

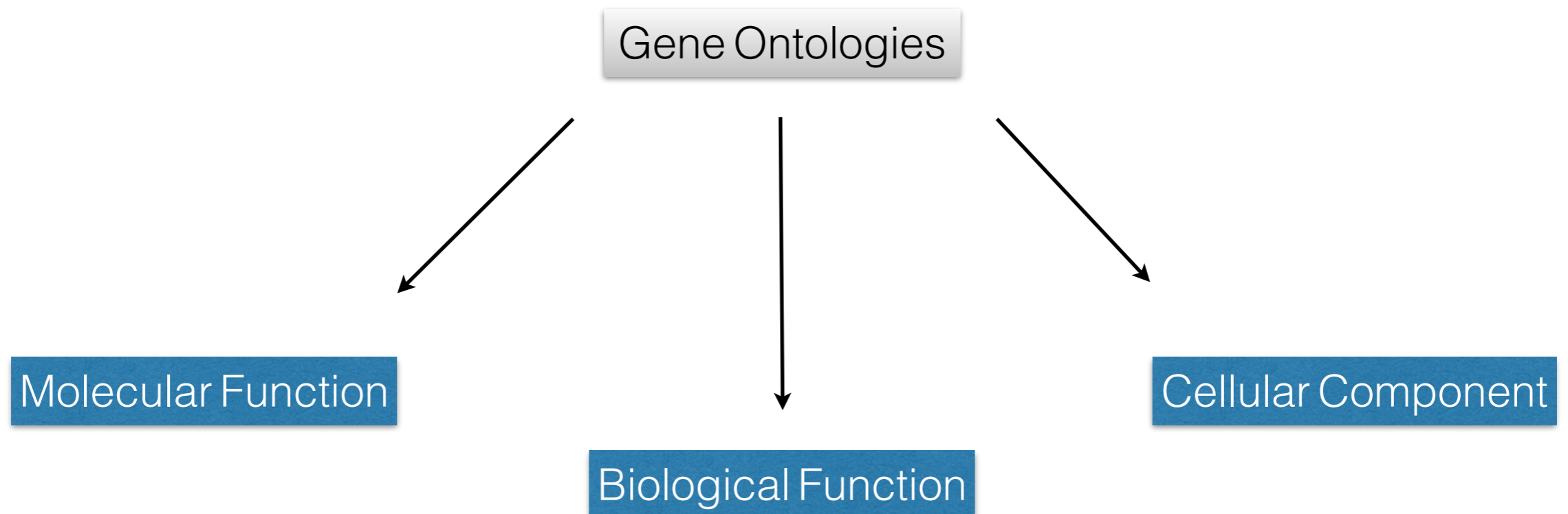
COL5A1: Gene for improved long distance running

