Key Area 8

- Genomic Sequencing

Vocabulary:

- Genomic sequencing
- Sequence data
- Bioinformatics
- Phylogenetics
- Molecular clock

Background Info:

- Human genome project ended in 2003
- Successfully sequenced 3 billion nucleotide bases
- Found that there are around 20'000 genes in the human genome

Genomic Sequencing

- Is a process where the exact order of the nucleotide bases along an organisms genome is determined.

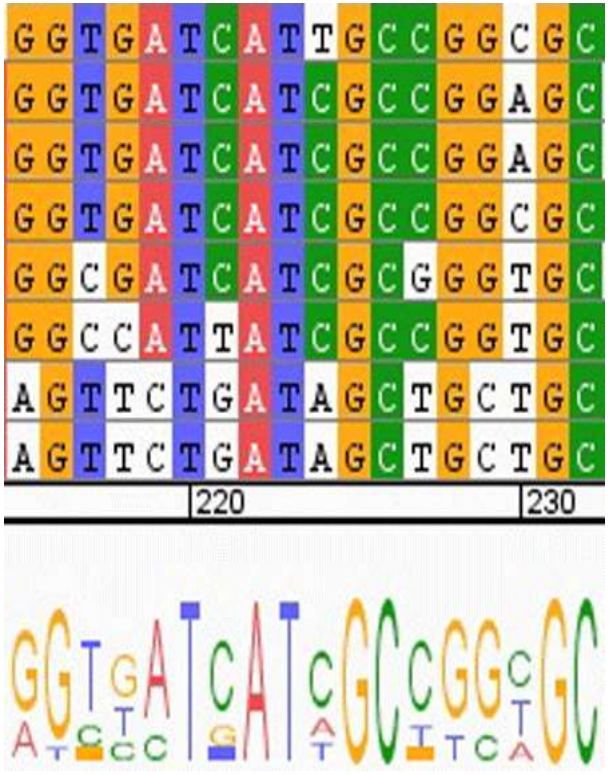
Useful:

- able to identify the genes and genomic sequences for particular diseases and conditions
- able to identify genomic mutations and variations

Sequence Data

Sequence of bases in nuceotides for:

1. genes
2. entire genome
- (all hereditory information in an individual's DNA)



More closely related species will have more similar nucleotides base sequences/genes.

