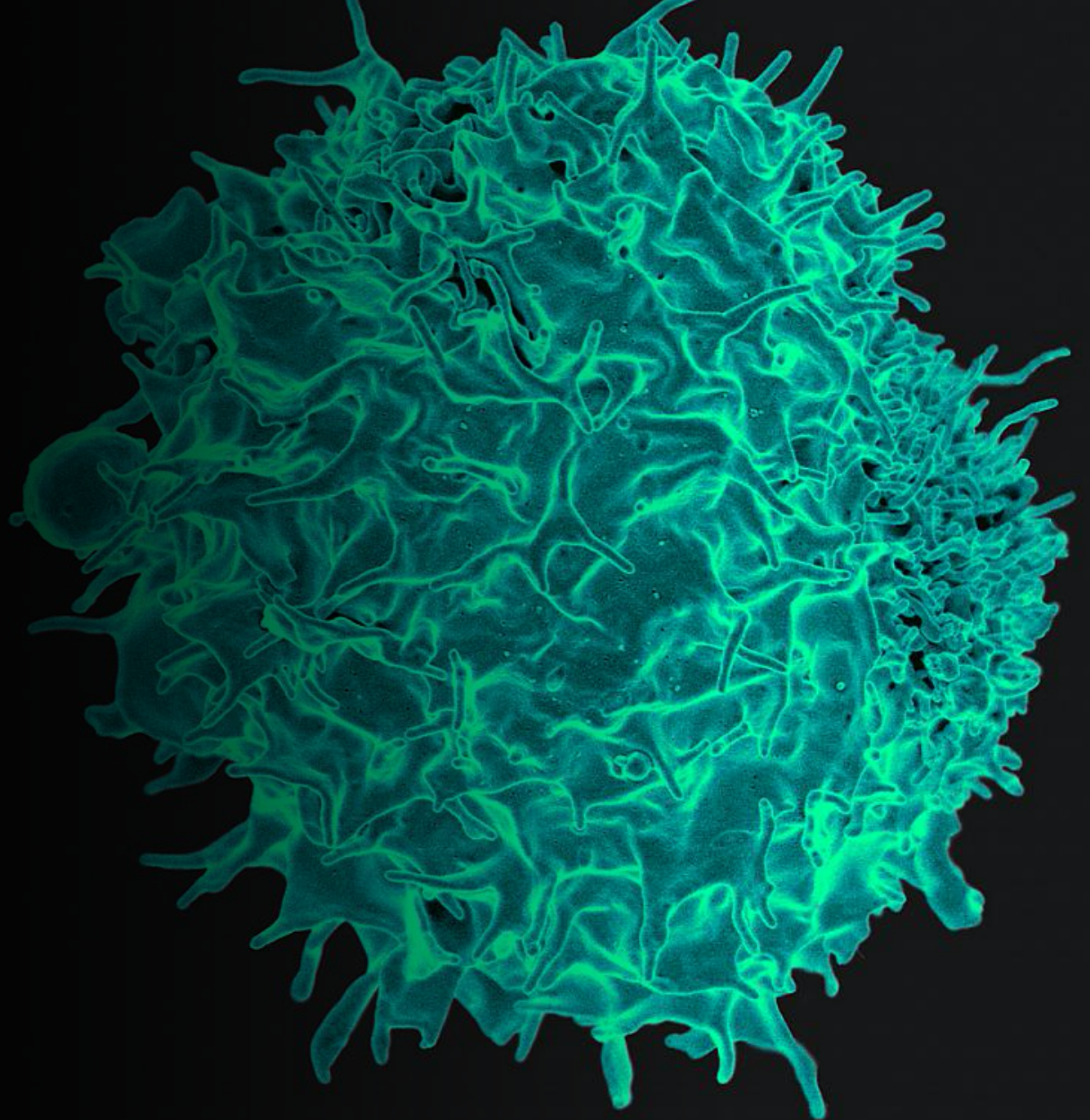


# Griscelli Syndrome Type II and Rab27A

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Presented By Gus Hermberg

