ERCC6 Gene & Cockayne Syndrome

by Zachary Beethem

https://www.emaze.com/@AFCWWORI/Cockayne

What is Cockayne Syndrome?

A rare, autosomal recessive, neurodegenerative disorder

Invariably fatal

Wide range of symptoms

Four types: I, II, III, and xeroderma pigmentosum (XP-CS)

Cockayne Syndrome Type II

Causative gene: ERCC6

Differences with I, III, XP: More severe, shorter life, congenital

http://bluepurpleandscarlett.com/about-cs

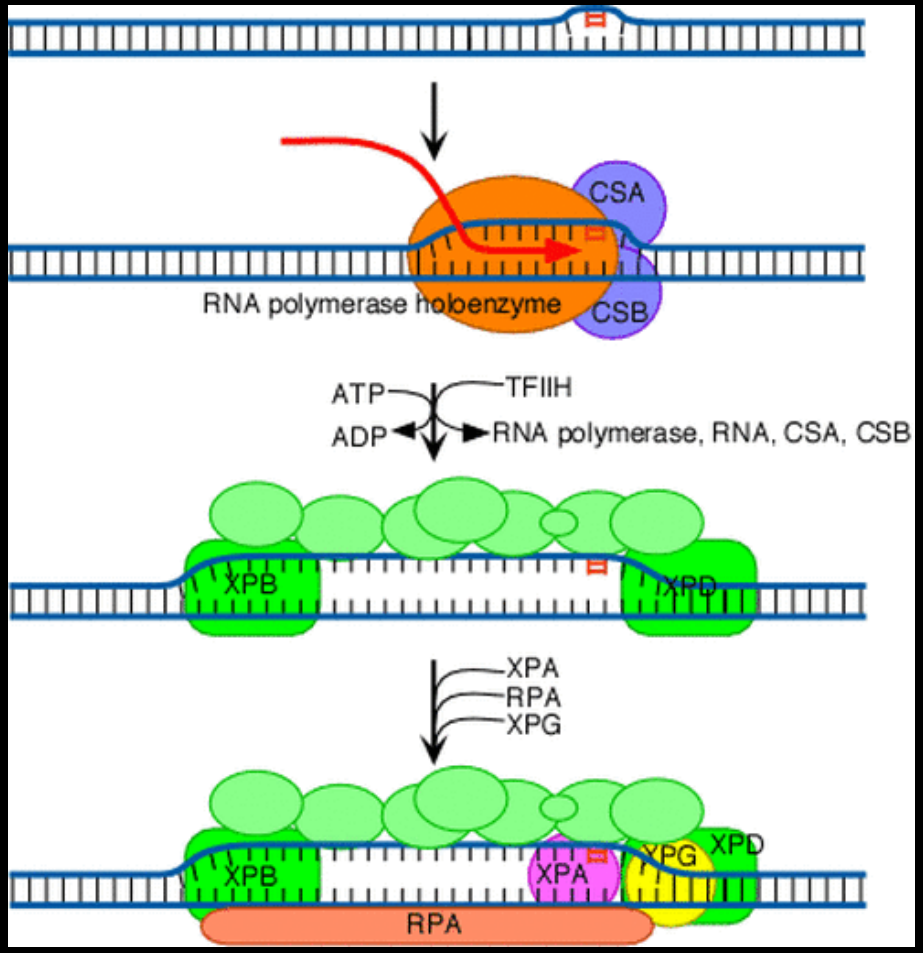
