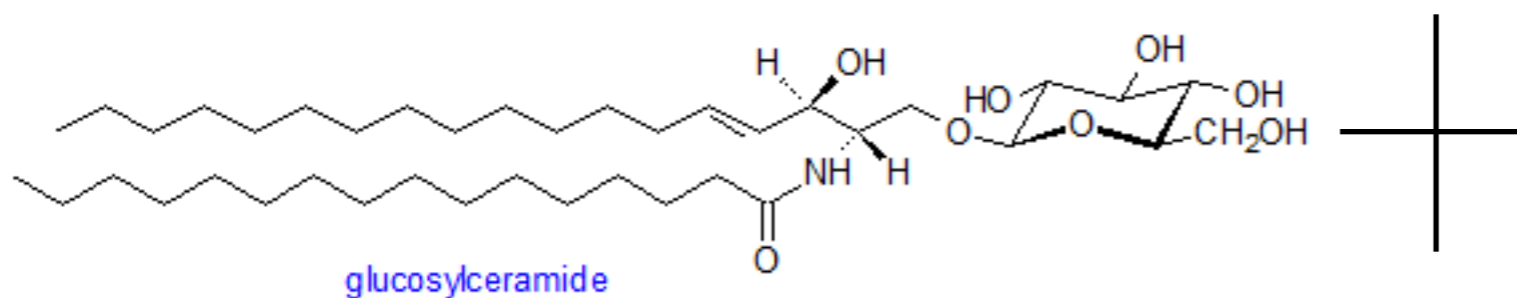
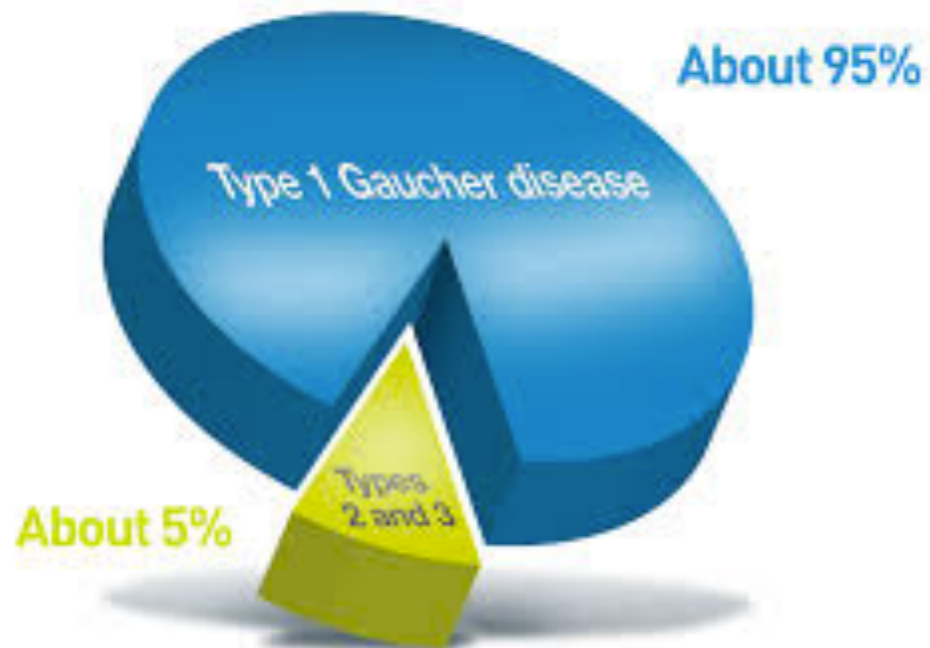