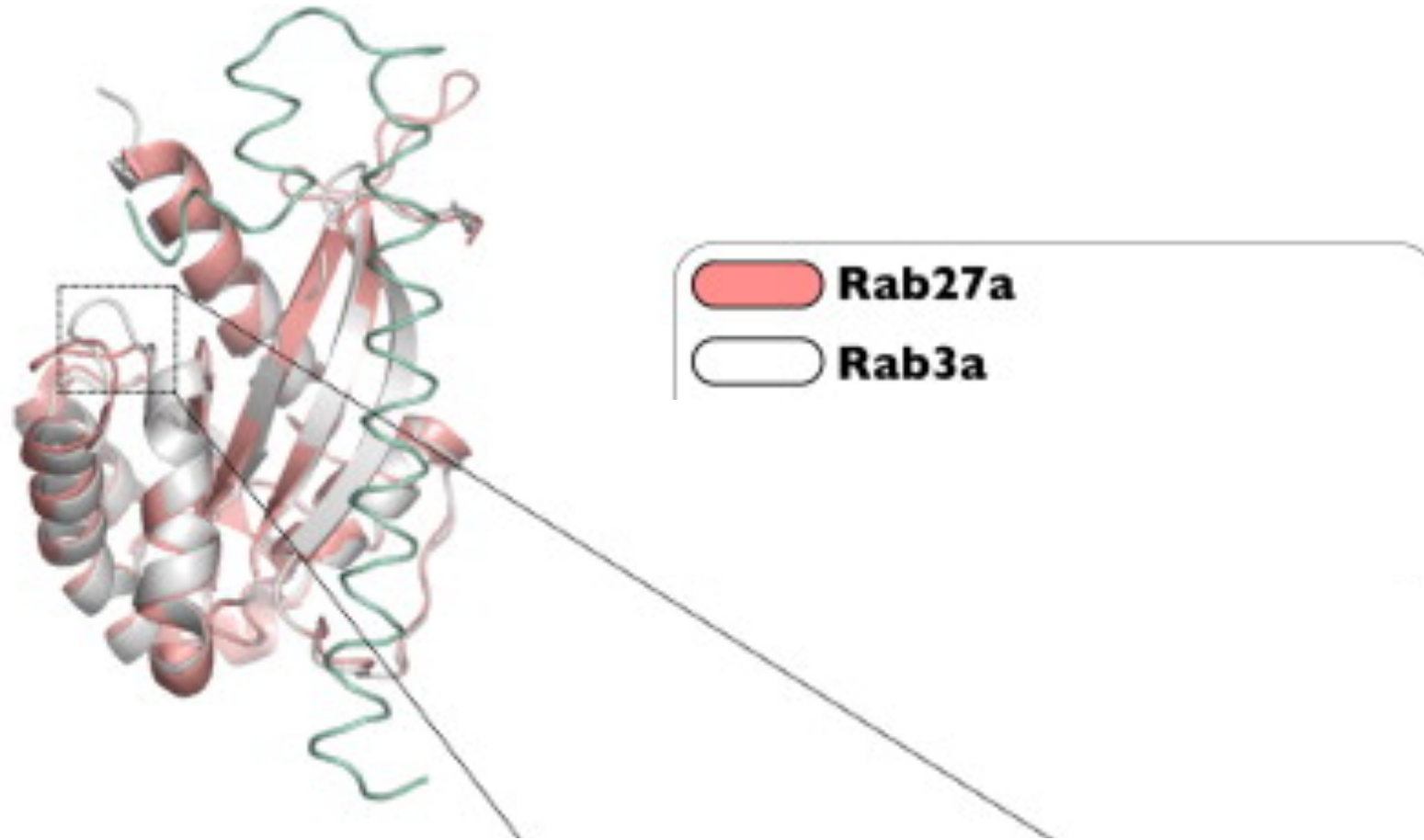


# What is Griscelli Syndrome?



It is a disease where the affected have partial albinism and immunodeficiency.

