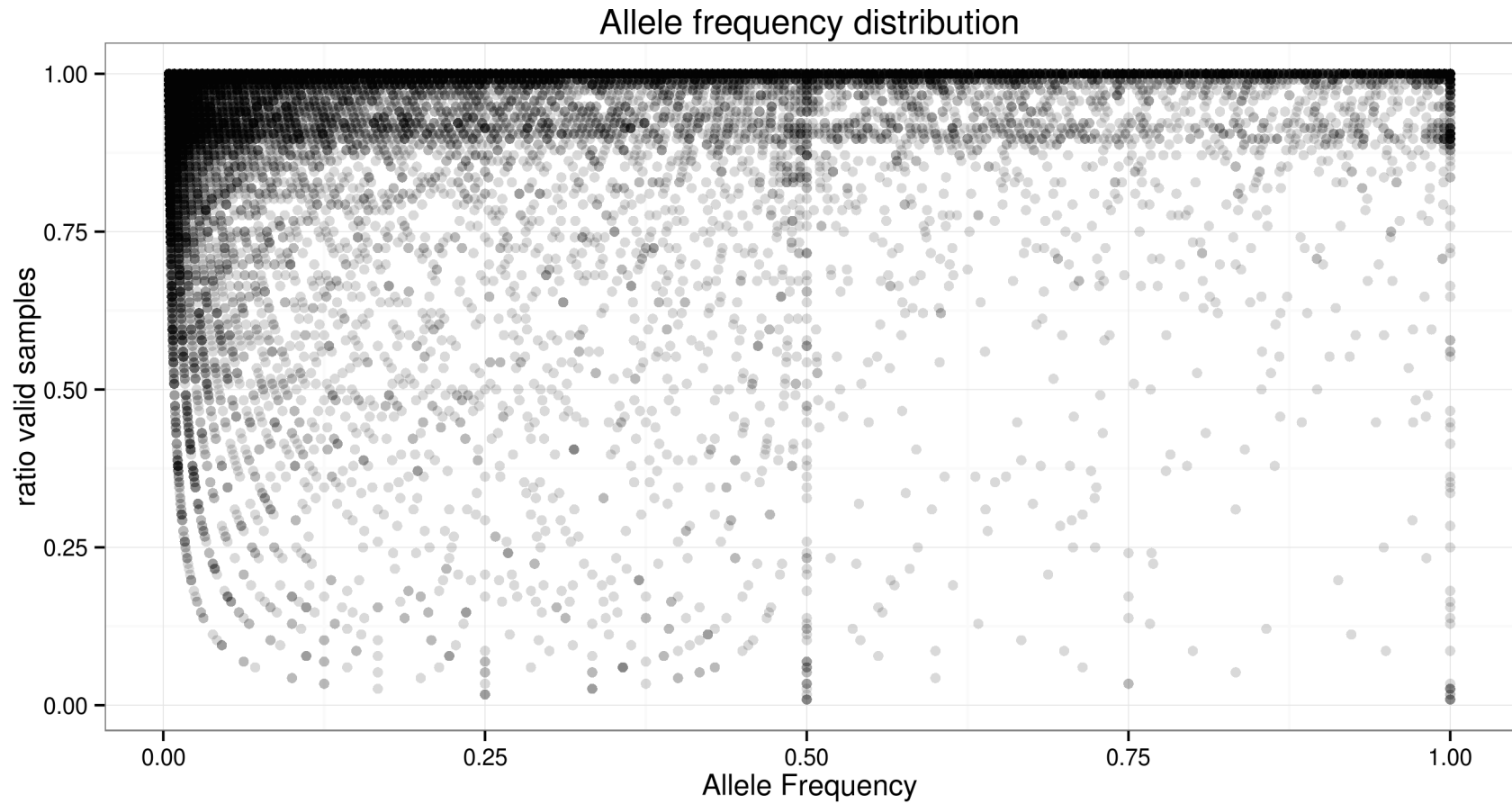
Some statistics



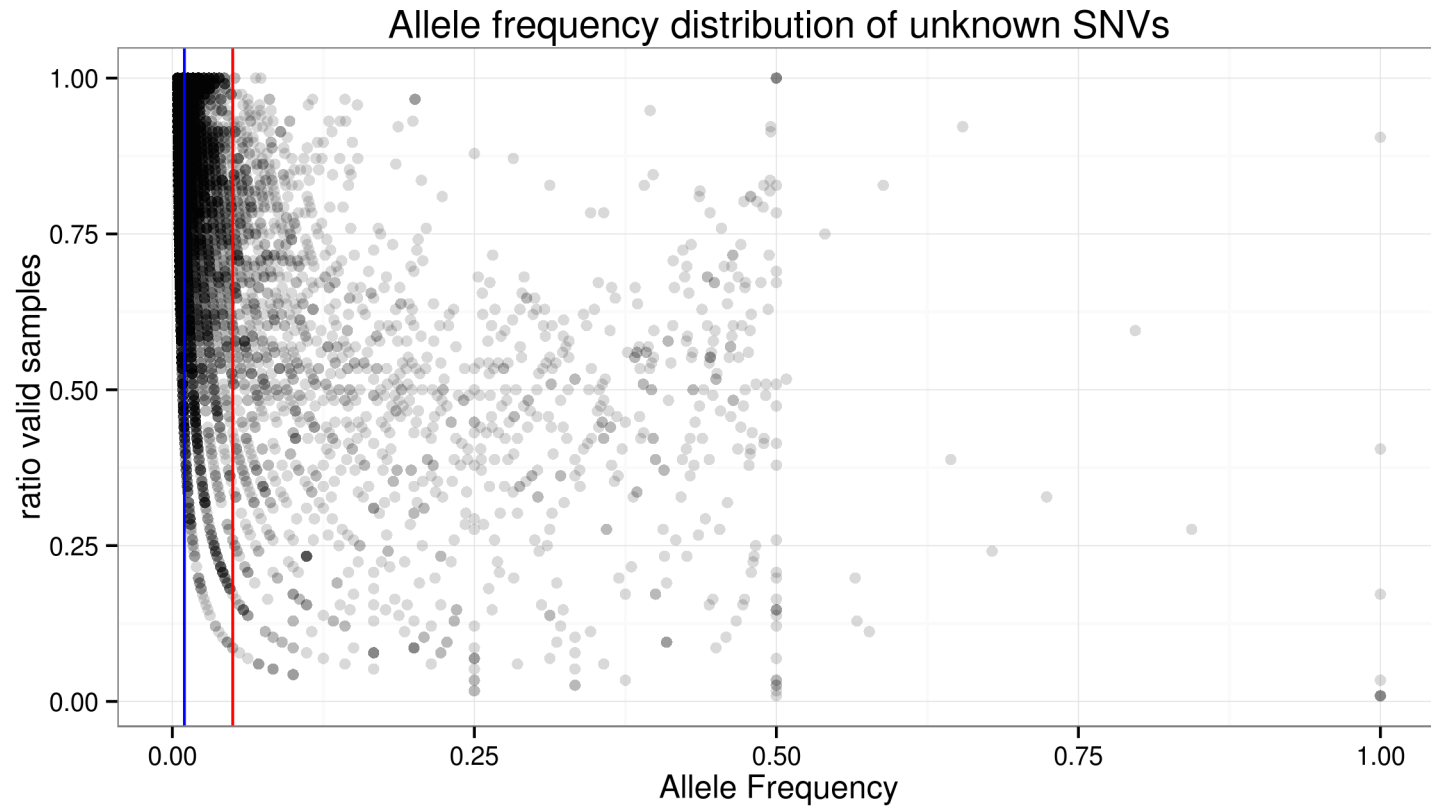
Some statistics



Some statistics



Some statistics



Questions?