# *PSAP* and Gaucher Disease

By Mitchell Coplan



# What is Gaucher Disease?

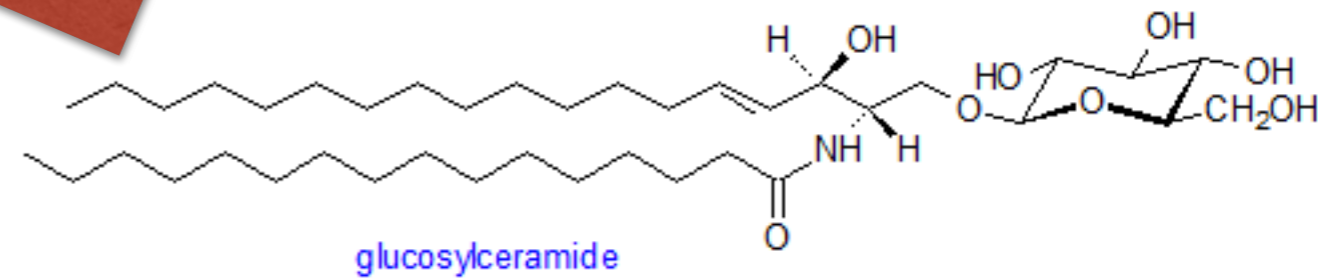
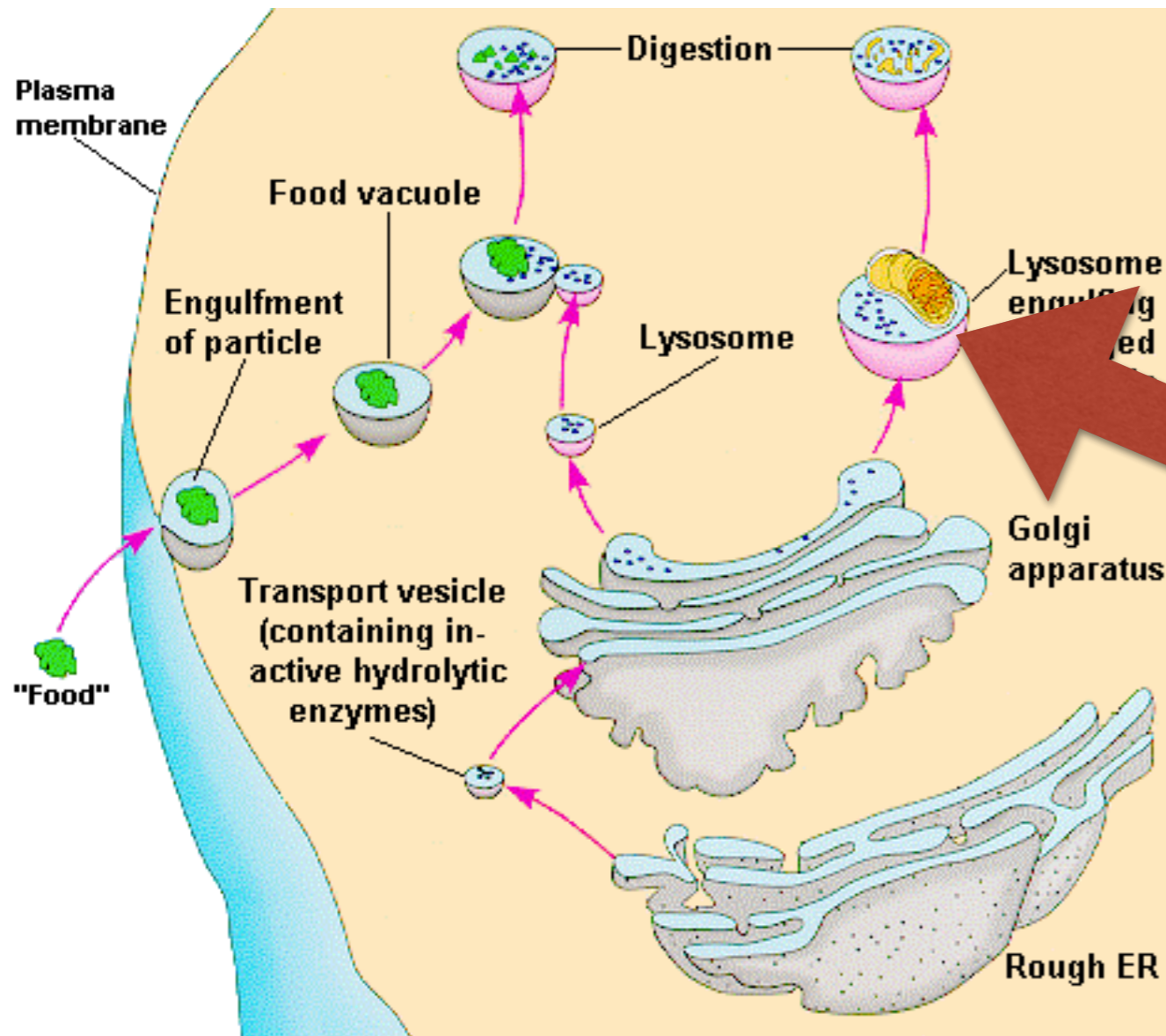