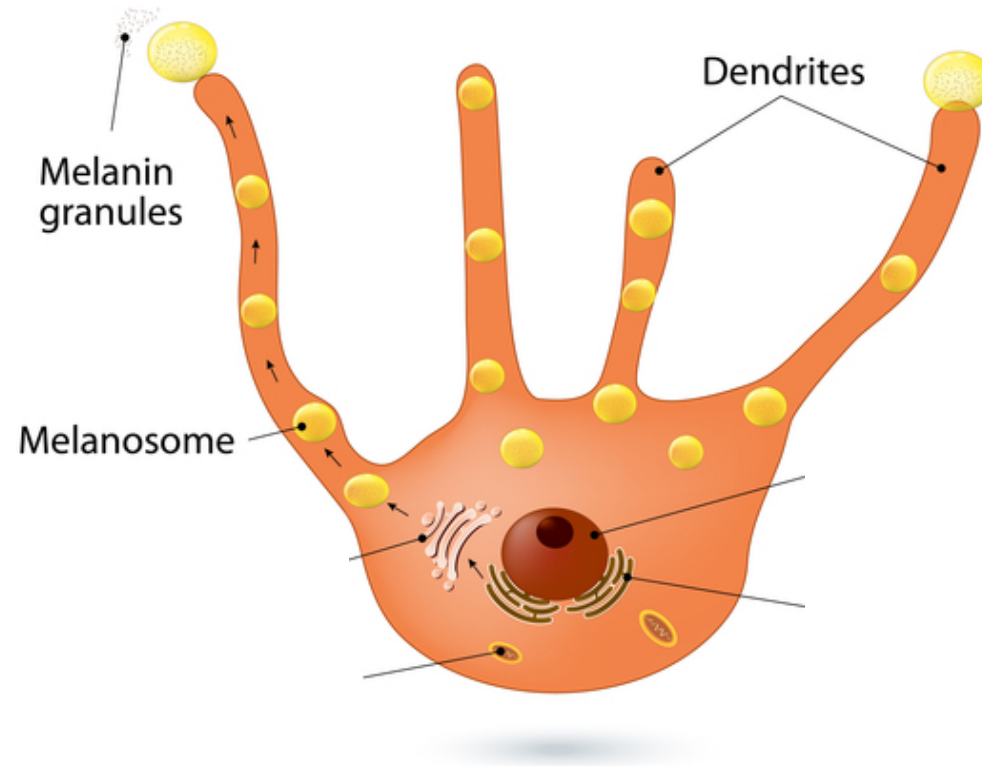
# What is type II?



Type II is associated specifically with mutations in Rab27A.