By: Logan Silber

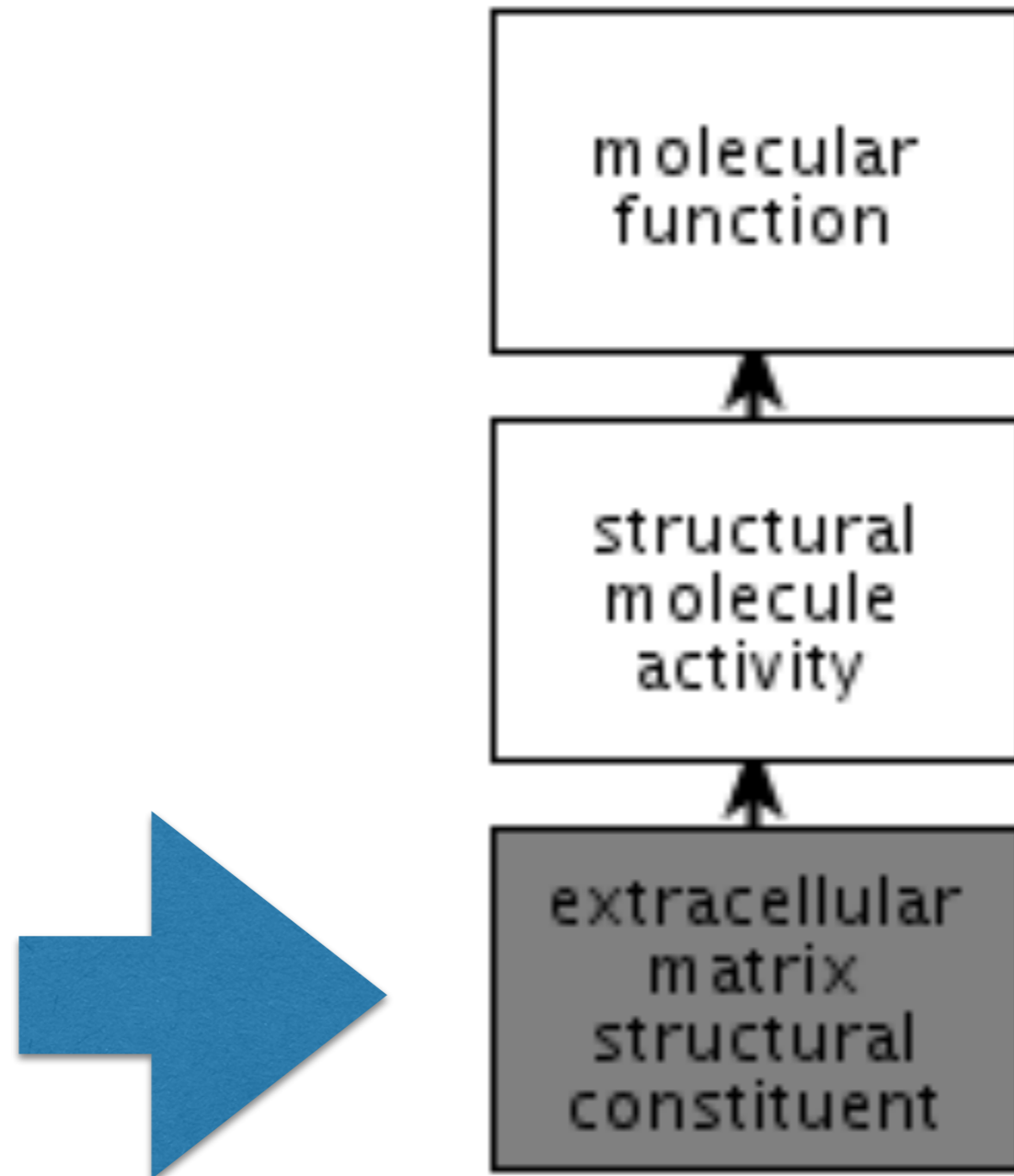
The trait: enhanced running economy



