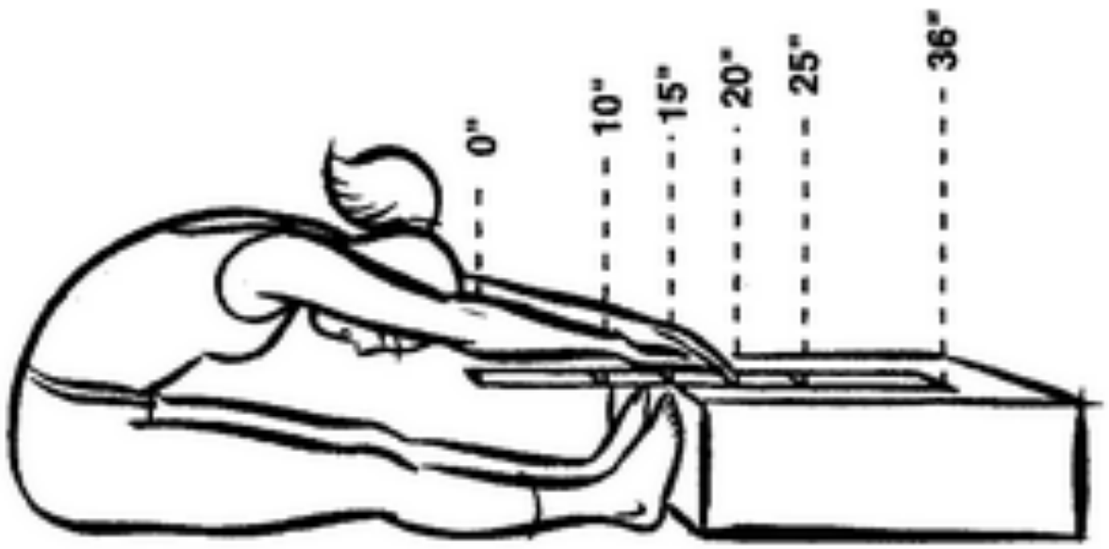
COL5A1: Gene for improved long distance running