ERCC6/CSB protein

ERCC6 protein also called CSB

Transcription coupled nucleotide excision repair protein

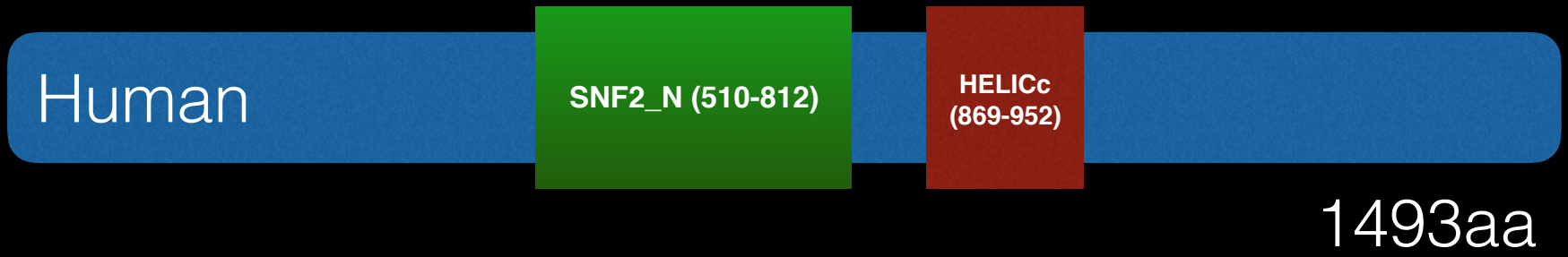
DNA lesion recognizer

Potentially involved in oxidative damage repair



http://www.acsu.buffalo.edu/~kowalsk/dnarepair/ner.html

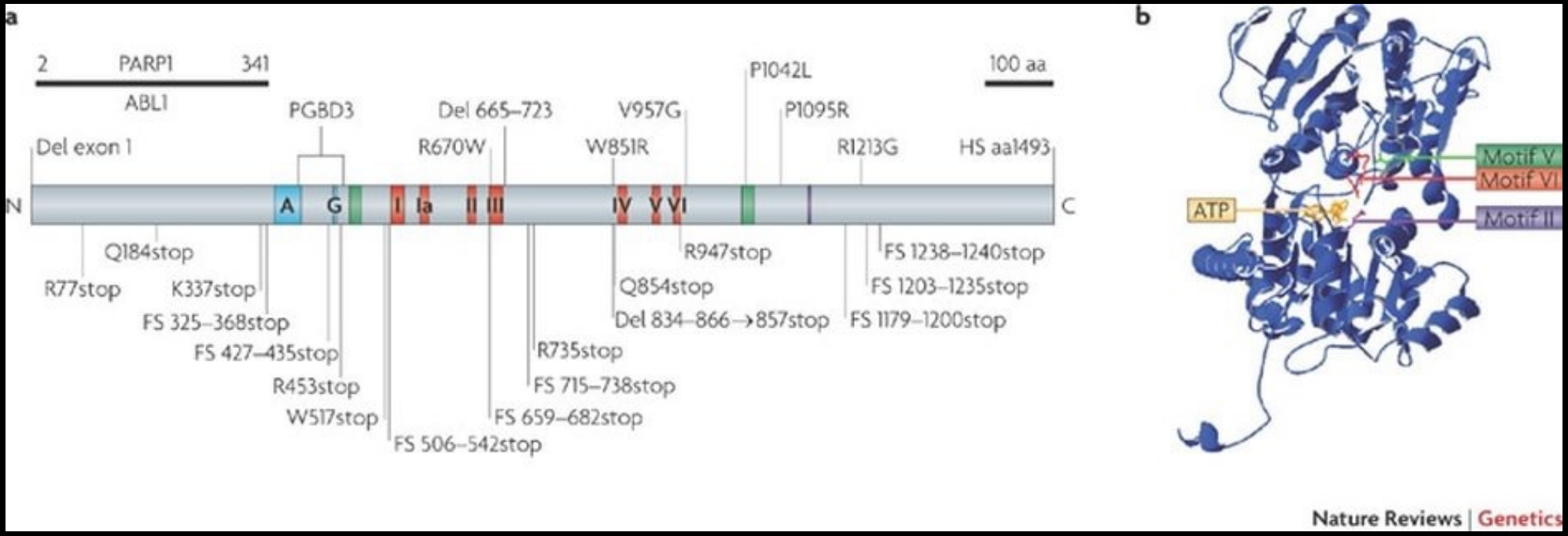
ERCC6 Gene



SNF2_N: Molecular function is to bind ATP. Proteins with this domain can disrupt histone-DNA interactions.