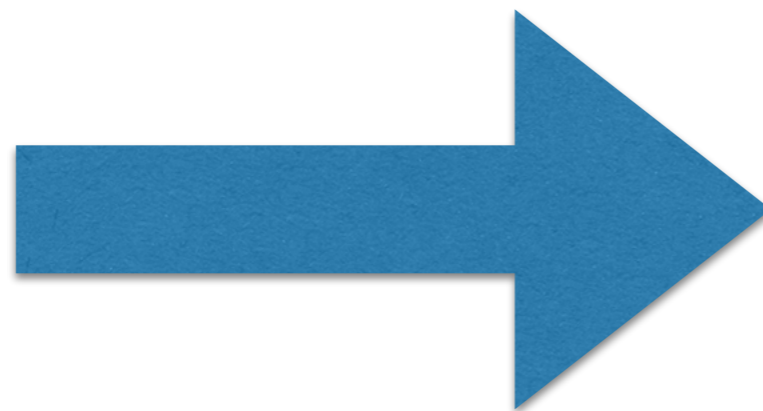
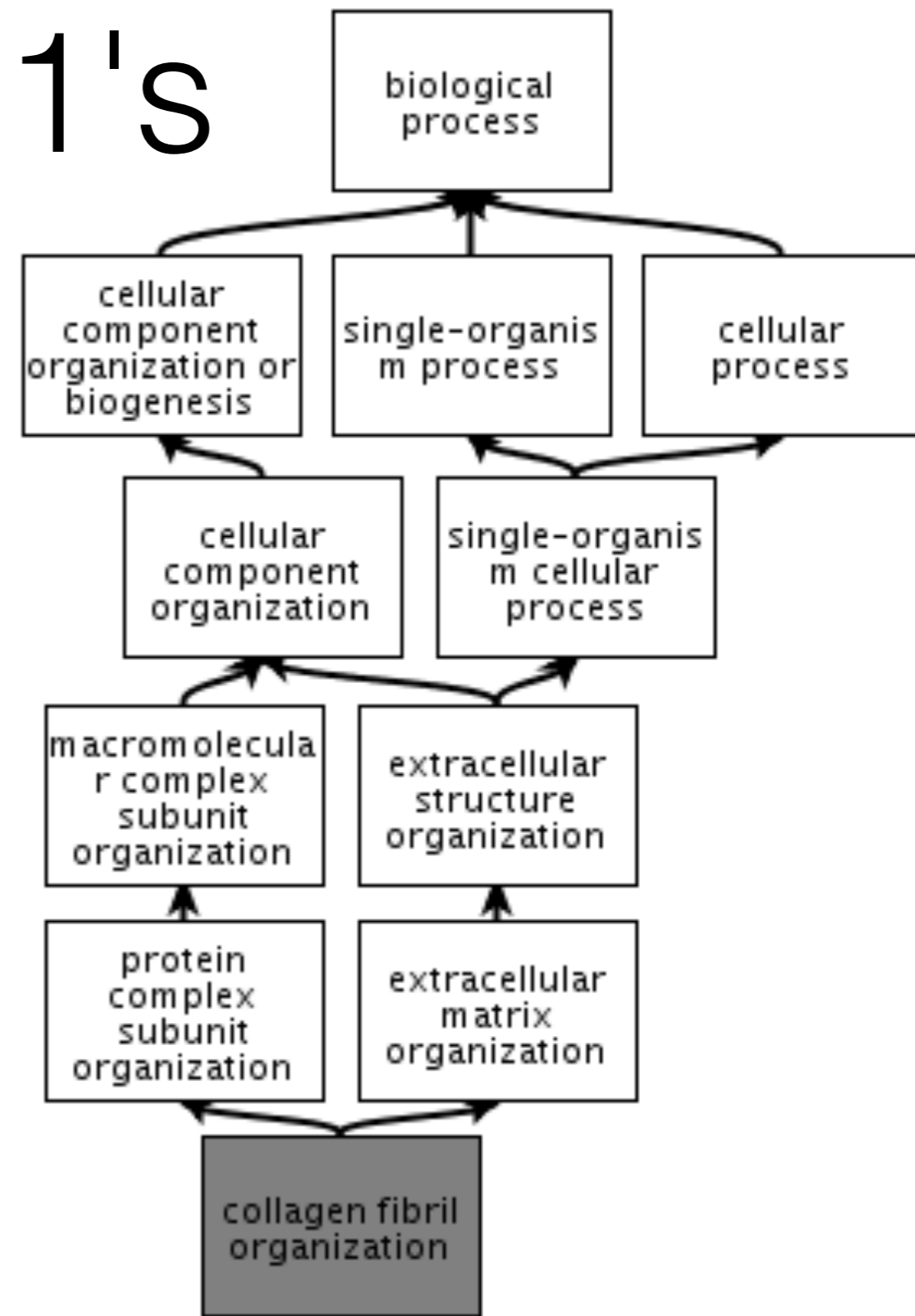
What gene is mutated in my trait?