By: Logan Silber

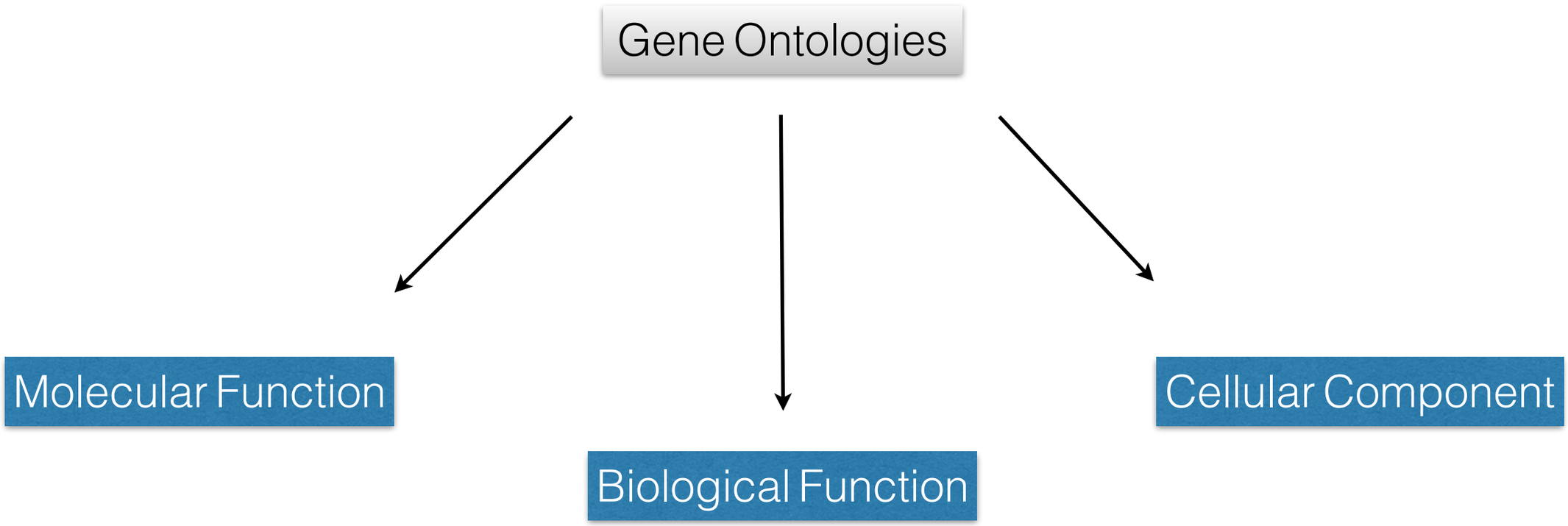
SNP of COL5A1: Rs12722

Reduced flexibility

