Assignment 3 / Biometry and bioinformatics I / 2013

Deadline for submitting: 31.12.2013

Topic: Human mitochondrial genome as a tool for tracing human population histories. Human mtDNA-database, http://www.mtdb.igp.uu.se/

- Collect ~5 mtDNA genome sequences from each continent, i.e. ~30 sequences in total

- Align the seqs and construct a UPGMA-tree (however: very soon, during Biometry and bioinformatics II, you get familiar with other phylogenetic methods and you may want to use one of them instead. Neighbor-joining, for example.)

- Interprete the tree by using the map below. The map is a story about human population histories, starting from Africa.

- Inspect your alignment and report what kind of polymorphisms there are and at what genes or other regions of the mitochondrial genome they are located. Note the facility *polymorphic sites* in http://www.mtdb.igp.uu.se/. However, if there are very very many polymorphic sites in your dataset, it is not necessary that you report them all. The idea is that you "learn to read information" (by using your eyes, "get a touch with a data and results") and it is enough that you report some of the polymorphic sites to show that you understand.

Mitochondrial genome of all mammals is a conserved entity: about 16 000bp long, 22 tRNA´s (transfer RNAs), two ribosomal genes (12S and 16S) 13 protein encoding genes, and the non-coding D-loop.

It is circle, but naturally linear as a database-sequence. But: not always cut from the same location. Most database seqs are such that they start from the D-loop and end at two tRNA-genes (tRNA-Thr and tRNA-Pro). Use only

this kind of seqs! Otherwise you run into practical problems! This mean that you should check each sequence from its description:

An example. The first seq in the database, EF064321.
After the general description, the start of seq details descrption looks like this:

```
D-loop          join(16024..16569,1..577)
     tRNA            578..648
                     /product="tRNA-Phe"
     rRNA            649..1602
                     /product="12S ribosomal RNA"
     tRNA            1603..1671
                     /product="tRNA-Val"
     rRNA            1672..3229
                     /product="16S ribosomal RNA"
     tRNA            3230..3304
                     /product="tRNA-Leu"
     gene            3307..4263
                     /gene="ND1"
     CDS             3307..4263
                     /gene="ND1"
                     /codon_start=1
                     /transl_table=2
                     /product="NADH dehydrogenase subunit 1"
.
.
```

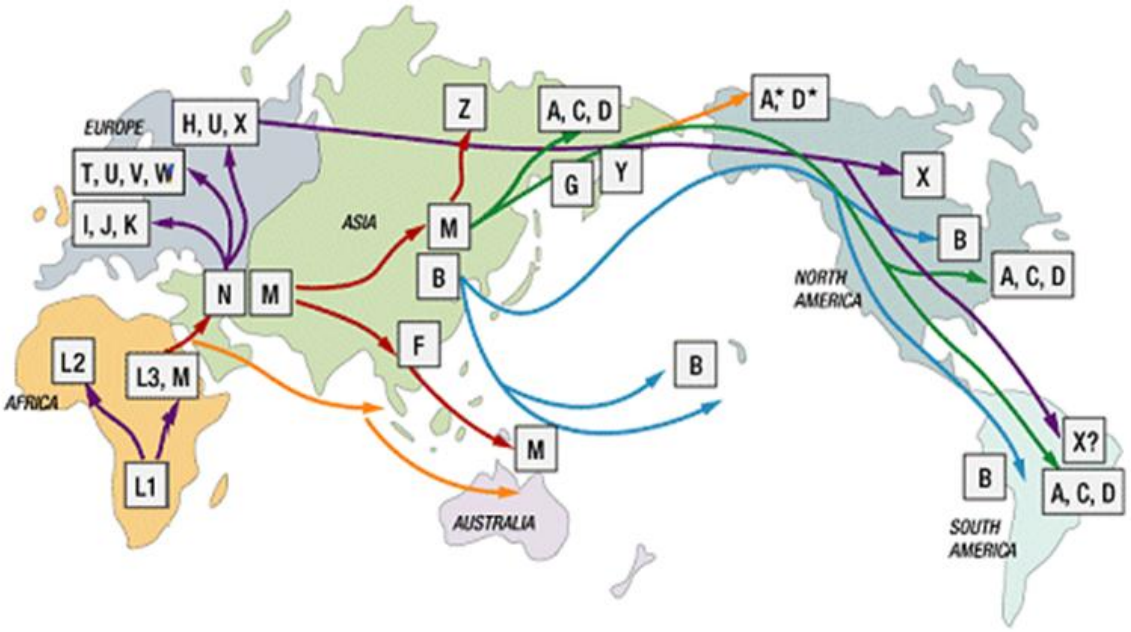
and ends like this:

```
TFHPYYTIKDALGLLLFLLSLMTLTLFSPDLLGDPDNYTLANPLNTPPHIKPEWYFLF

AYTILRSVPNKLGGVLALLLSILILAMIPILHMSKQQSMMFRPLSQSLYWLLAADLLI
                     LTWIGGQPVSYPFTIIGQVASVLYFTTILILMPTISLIENKMLK"
     tRNA            15888..15953
                     /product="tRNA-Thr"
     tRNA            complement(15955..16023)
                     /product="tRNA-Pro"
```

All seqs which you collect should have this configuration.

Alignment is very easy (there are probably only 1-3 gaps) but must be done. Recommendation is that you use the MAFFT-server because it is so fast. The sequences are rather long => Clustal quite slow. (Or try both, that will be a piece of learning useful practical issues.)



EXPANSION TIMES *(years ago)*	
Africa	120,000 - 150,000
Out of Africa	55,000 - 75,000
Asia	40,000 - 70,000
Australia/PNG	40,000 - 60,000
Europe	35,000 - 50,000
Americas	15,000 - 35,000
Na-Dene/Esk/Aleuts	8,000 - 10,000

FamilyTreeDNA

mtDNA Migrations Map