<http://www.vpriv.com/about-vpriv/what-is-type-1-gaucher-disease.php>

<http://www.gauchercare.com/en/healthcare.aspx>

<http://www.vpriv.com/about-gaucher-disease/>

# What causes these phenotypes?

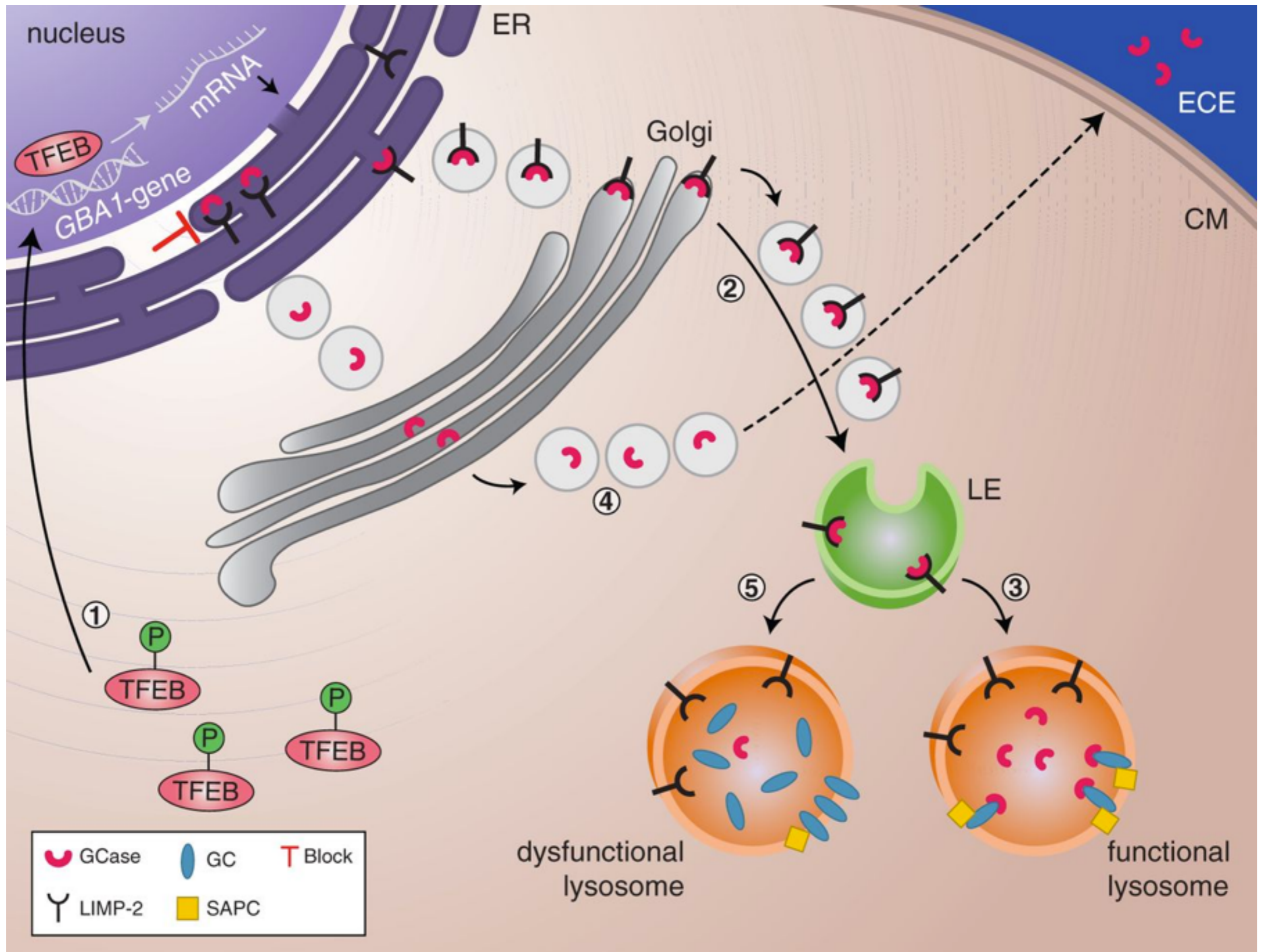


Lysosomal storage disease  
Cells can't break down glucocerebrosides

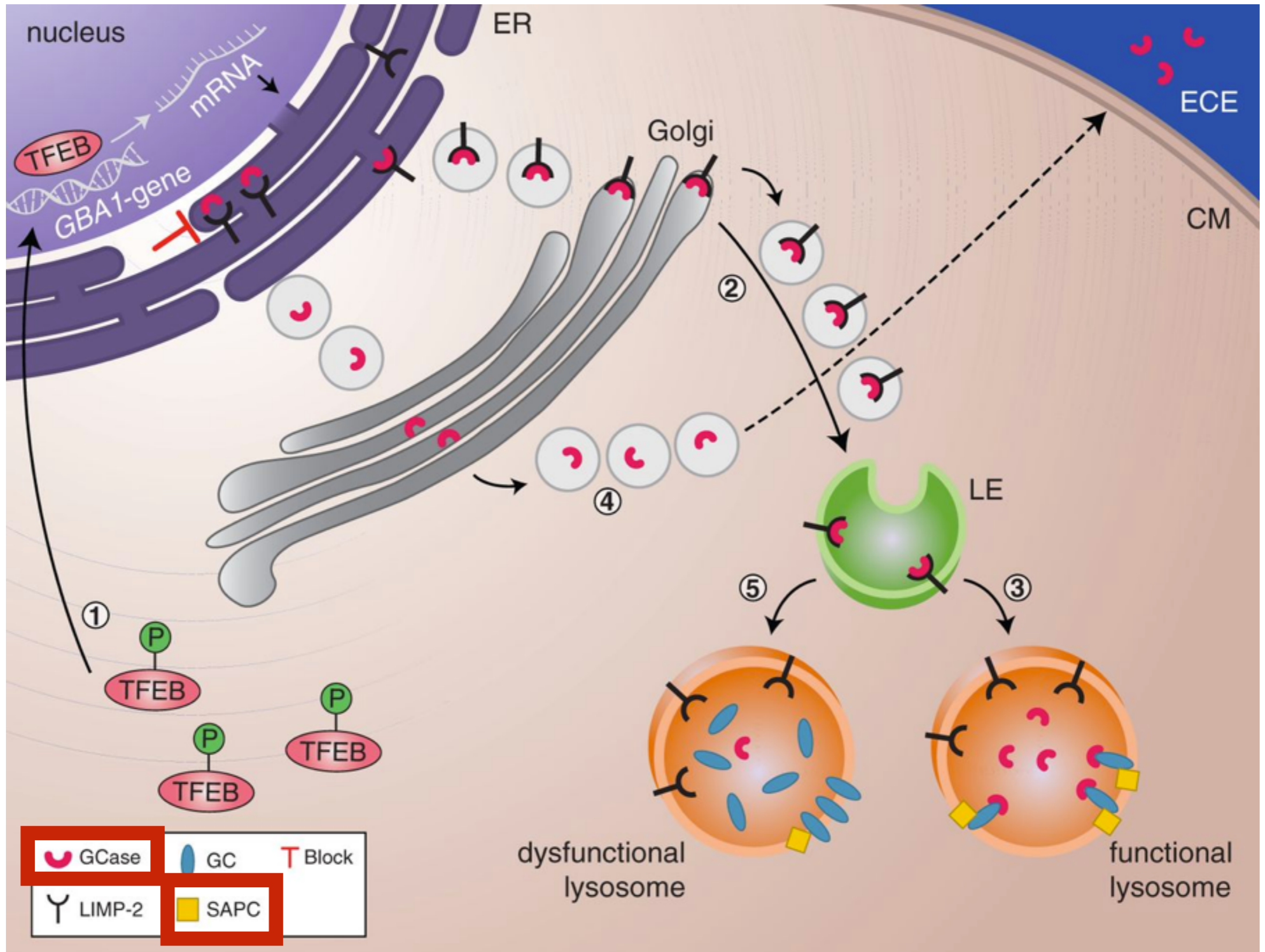