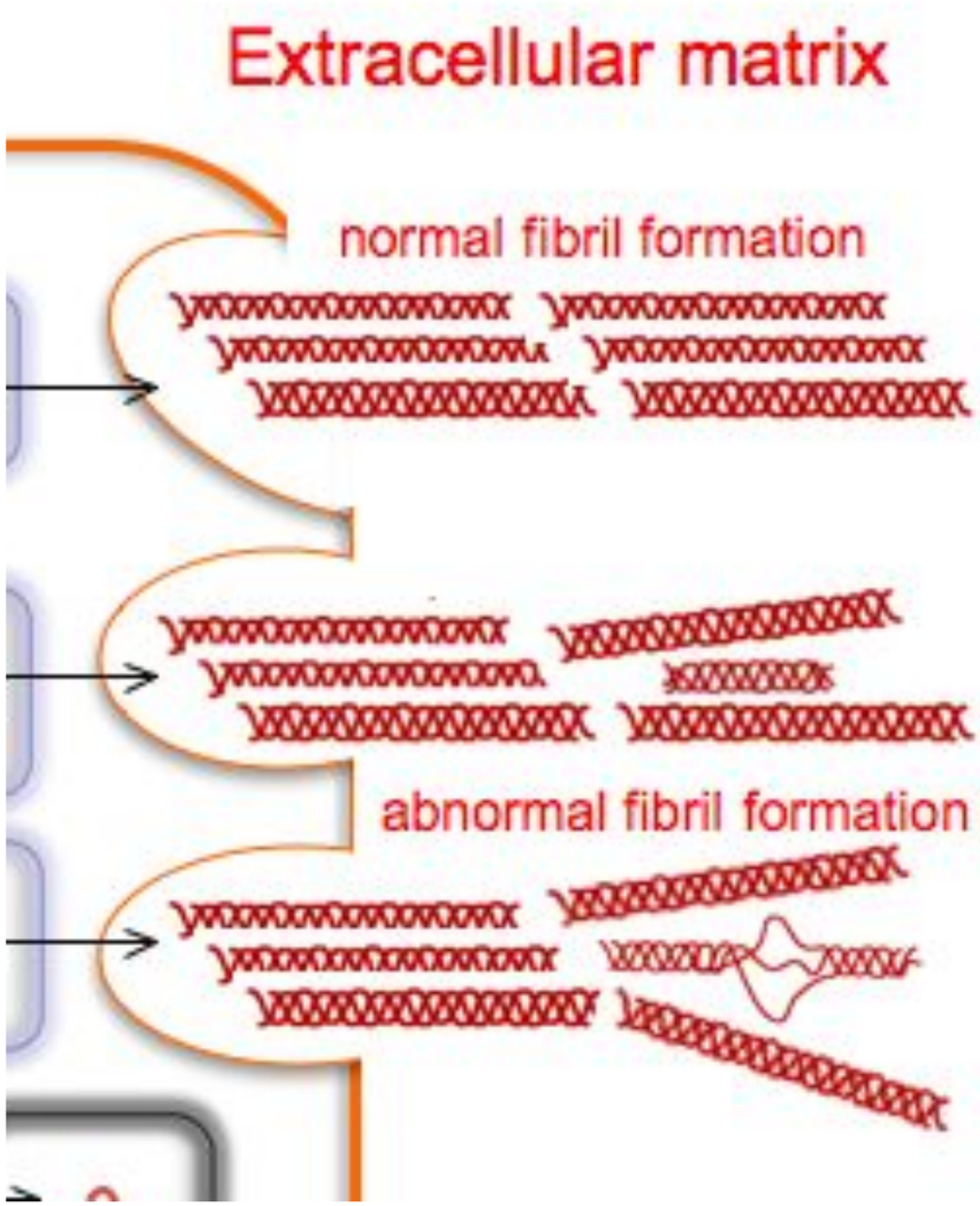


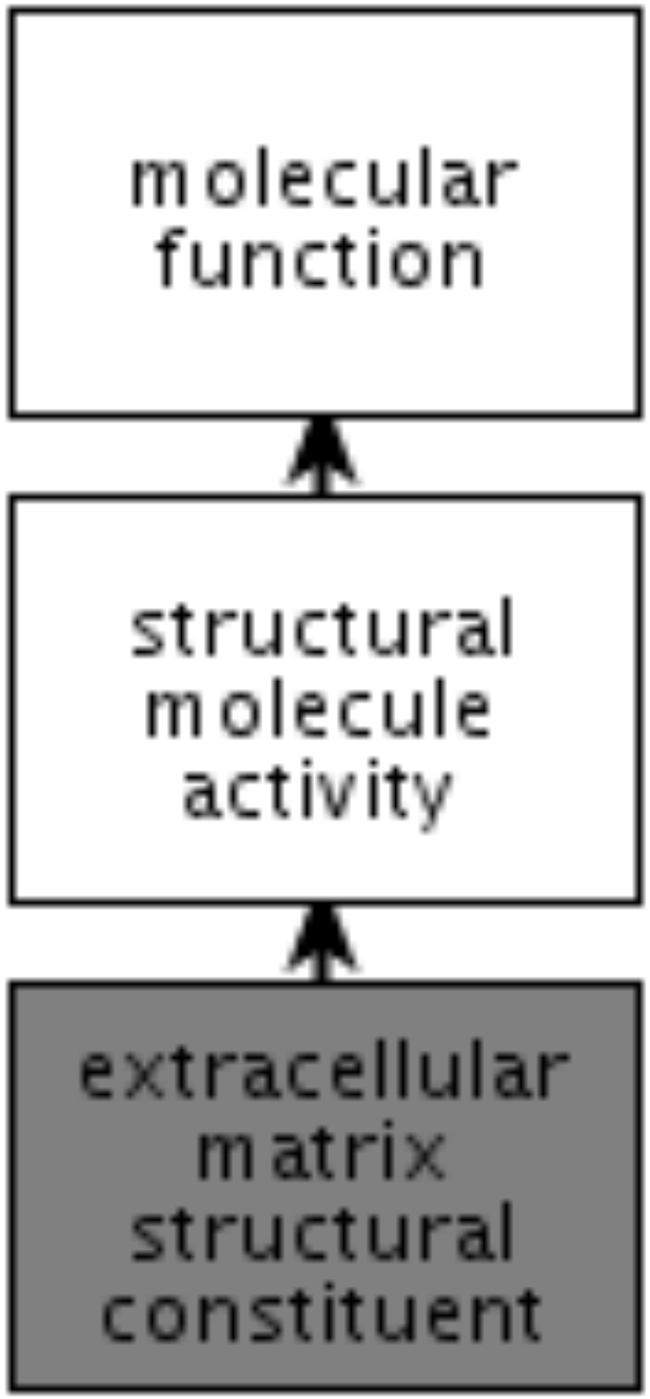
Enhanced running economy

What gene is mutated in my trait?