Bioinformatics

The **analysis** of sequence data using

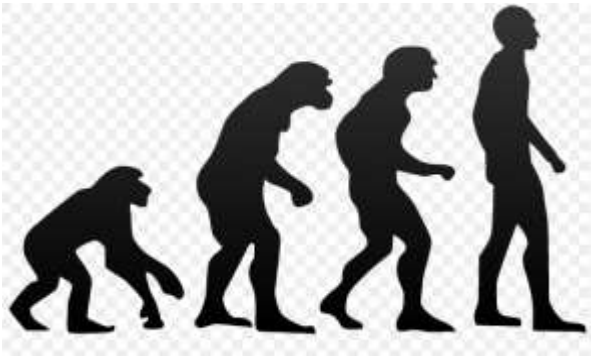
1. Computer

2. Statistical tests

Bioinformatics

- Two main uses
- 1. Evolution

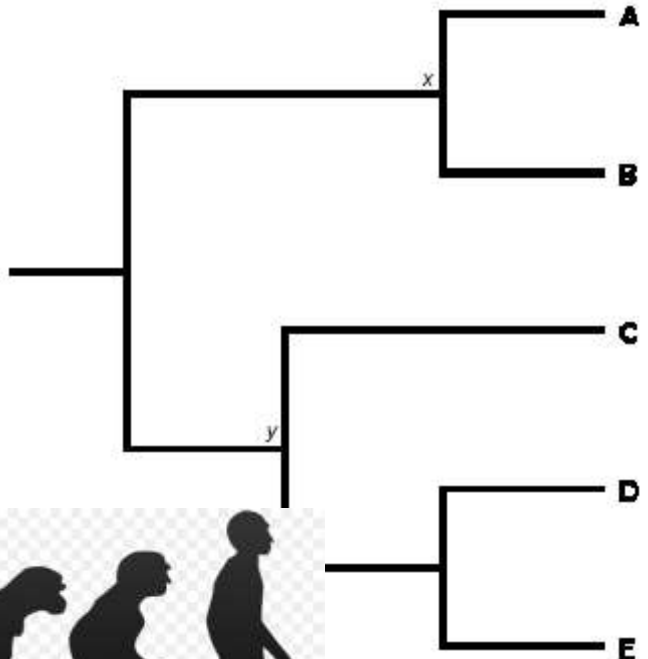
(relatedness of species)

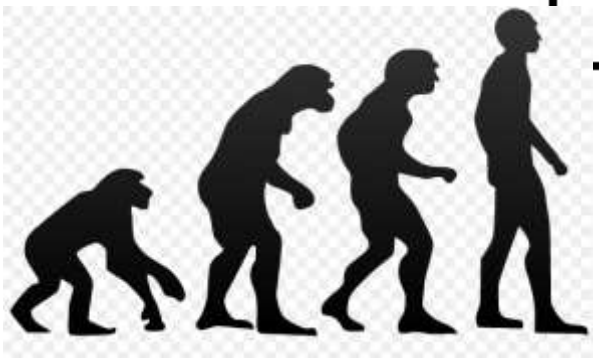