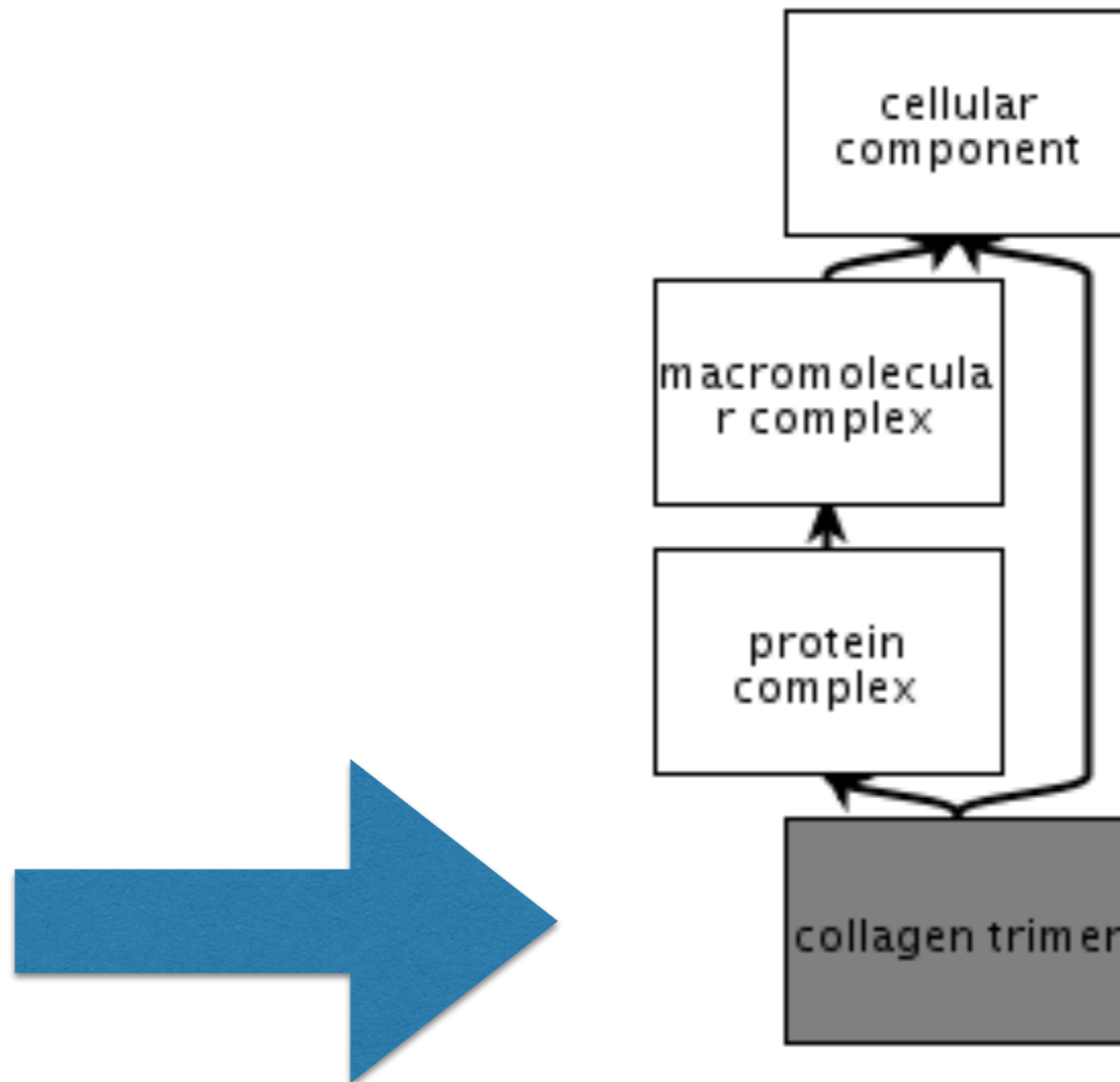
What is COL5A1's molecular function?