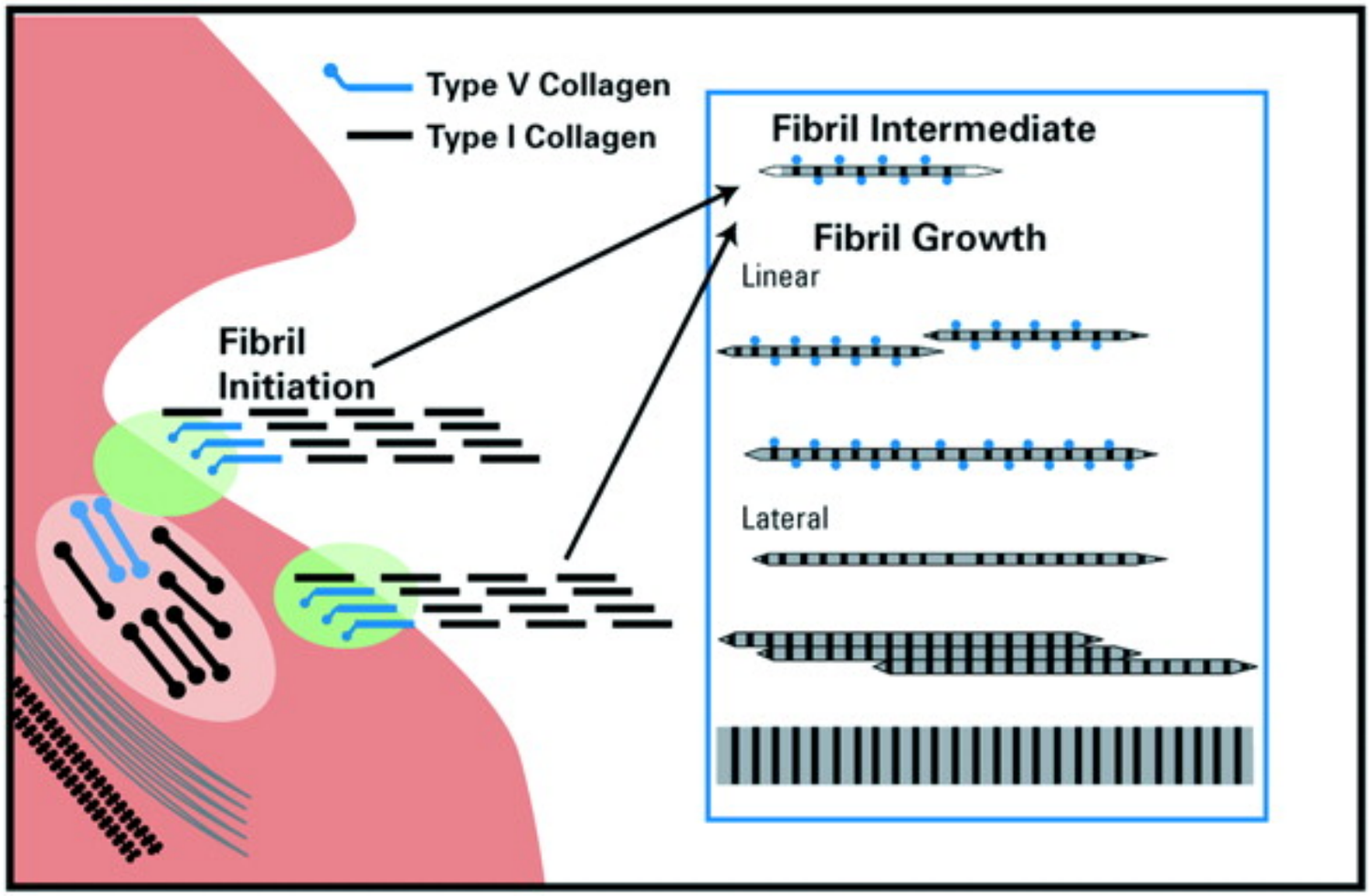


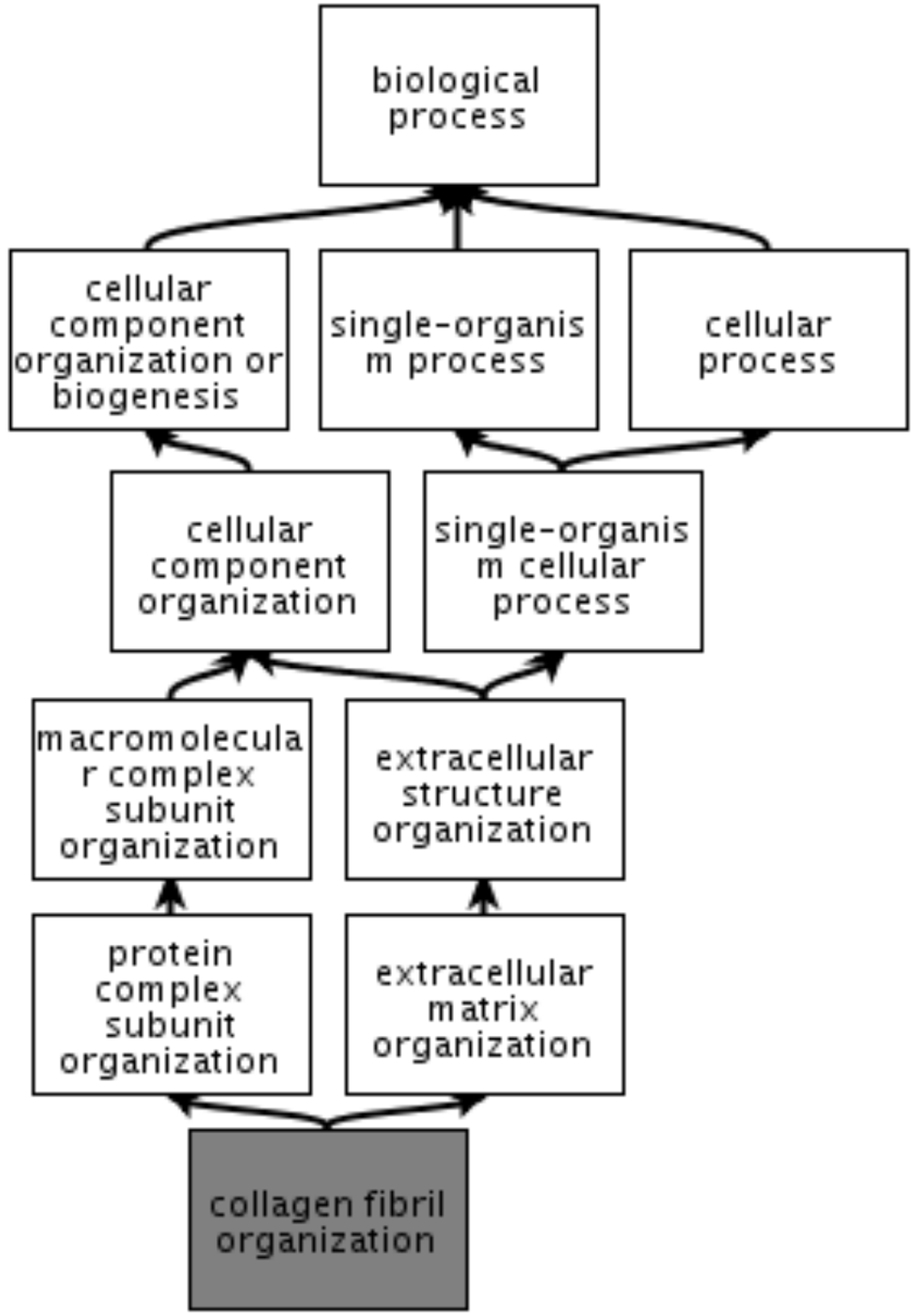
What is COL5A1's molecular function?