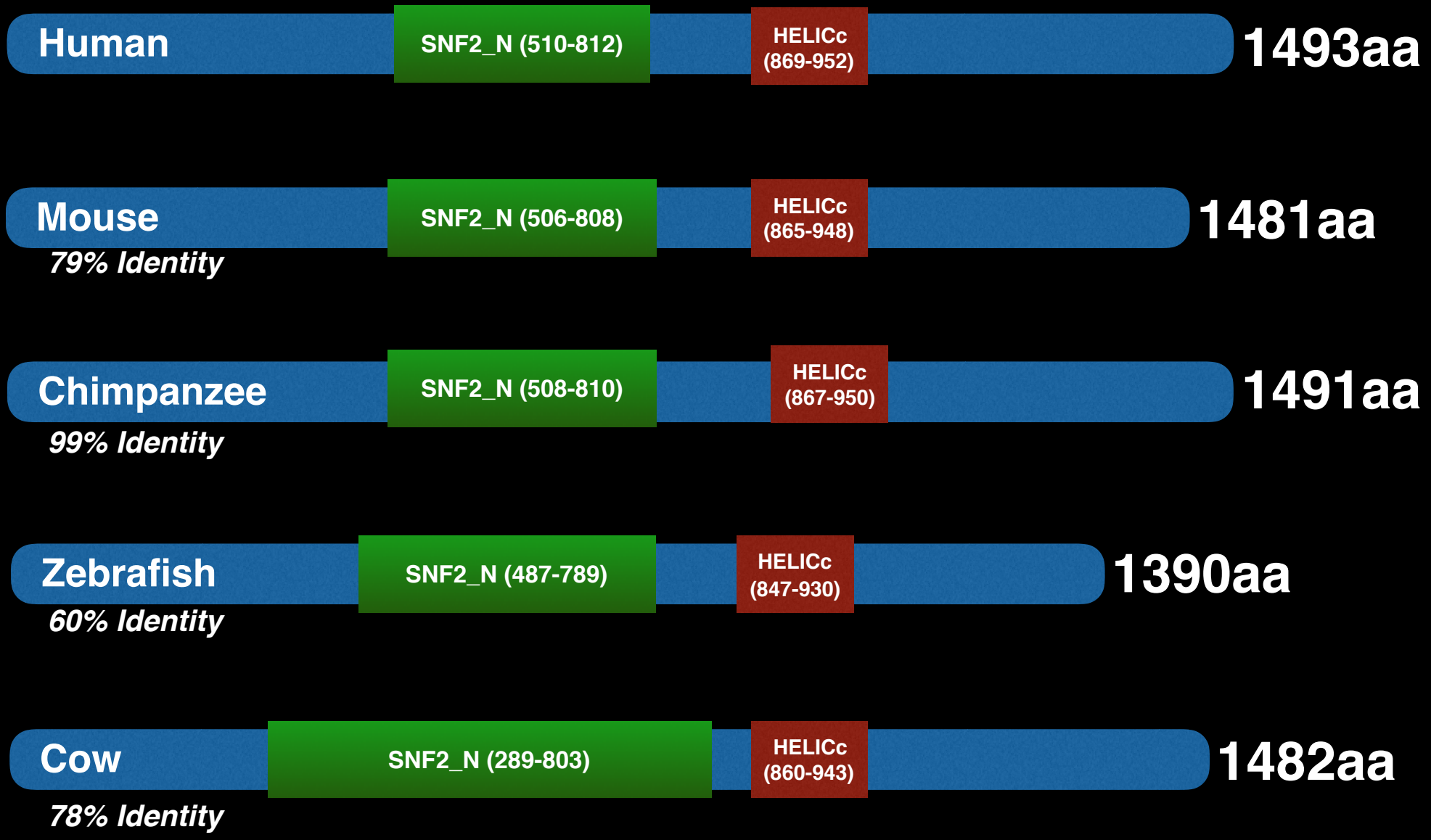
HELICc: C-terminal domain helicase. Molecular function is ATP binding and helicase activity.

http://www.ebi.ac.uk/interpro/entry/IPR000330
http://www.uniprot.org/uniprot/Q60ER9



http://www.nature.com/nrg/journal/v10/n11/fig_tab/nrg2663_F2.html

Domains and Identity in Other Organisms



Homology: Sequence Similarity

ERCC6 in 15 non-human species:

Orangutan, 97.98% identity

Gorilla, 99.13% identity

Chimpanzee, 99.46% identity

Mouse, 79.07% identity

Cow, 78.27% identity

Zebrafish, 59.86% identity

Rat, 78.97% identity

Wild Boar, 84.88% identity

Elephant, 82.19% identity

Panda, 84.61% identity

Sheep, 81.77% identity

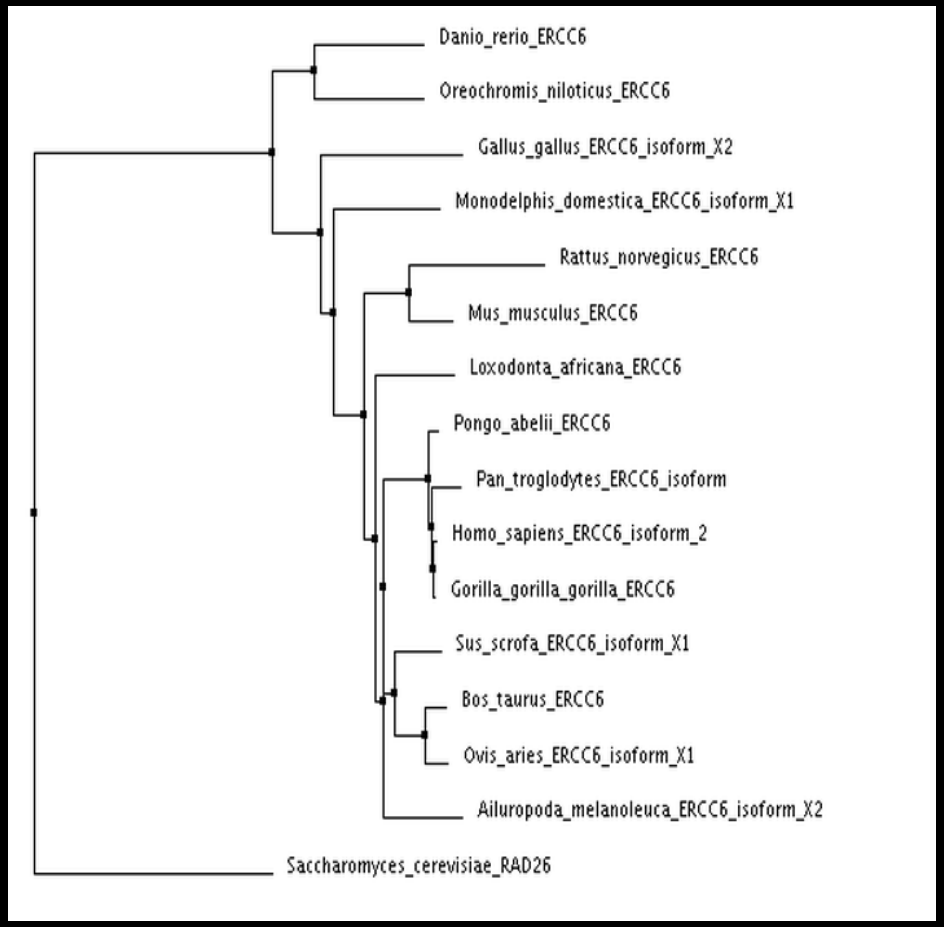
Red Junglefowl, 64.55% identity

Tilapia, 56.65% identity