- 2 Personal genomics
- (analysing individual's sequence data)

Phylogenetics (evolution studies)

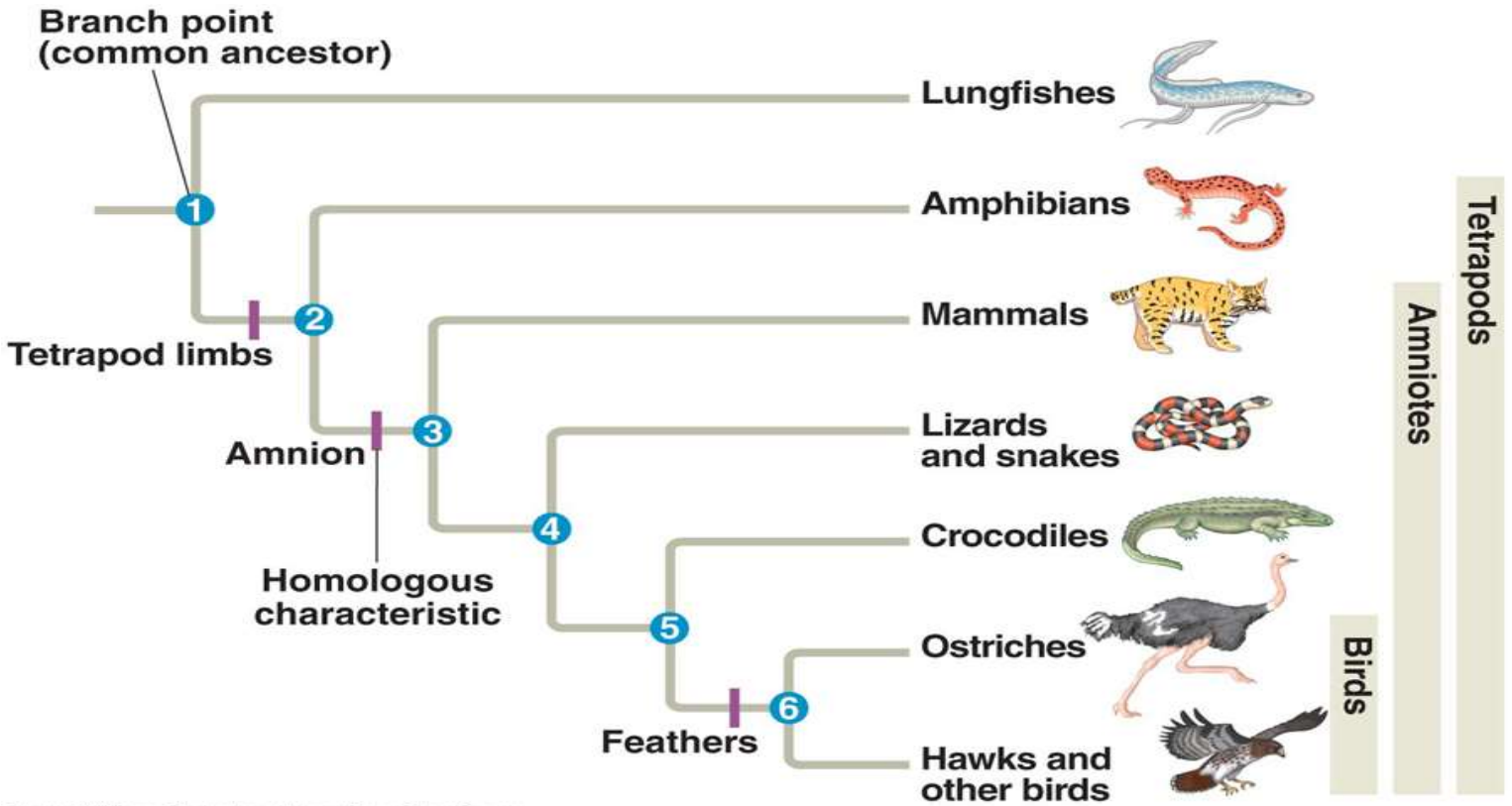
Sequence data & fossil data used to draw phylogenetic trees to compare divergence of lineage of related species.