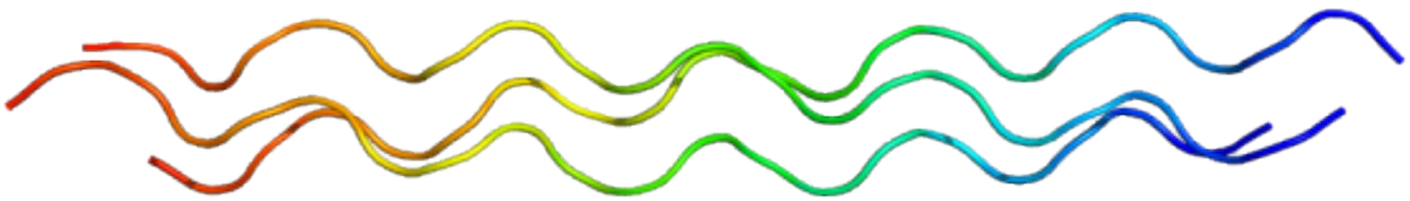
What is COL5A1's biological function?

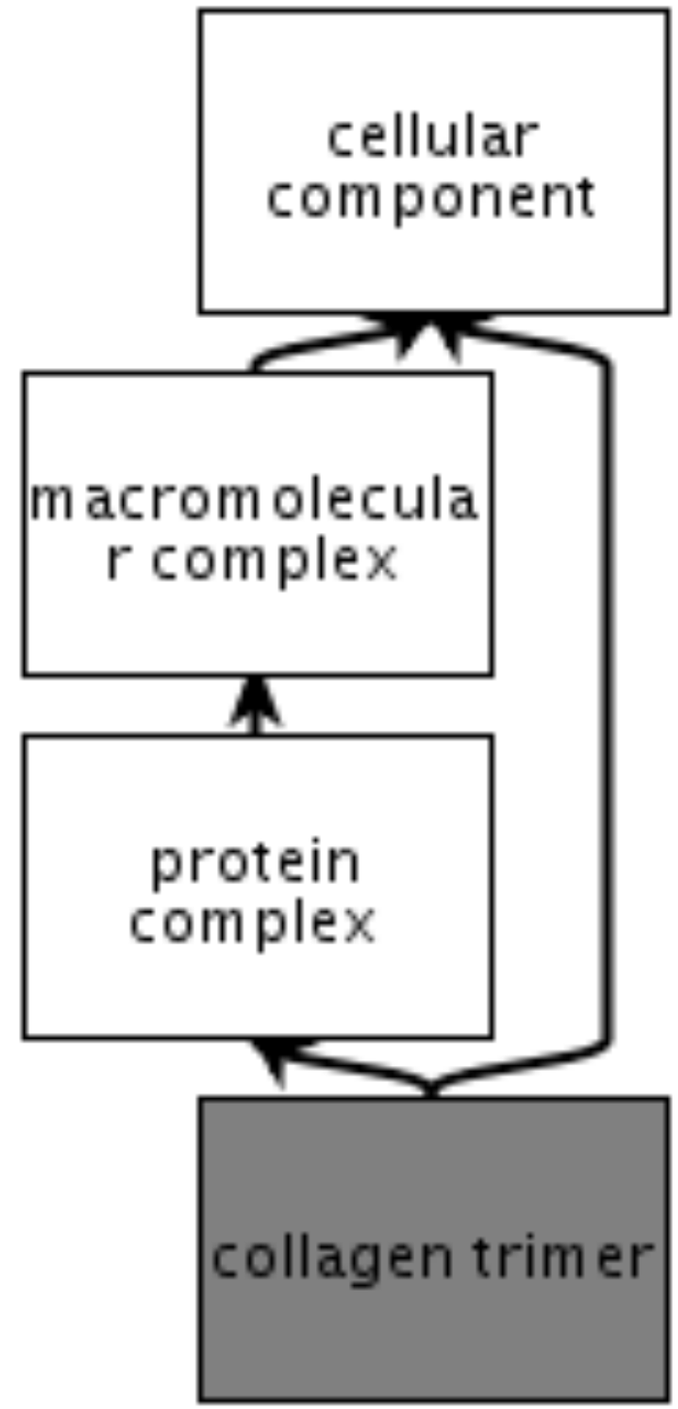




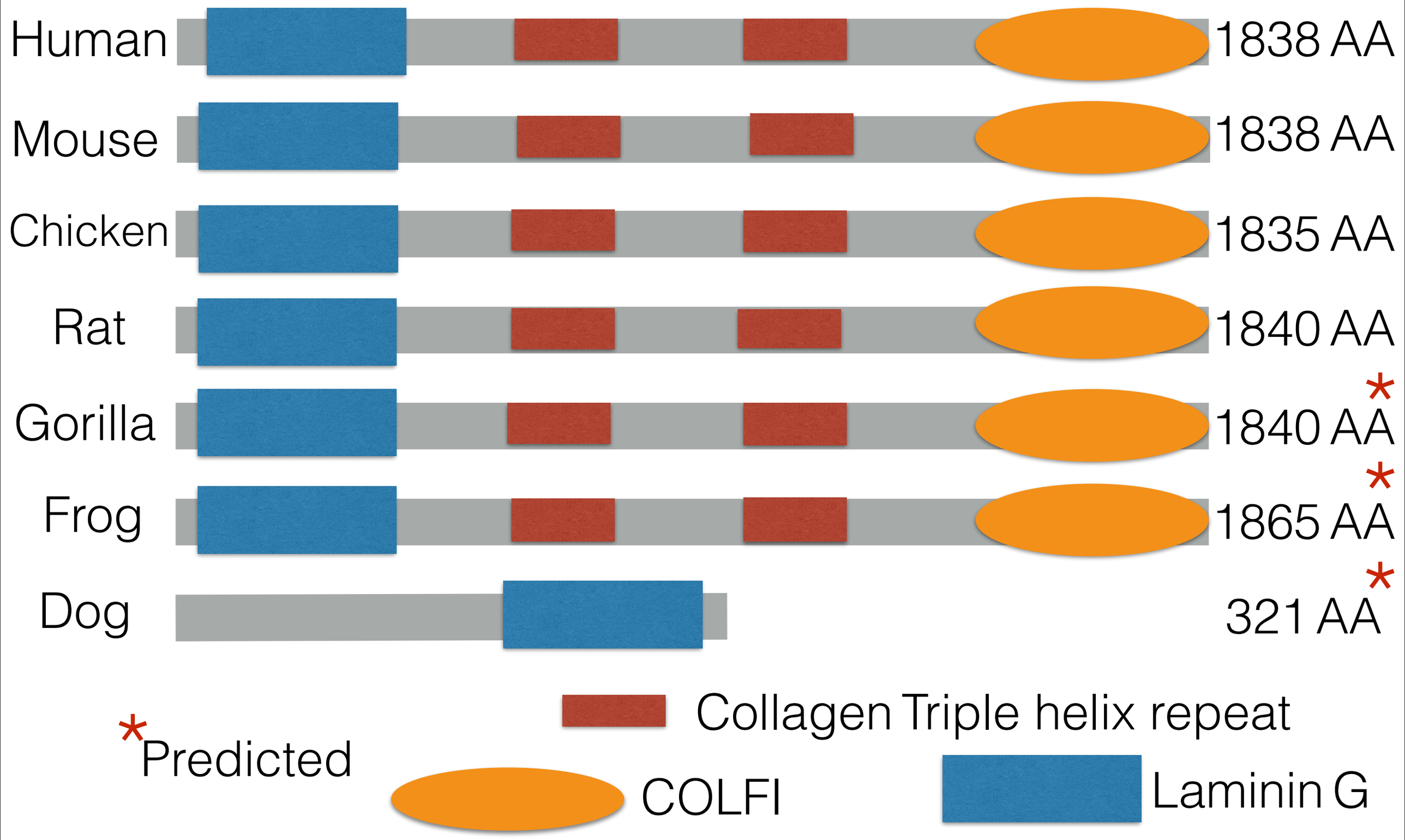
What is COL5A1's cellular component?