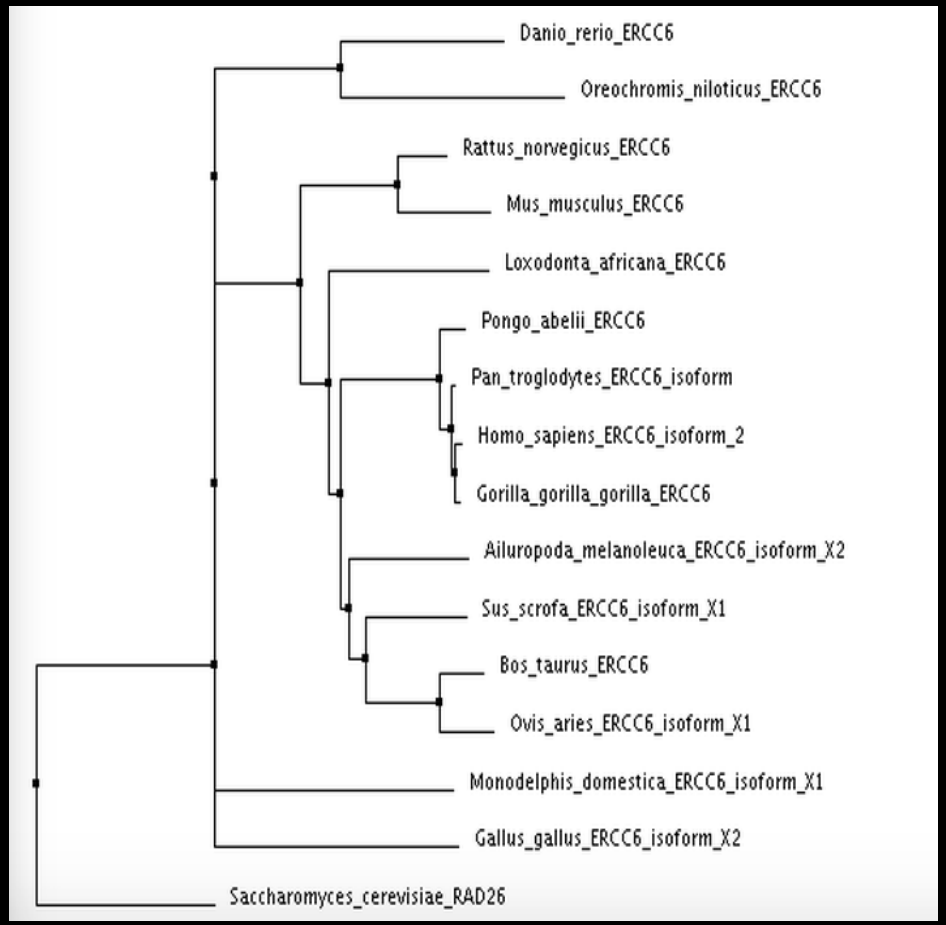
Possum, 69.24% identity

Saccharomyces cerevisiae, 38.52% identity

Phylogenetic Trees: Neighbor Joining

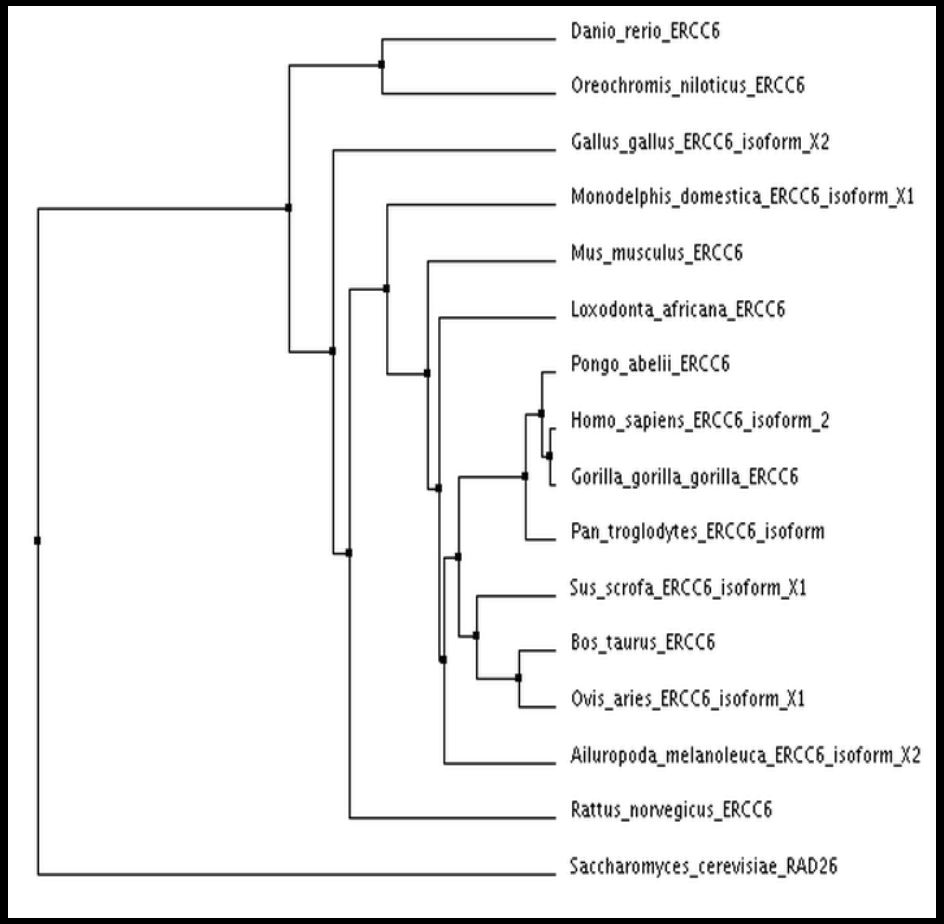


BLOSUM62 NJ

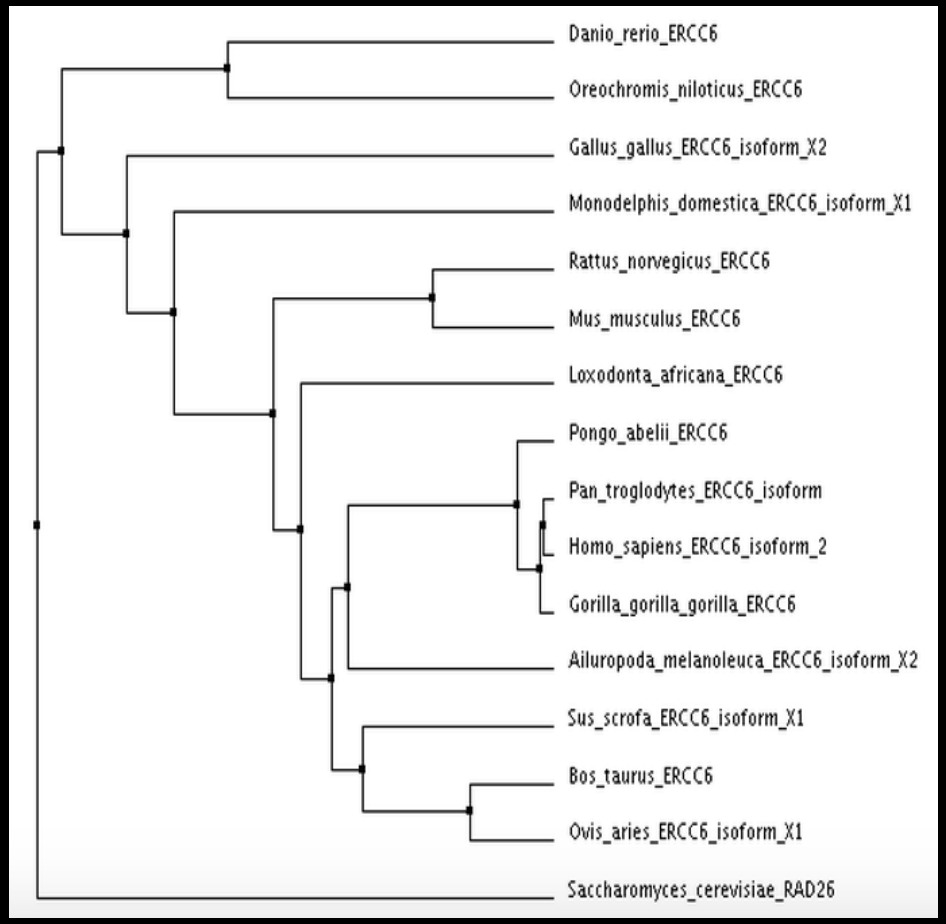


Percent Identity NJ

Phylogenetic Trees: Average Distance



BLOSUM62 Average Distance



Percent Identity Average Distance

Gap in Knowledge

A major symptom of CS is progeria, or premature aging. Aging is caused by oxidative damage. CSB is thought to play a role in processing and possibly fixing oxidative damage. *What is this role?*

Aim 1: To determine XXX

Approach:

Hypothesis:

Rationale:

Aim 2: To determine XXX

Approach:

Hypothesis:

Rationale:

Aim 3: To determine XXX

Approach:

Hypothesis:

Rationale:

References