Divergence of Lineage dependent on sequence data

- Used to determine time since divergence of lineage



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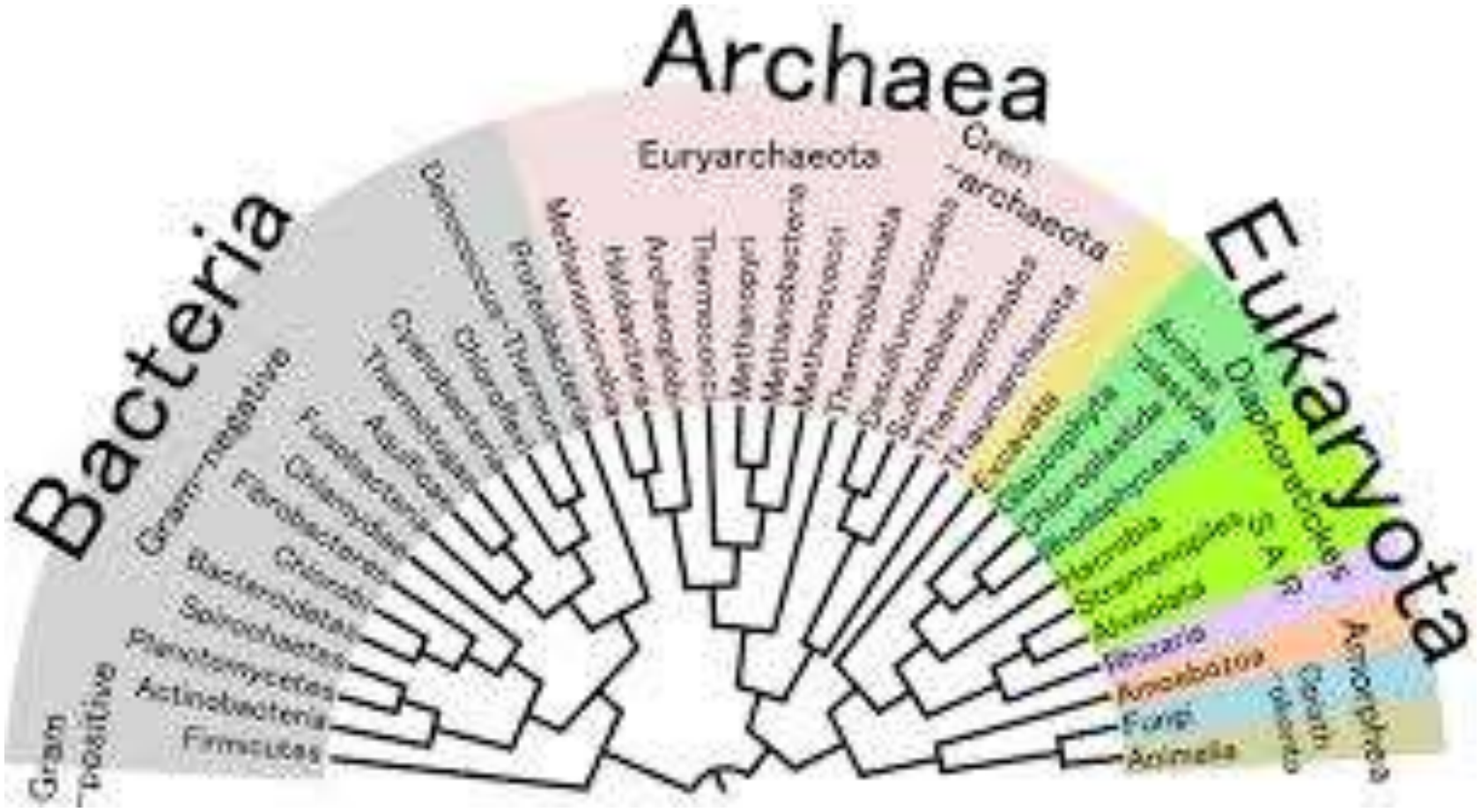
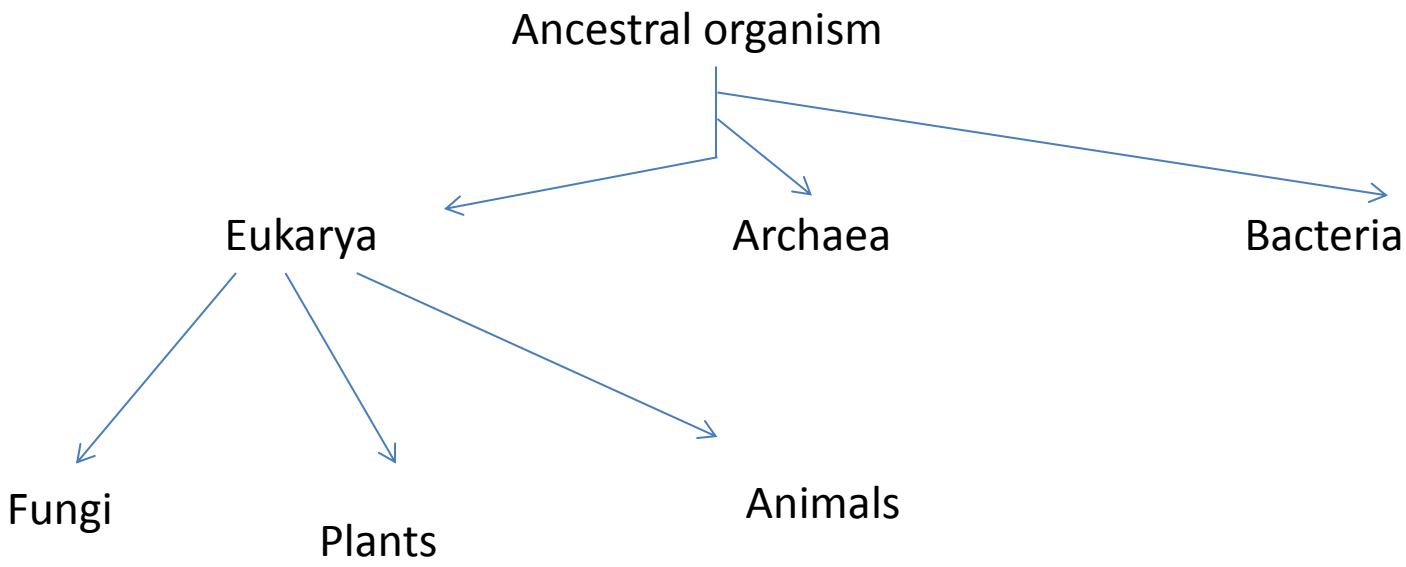
3 Domains of Life Today