Subcomponent of type V collagen

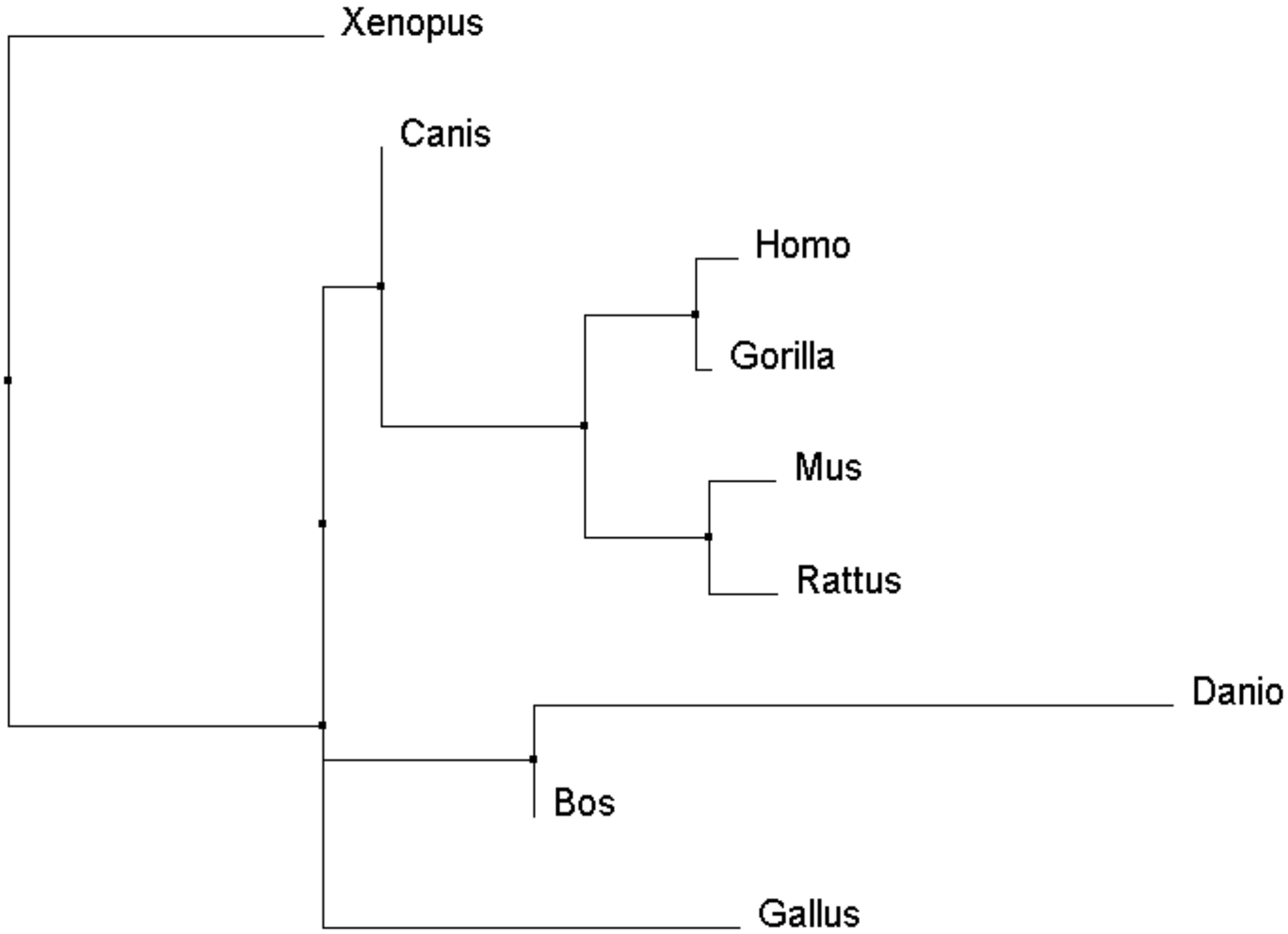




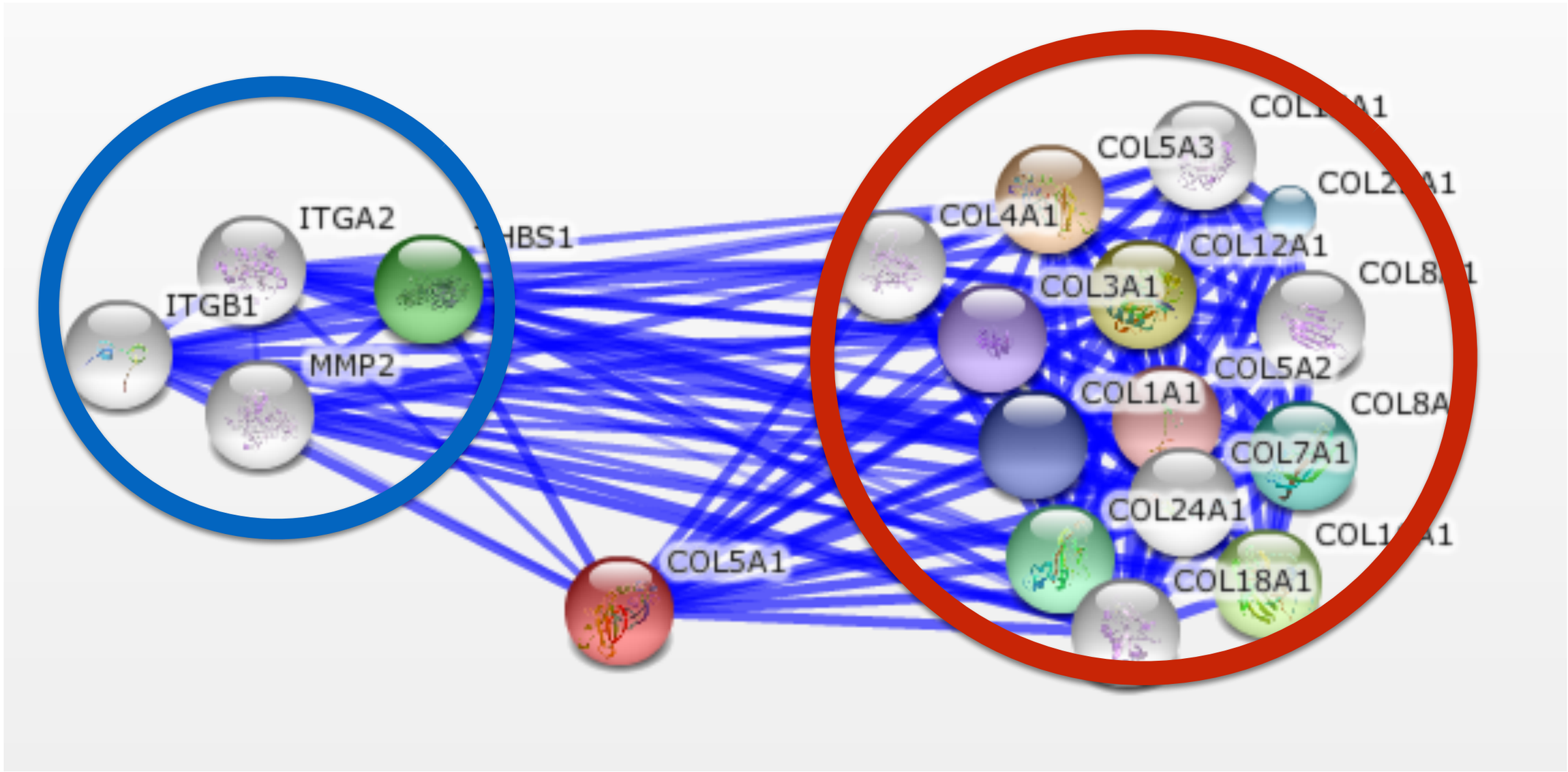
How well conserved is my protein amongst other organisms?