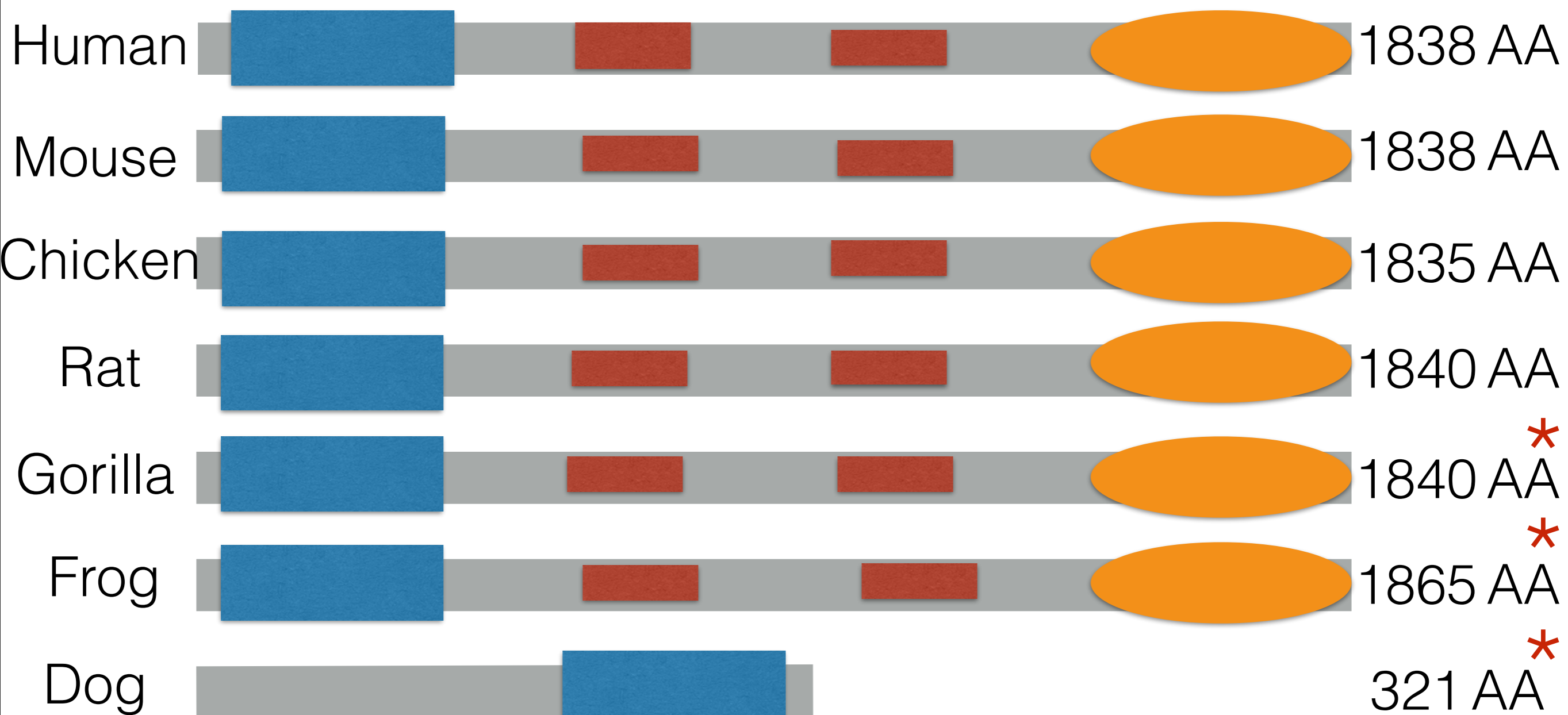
What is COL5A1's biological function?



What is COL5A1's cellular component?



How well conserved is my protein amongst other organisms?