3 Domains of Life Today

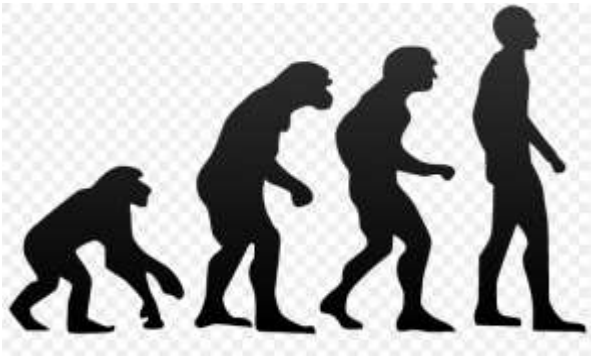


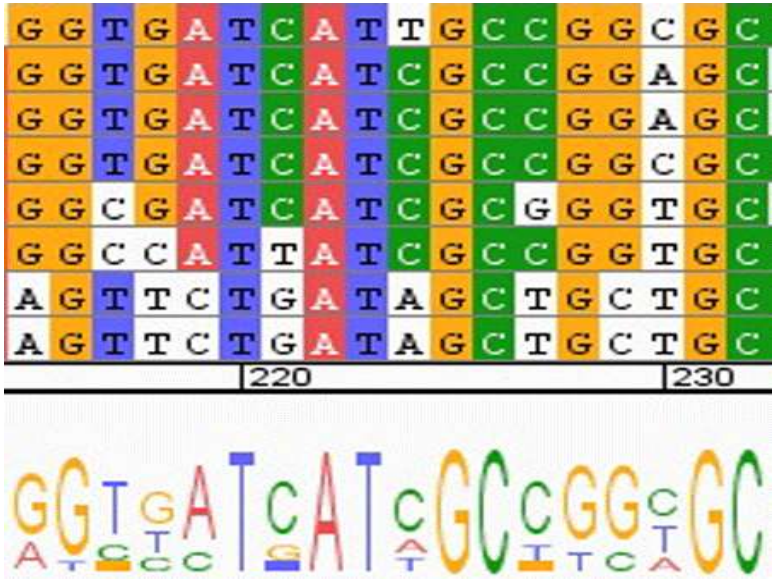
Evolution of life

- Common ancestor
- Photosynthetic organisms (cyanobacteria)
- Eukaryoates
- Multicellular organisms
- Animals
- Vertebrae animals
- Land plants

Comparing Genomes

- Comparing sequence data via computer and statistical tests suggests that across many closely related species, genes are found in a conserved state.