Phylogeny of gene amongst other organisms



COL5A1 product interacts with proteins needed for collagen fibril synthesis



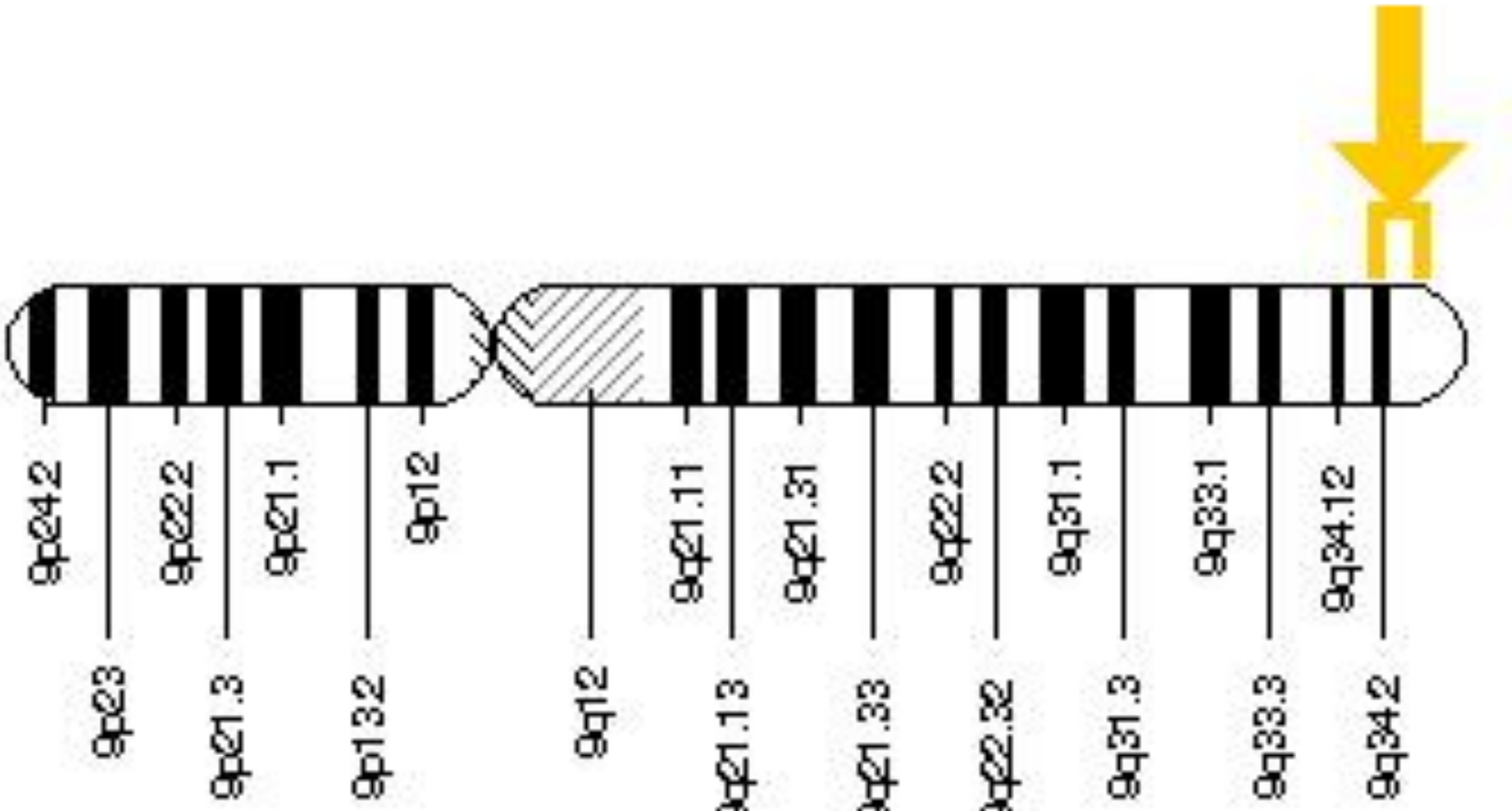
Gap in knowledge

How the COL5A1 mutation, rs12722, affects cellular functionality to produce an enhanced running economy

Hypothesis

Type V collagen produced by rs12722 enhances running economy through formation of more durable collagen fibrils and a more stable interaction with type I collagen.

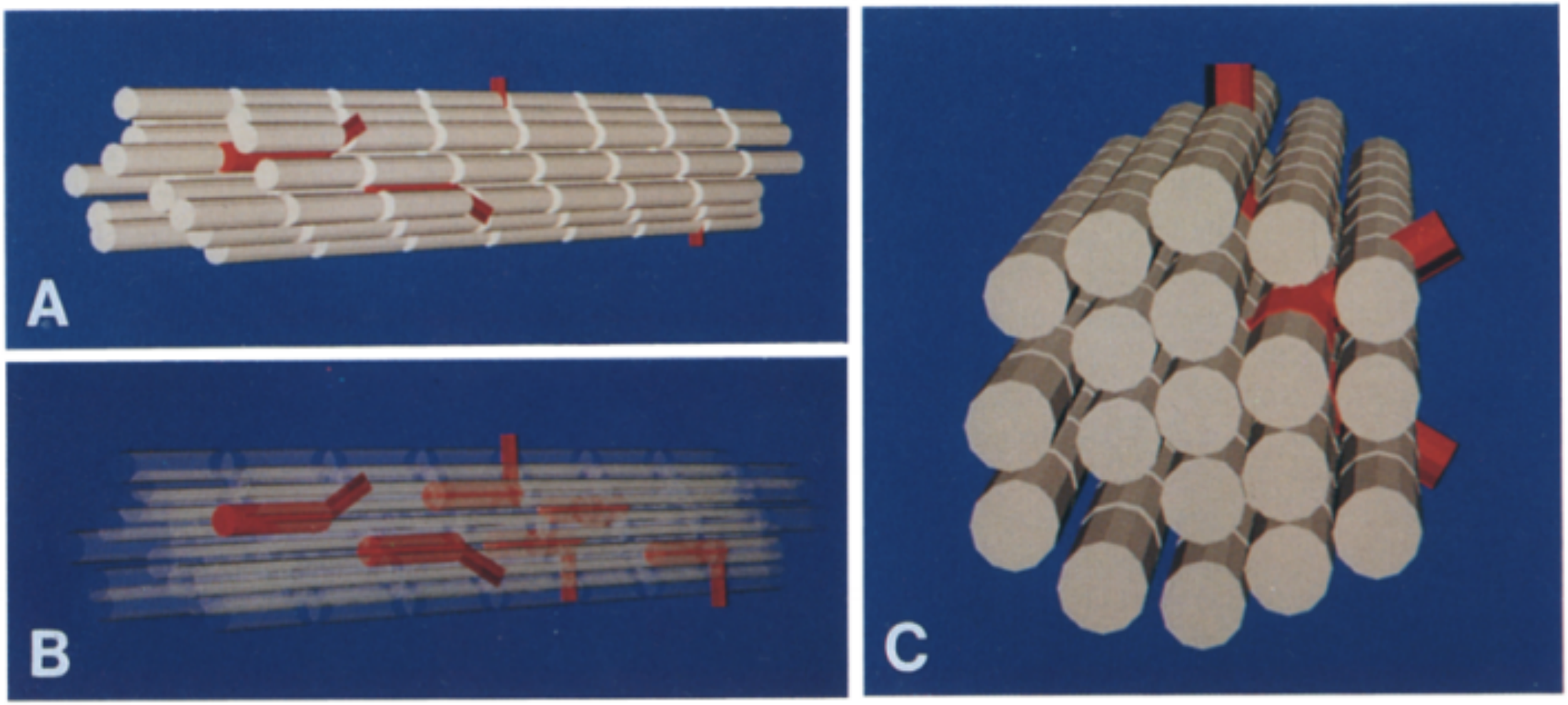
Aim 1: To determine if the SNP is within a DNA motif or regulatory region that effects transcription