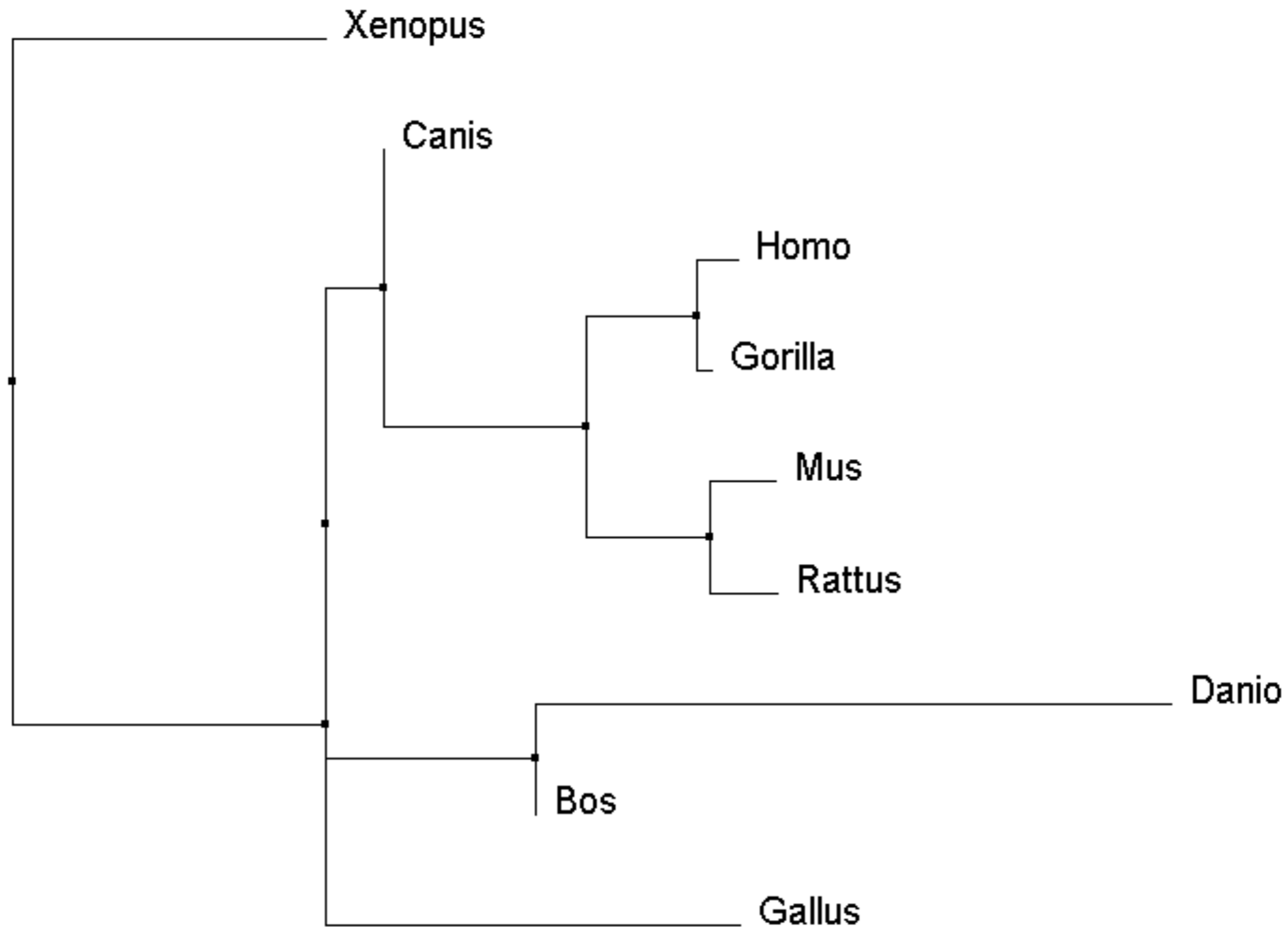
* Predicted

Collagen Triple helix repeat

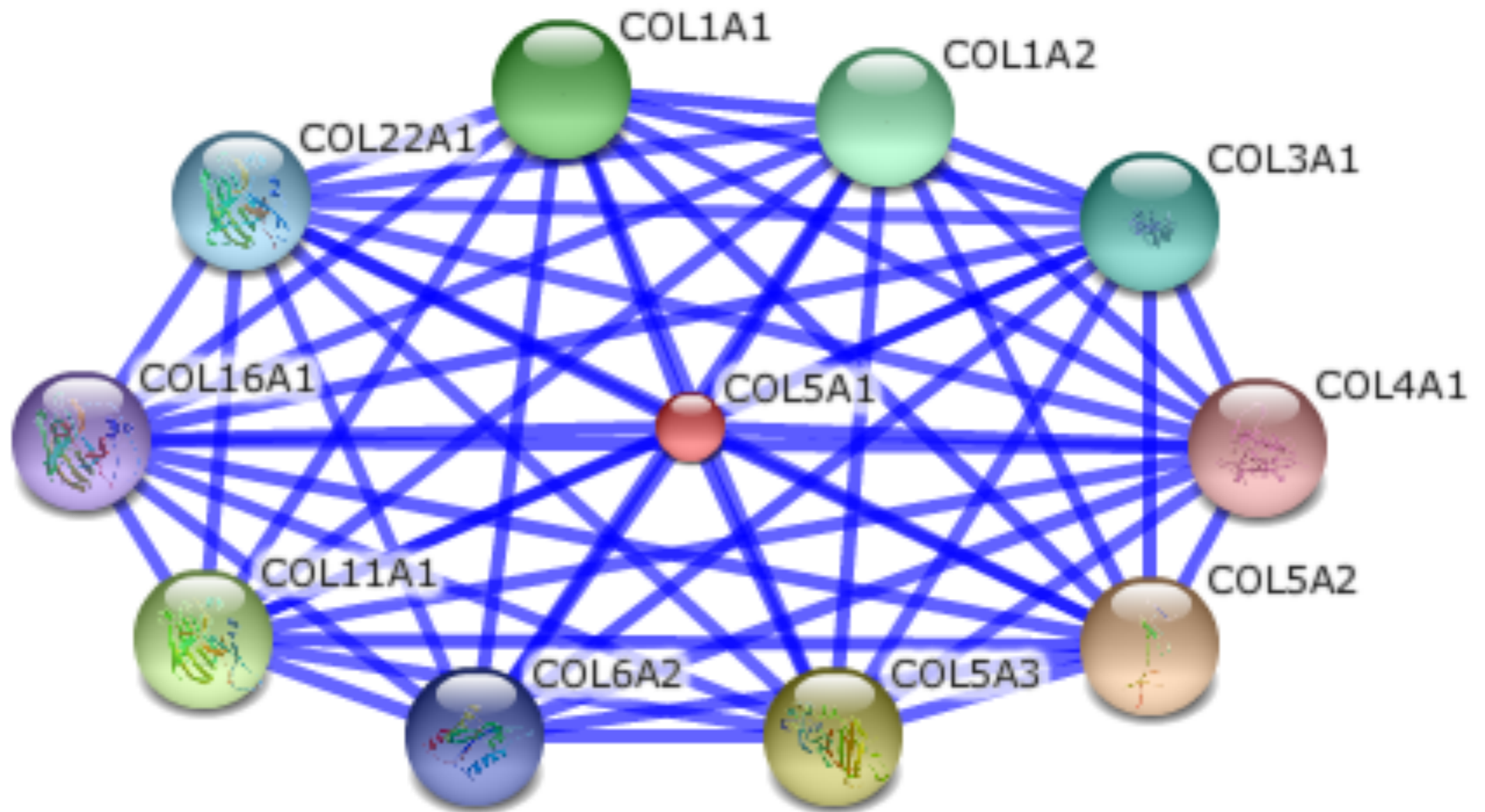
COLFI

Laminin G

Phylogeny of gene amongst other organisms



Protein interactions: COL5A1 product interacts with proteins needed for collagen fibril synthesis



