

Genomic data and bioinformatics

Jason Hodgson

6/11/2017

1 Data

DNA data is easy to display visualise.

Biggest problem at the moment is that there is so much data.

Up to 900 billion bases in a single experiment now.

Often presented in FASTA files:

```
!Taxa1
ATCGTAGCTACGTTTACCAGAAC
GGATCATTATTCTATATGCGGGA
etc.
```

FASTQ files:

DNA or RNA sequence data, includes a quality score.

VCF (variant call format)

Just variable sites mapped to a genome build. Meta data including some quality information.

PLINK

SNP genotype data. Either 2 or 3 files per a dataset, a genotype file, a family file and a marker file.

2 How to analyse the data

Common software for genomic analysis in R: Genetic packages (popgen etc).

Advantages: easy to integrate with other statistical analysis. Excellent plot-

ting capabilities.

Disadvantages: Extremely memory intensive. Often not possible with very large datasets. Slow.

Because of this lots of stand alone programmes.

PLINK: SNP data, designed for GWAS(genome wide association study) Powerful, fast, supports large datasets. Basic population genetics. Many models for testing genotype, phenotype associations.

Admixture: SNP or microsatellite data, model based method for inferring population structure and admixture proportions. Can handle large datasets.

Others: ALDER (admixture dating using LD), CHROMOPAINTER (local ancestry assignment), SAMTOOLS (NGS data), USEARCH (analysing metagenomic data), TREEMIX (inferring migrations) plus many more.

Work in UNIX - fast, simple and repeatable.

Script your analysis: automates it making it faster to run, makes it faster to repeat, makes a record of exactly what was done. REPEATABILITY.