Approach: Using DNA Motif databases

Hypothesis: The SNP is located on a transcription regulatory region

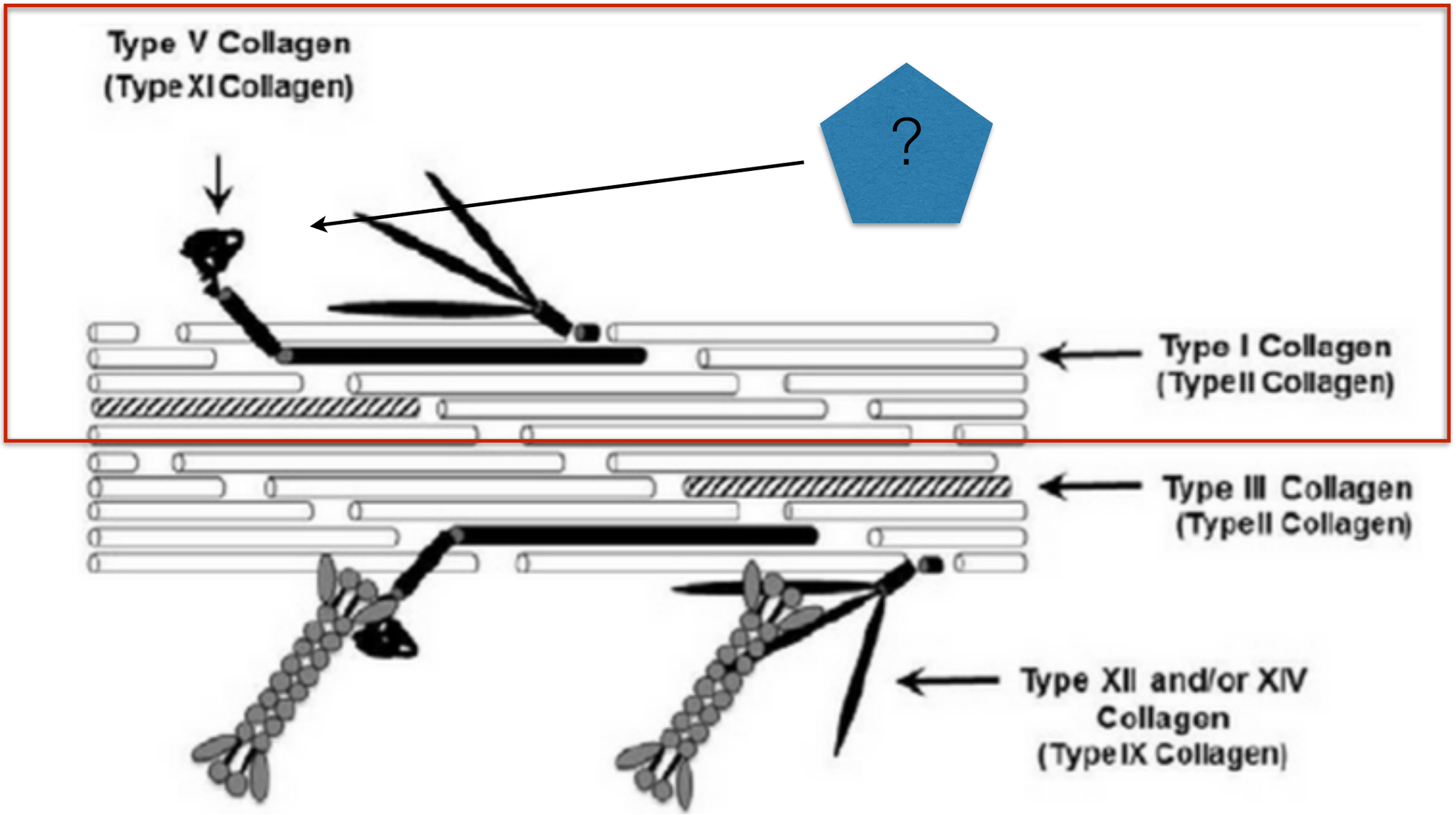
Aim 2: To determine if more of rs12722 is being produced in ligaments



Approach: RNA Sequencing

Hypothesis: Less rs12722 protein is being produced and added to ligaments.

Aim 3: Use a yeast two-hybrid to see what proteins interact with rs12722



Approach: Yeast two-hybrid library and matrix approach

Hypothesis: There is a new protein that interacts with the rs12722 protein that promotes a more stable collagen fibril

Future Directions

Larger marathon studies

More indepth analysis of type V interactions with type I

References

1. Type V collagen: molecular structure and fibrillar organization of the chicken alpha 1(V) NH2-terminal domain, a putative regulator of corneal fibrillogenesis. The Journal of Cell Biology. 1993;121(5):1181-1189.
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4. (2006) COL5A1 gene. Genetics Home Reference. http://ghr.nlm.nih.gov/gene/COL5A1