# What causes the albinism?

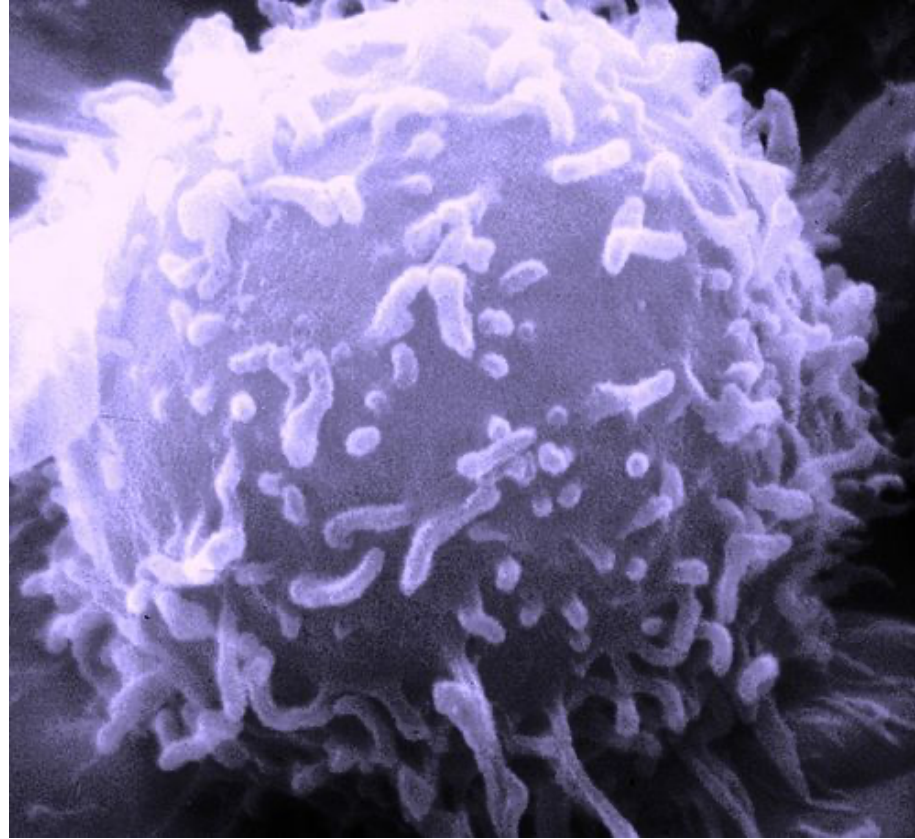
## MELANOCYTE



Inability to transport melanin outside of cells results in albinism.

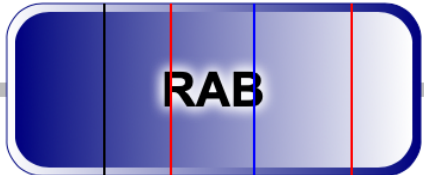


# What causes the immunodeficiency?



Disruption in cell transport makes lymphocytes ineffective at fighting infections.

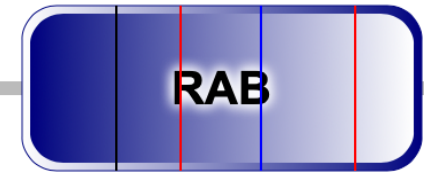
# How conserved is the gene?