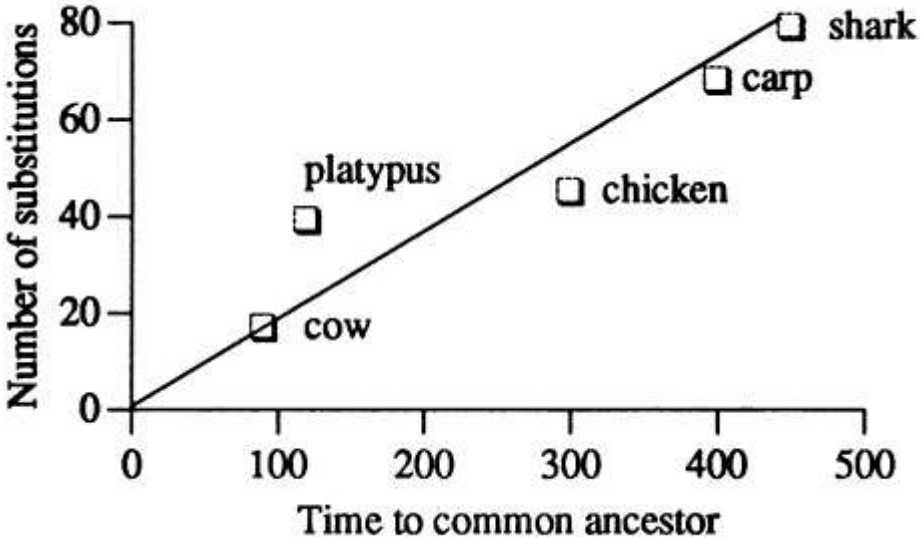




Molecular Clocks

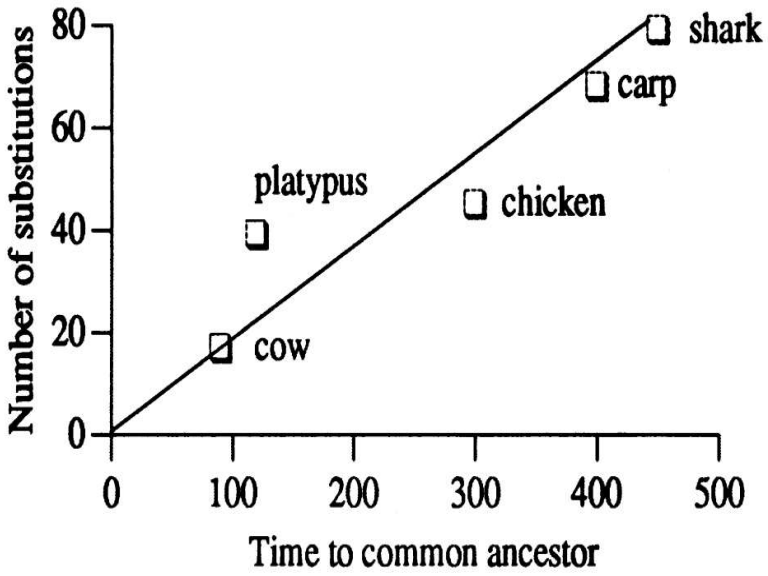
Graphs showing how many amino acids in a protein differ against time showing how related species are.