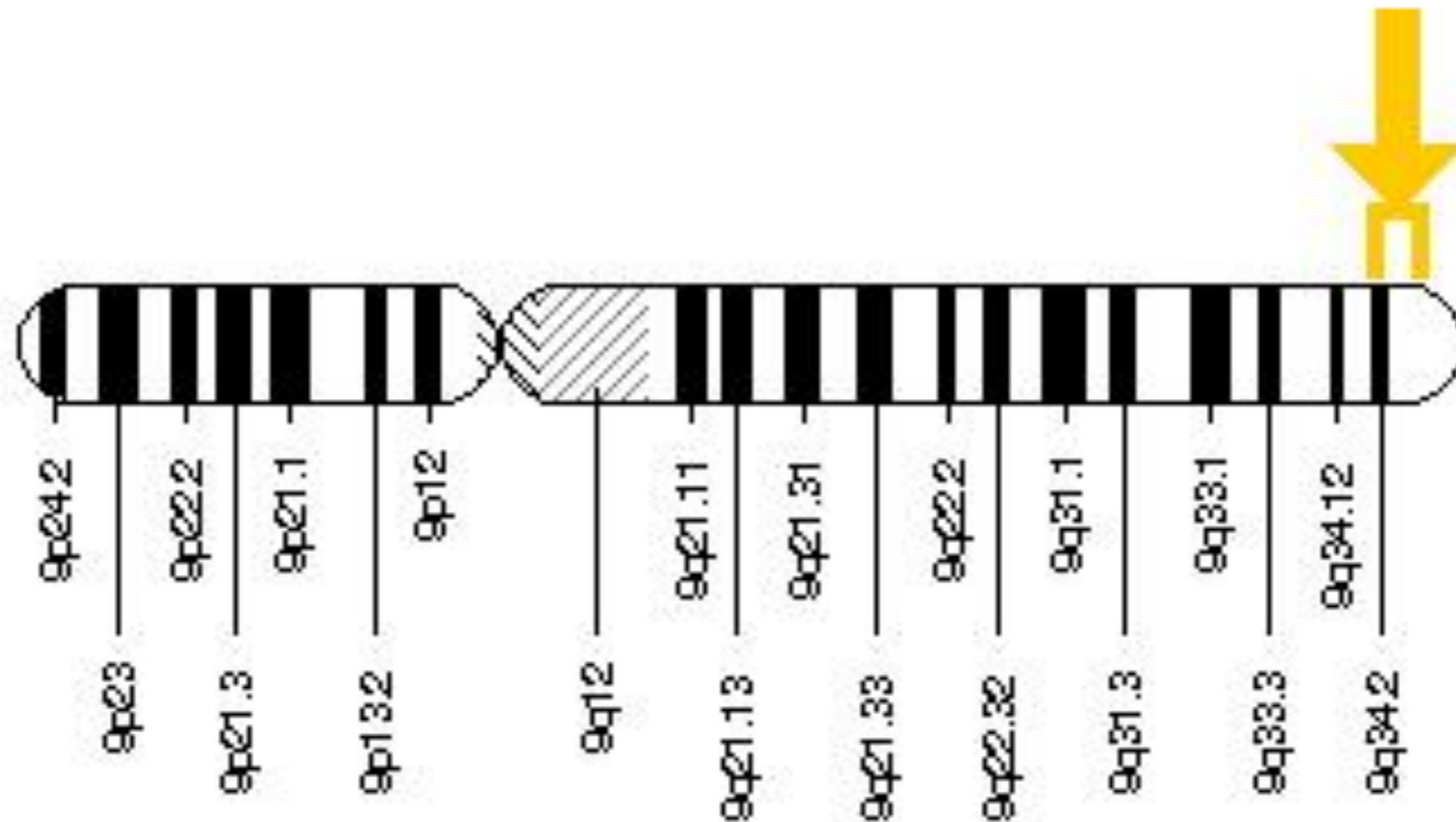
Gap in knowledge

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

Hypothesis

Type V collagen produced by rs12722 does enhance running economy due to a more durable collagen fibrils and a more stable interaction with type I collagen

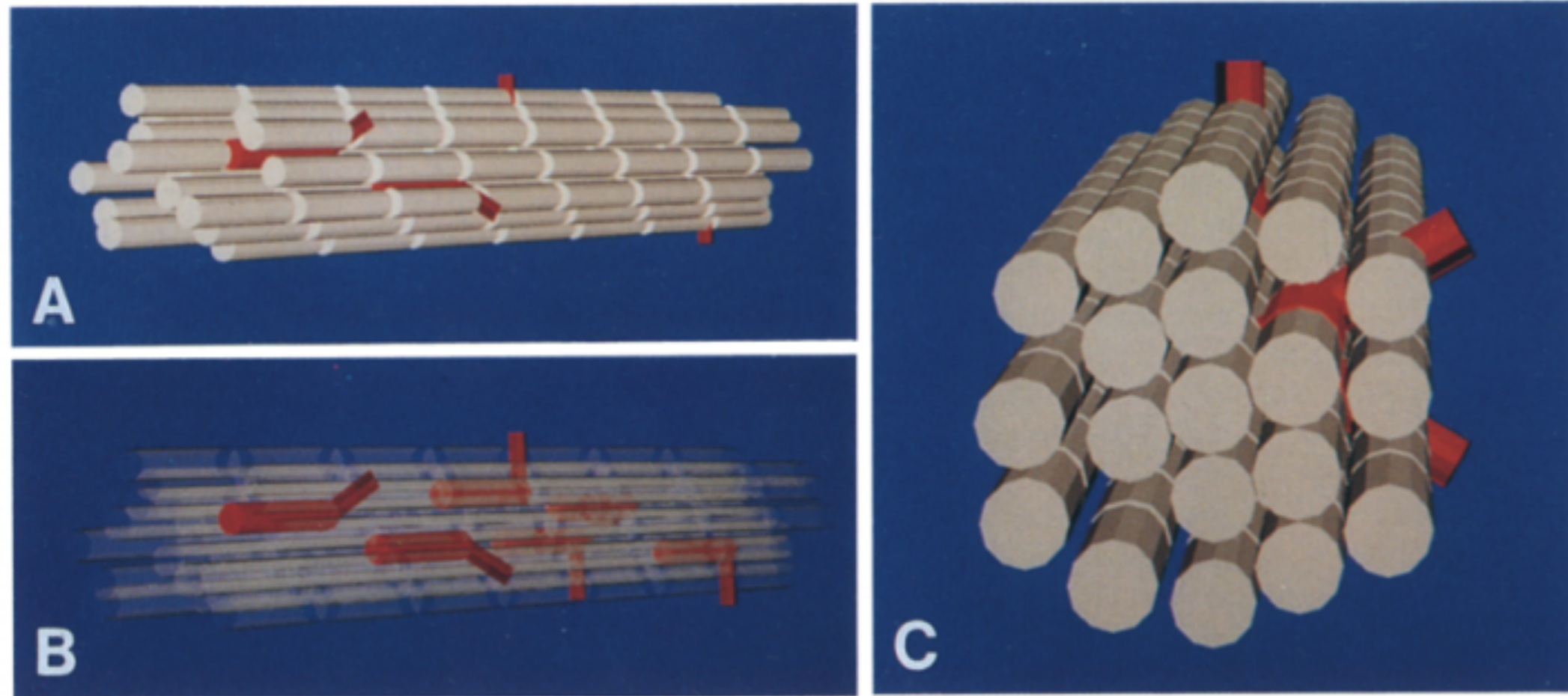
Aim 1: To determine if the SNP is within a DNA motif or regulatory region that effects transcription



