ceu1kg: resources for exploring the 1000 genomes data on individuals of central European ancestry in Bioconductor

VJ Carey

August 10, 2010

1 Introduction

Using results of next generation sequencing experiments, a consortium of geneticists produced calls for SNP at approximately 8 million loci of the genomes of individuals of central European ancestry.

Genotype calls are held in a list of snp.matrix instances:

> library(ceu1kg)

```
Loading package ff 2.1-2
```

- getOption("fftempdir")=="/var/folders/4D/4DI98FkjGzq0K2niUTEHSE+++TM/-Tmp-//RtmpTQNdf
- getOption("ffextension")=="ff"
- getOption("ffdrop")==TRUE
- getOption("fffinonexit")==TRUE
- getOption("ffpagesize")==65536
- getOption("ffcaching")=="mmnoflush" -- consider "ffeachflush" if your system stalls
- getOption("ffbatchbytes")==16777216 -- consider a different value for tuning your sys Attaching package ff
- > if (!exists("ceu1KG.sml")) data(ceu1KG.sml)
- > names(ceu1KG.sml)
- [1] "chr1" "chr2" "chr3" "chr4" "chr5" "chr6" "chr7" "chr8" "chr9"
- [10] "chr10" "chr11" "chr12" "chr13" "chr14" "chr15" "chr16" "chr17" "chr18"
- [19] "chr19" "chr20" "chr21" "chr22"
- > ceu1KG.sml[[1]]

A snp.matrix with 60 rows and 605756 columns

Row names: NA06985 ... NA12874

Col names: chr1:533 ... chr1:247196267

Metadata about the loci are provided in GRanges instances:

```
> if (!exists("ceu1kgMeta_20")) data(ceu1kgMeta_20)
> ceu1kgMeta_20[1:3]
```

GRanges with 3 ranges and 5 elementMetadata values

chr20

	seqnames		ranges	strand	ref	alt	depth
	<rle></rle>	<ii< td=""><td>Ranges></td><td><rle></rle></td><td><character></character></td><td><character></character></td><td><character></character></td></ii<>	Ranges>	<rle></rle>	<character></character>	<character></character>	<character></character>
rs6078030	chr20	[9098,	9098]	*	C	T	192
rs4814683	chr20	[9795,	9795]	*	G	T	332
rs34147676	chr20	[10731,	10731]	*	C	Α	228
	ance	est all	LeleCnt				
	<characte< td=""><td>er> <char< td=""><td>cacter></td><td></td><td></td><td></td><td></td></char<></td></characte<>	er> <char< td=""><td>cacter></td><td></td><td></td><td></td><td></td></char<>	cacter>				
rs6078030		•	24				
rs4814683		•	40				
rs34147676			14				
seqlengths							

The Bioconductor *GGdata* package includes HapMap phase II genotypes on 90 CEU individuals in 30 trios, coupled with expression data as distributed at the Sanger GENEVAR project (ftp://ftp.sanger.ac.uk/pub/genevar/). The 1KG genotypes are available for 41 of these 90 and the associated genotype plus expression data for these 41 are stored in an 'smlSet' container:

```
> data(ceu1kg)
> ceu1kg

snp.matrix-based genotype set:
number of samples: 41
number of chromosomes present: 22
annotation: illuminaHumanv1.db
Expression data dims: 47293 x 41
Phenodata: An object of class "AnnotatedDataFrame"
    sampleNames: NA06985, NA06994, ..., NA12874 (41 total)
    varLabels and varMetadata description:
        famid: hapmap family id
        persid: hapmap person id
        ...: ...
        male: logical TRUE if male
        (7 total)
```

2 Imputation of 1KG genotypes to 19 'phase II only' CEU individuals

have been included in the