- Use sequence and fossil data



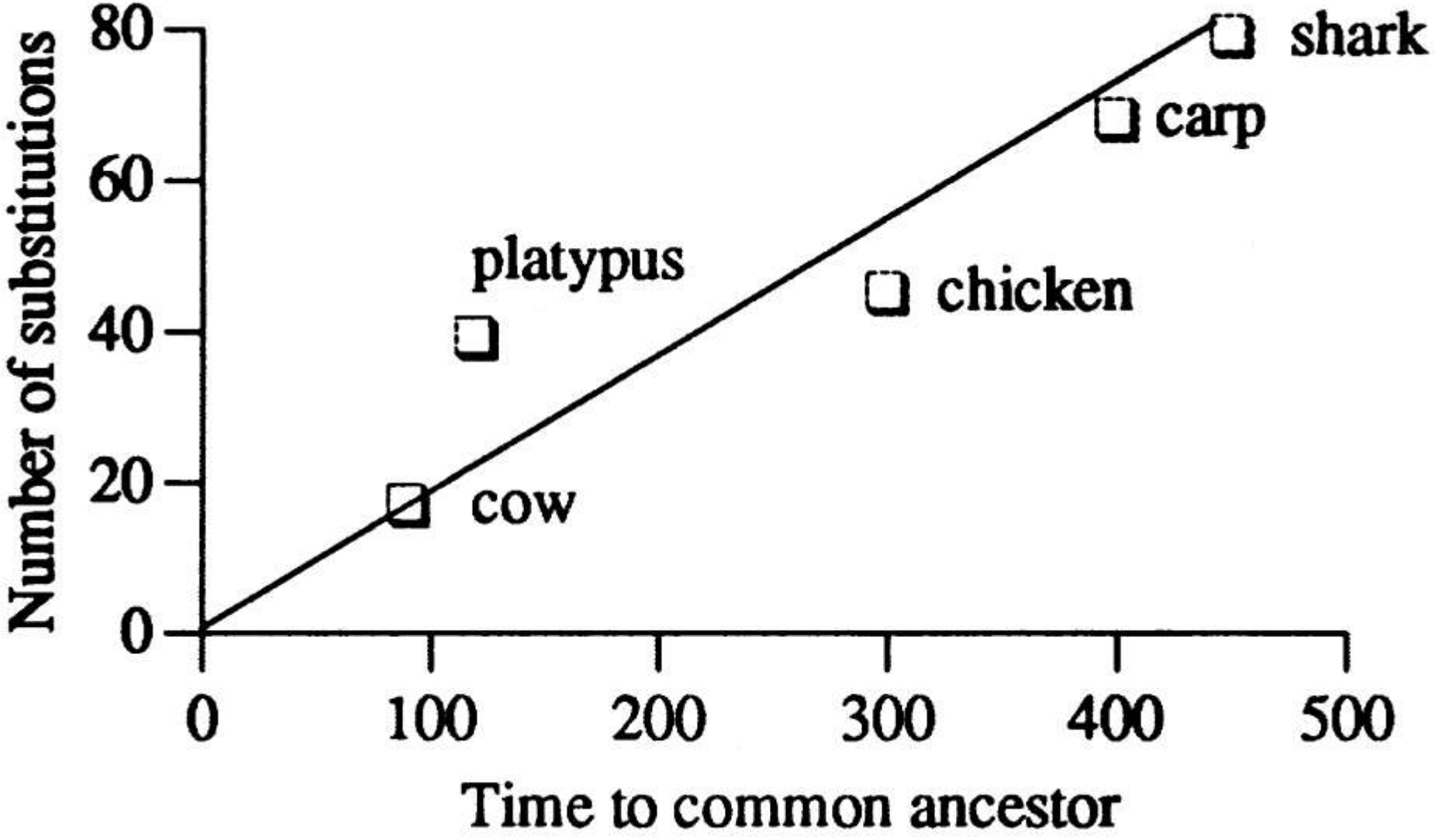
- This graph shows differences in the number of amino acids found in the protein haemoglobin against time.

Molecular Clock Graph



Y axis- number of Amino acid differences between species & humans

X axis – timescale since divergence of lineage since common ancestor


Number of substitutions
80
60
40
20
0
shark
carp
platypus
chicken
cow
0
100
200
300
400
500
Time to common ancestor

Personal Genomics

- The analysis of an ***individuals*** DNA (genome) by using sequence data via computer and statistical tests.

1. Pharmacogenetics

2. Drug trials

Personal Genomics (pharmacogenetics)

a) Pharmacogenetic drugs designed to best suit individuals with certain sequence data

Drug Trials

Patient groups with varied genotypes receiving the same diagnosis and the same prescription.

Group A
- Drug toxic but beneficial

Group B
- Drug toxic & not beneficial

Group C
- Drug not toxic & beneficial

Group D
- Drug not toxic & not beneficial

Drug Trial

Results mixed.

Worked for some & not others & caused toxic side effects to some.

<u>Solution</u>

- Pharmacogenetics

= Sequence data all subjects before starting

Problem with Pharmacogenetics

Difficulty in distinguishing between:

1. neutral &
2. harmful mutations

makes pharmacogenetics more difficult.