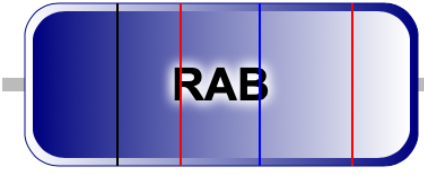
Humans



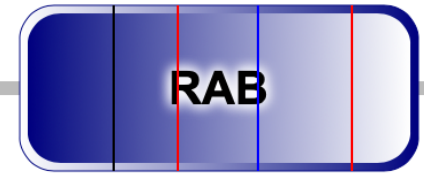
Rats



Dogs



Zebrafish



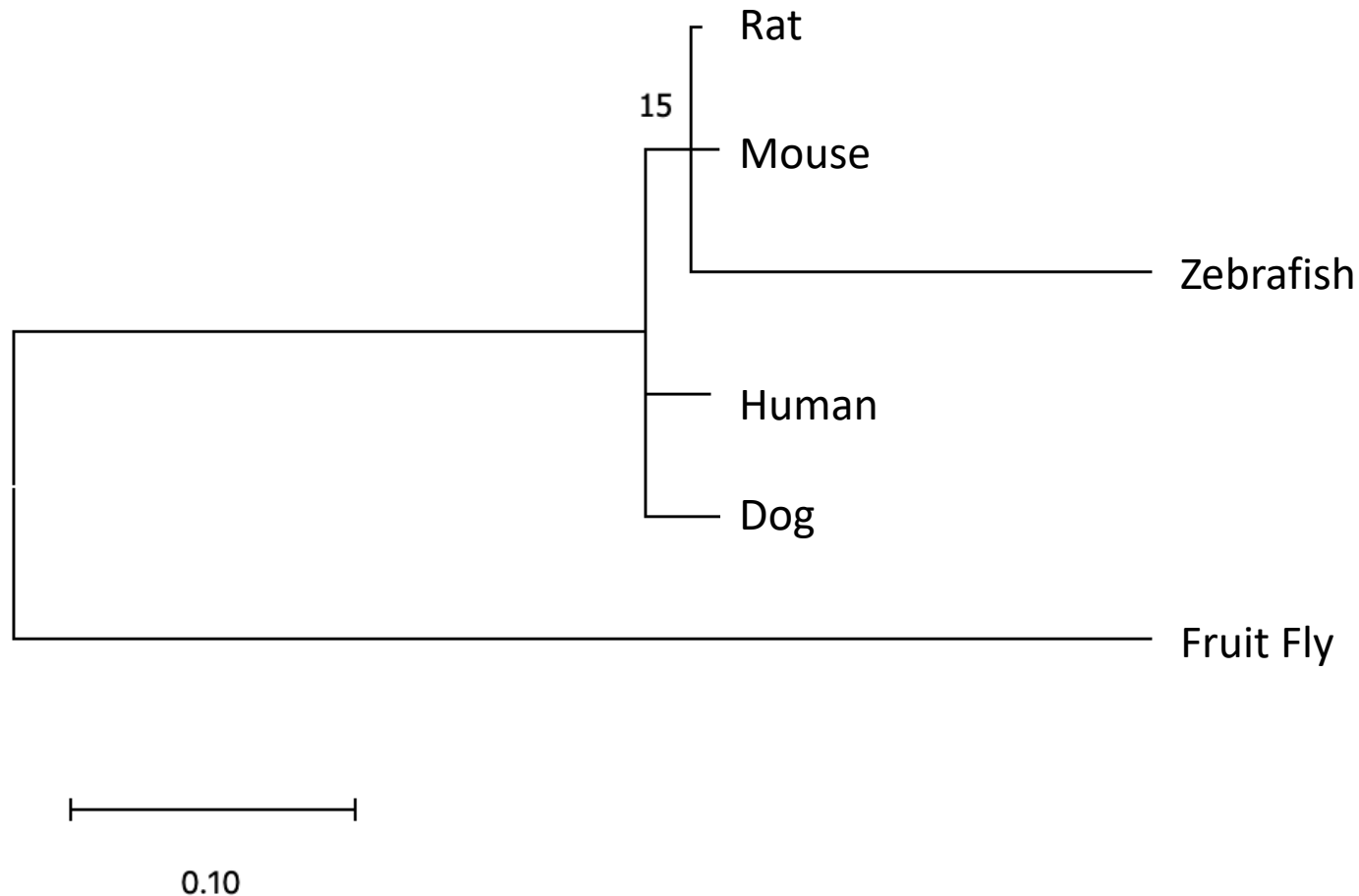
Mouse



Fruit Flies

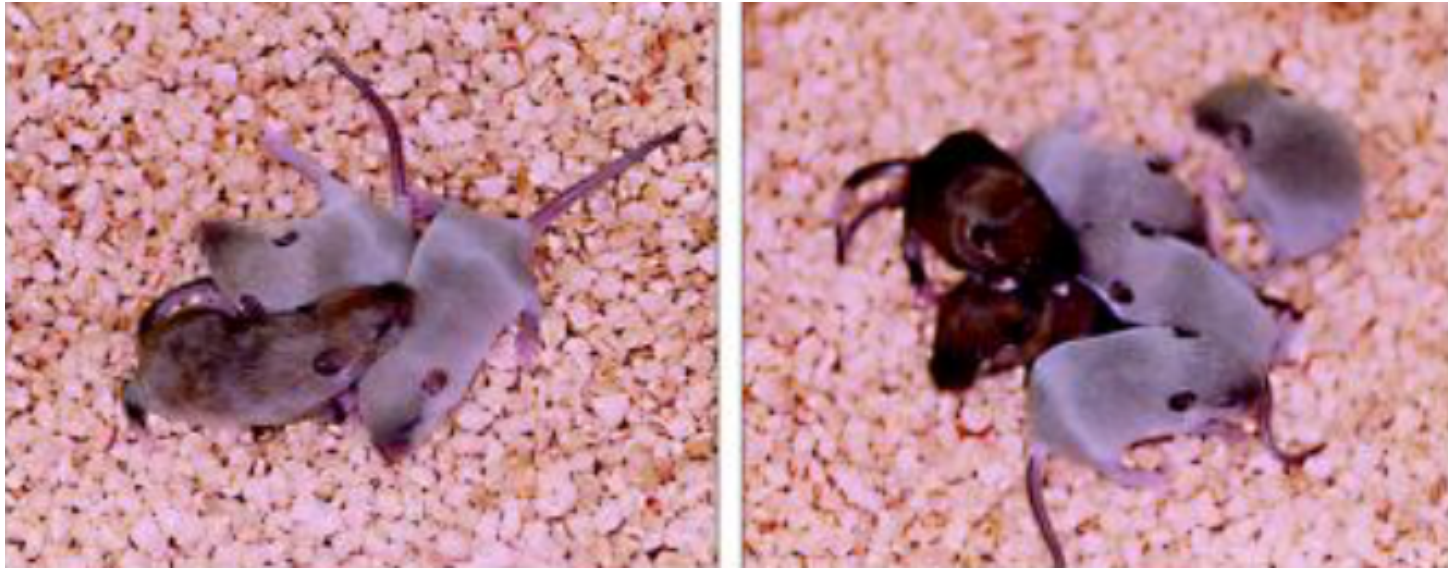
The structure is highly conserved between humans and model organisms.

