Summary Part 3

- Location => Look out for Feature
- Object creation from Adaptor or from API object
- No allele strings for structural variations, only coordinates
- VariationFeature already contains a lot of useful information to study consequences BUT:
  - Go further down to TranscriptVariation
    - Location specific information of Variant overlapping a Transcript
  - And TranscriptVariationAllele
    - Allele specific information on Transcript
Phenotype data

- **NEW** representation in API from e!71
- Annotation from different sources
  - EGA, NHGRI GWAS, OMIM, UniProt, HGMD, COSMIC
Exercise 5

• Find all phenotypes associated with the human SNP named rs12913832 and give:
  • Phenotype description, source and p-value
  • Find frequencies in 1000 Genomes populations for the (risk) allele associated with phenotype ‘Eye color’.

HINT:
• You might find the perl split function useful:
  my ($name, $allele) = split(‘-’, ‘rs9894429-T’);
• 1000 genomes populations start with: ‘1000GENOMES’