Approach: Using DNA Motif databases

Hypothesis: The SNP is located on a transcription regulatory region

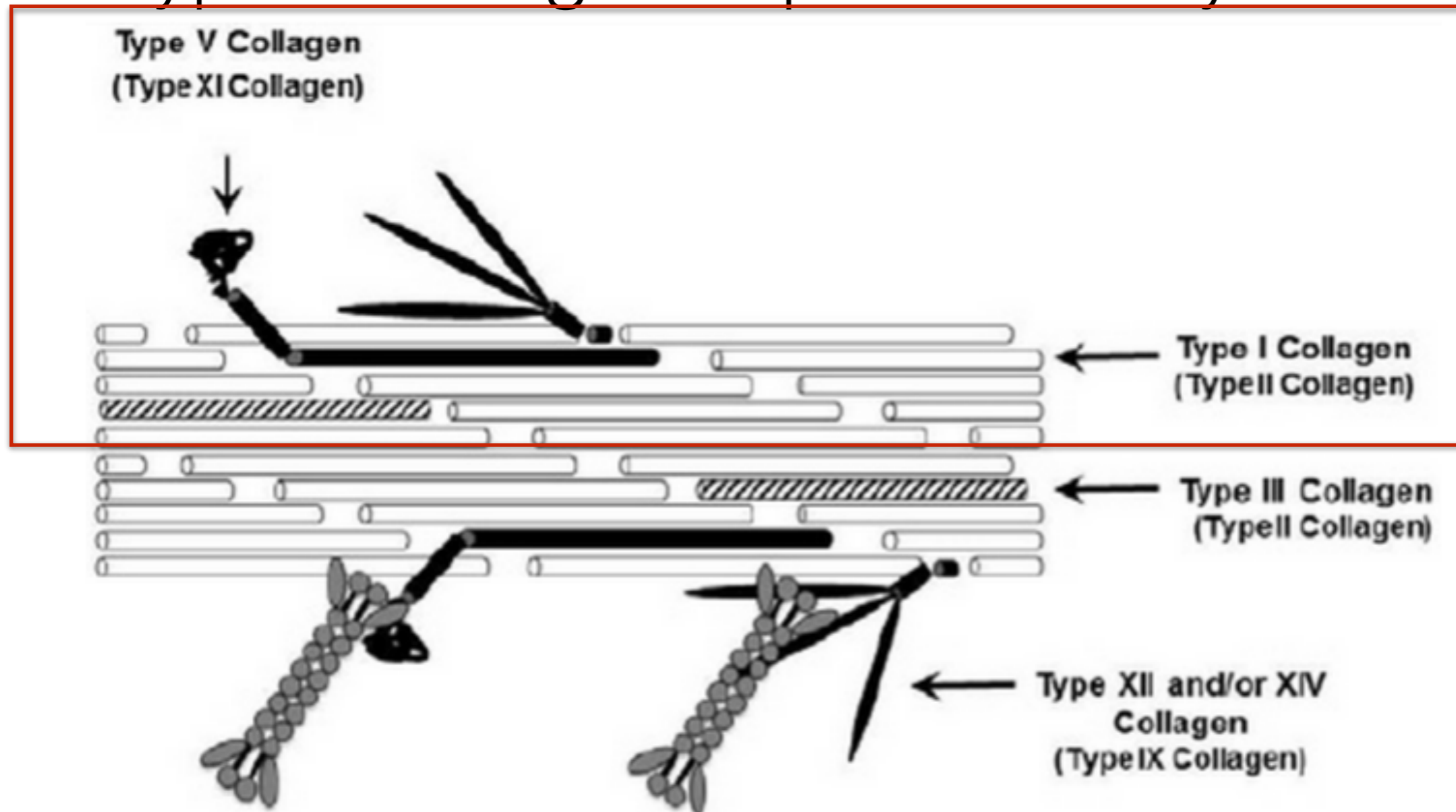
Aim 2: To determine if more of rs12722 is being produced in ligaments



Approach: RNA Sequencing

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

Aim 3: to determine if the ratio of type I collagen to type V collagen is lower when the type V collagen is produced by rs12722



Approach: Mass Spectroscopy

Hypothesis: There is a lower ratio of type V/type I collagen in ligaments in those with rs12722

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.
2. <http://www.snpedia.com/index.php/Rs12722>
3. COLLAGEN GENE SEQUENCE VARIANTS IN EXERCISE-RELATED TRAITS. *Central European Journal of Sport Sciences and Medicine* | Vol. 1, No. 1/2013: 3–17
4. (2006) COL5A1 gene. Genetics Home Reference. <http://ghr.nlm.nih.gov/gene/COL5A1>