# What is the pattern of descent for Rab27A?

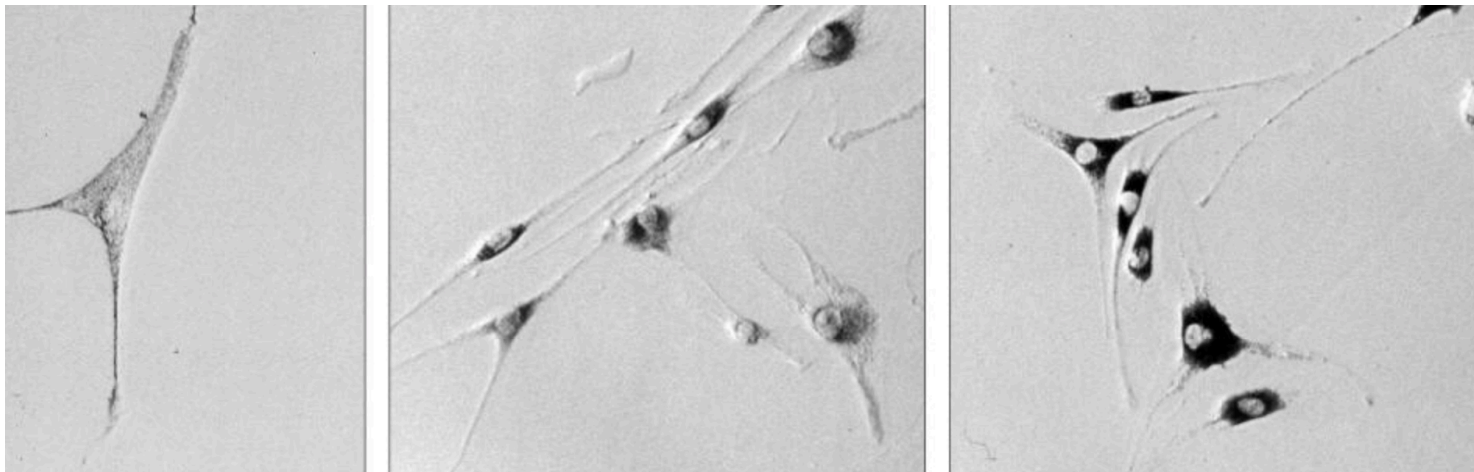


Phylogenetics show that the dog copy of the gene is most closely related to the human copy and the fruit fly copy is the outgroup.

# What could potentially be used to study GStII?



Mutant ashken mice provide an easy to detect phenotype.



Cultured cell lines are easier to grow and maintain and allow membrane transport to be studied.



# References

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