<http://wiki.pingry.org/u/ap-biology/index.php/Lysosomes>

<http://lipidlibrary.aocs.org/Lipids/whatlip/index.htm>

# How are glucocerebrosides(GC) normally broken down?

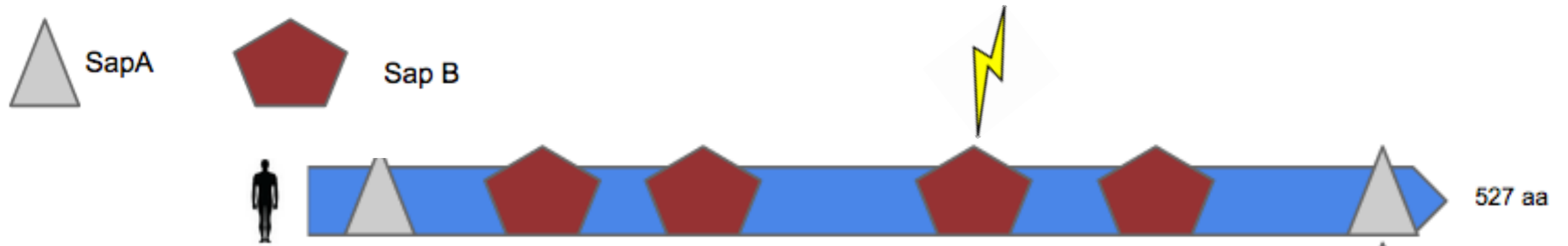


# What gene(s) is mutated in Gaucher Disease?





***GBA* (GCase) and *PSAP* (SAPC)**

# *PSAP* is mutated



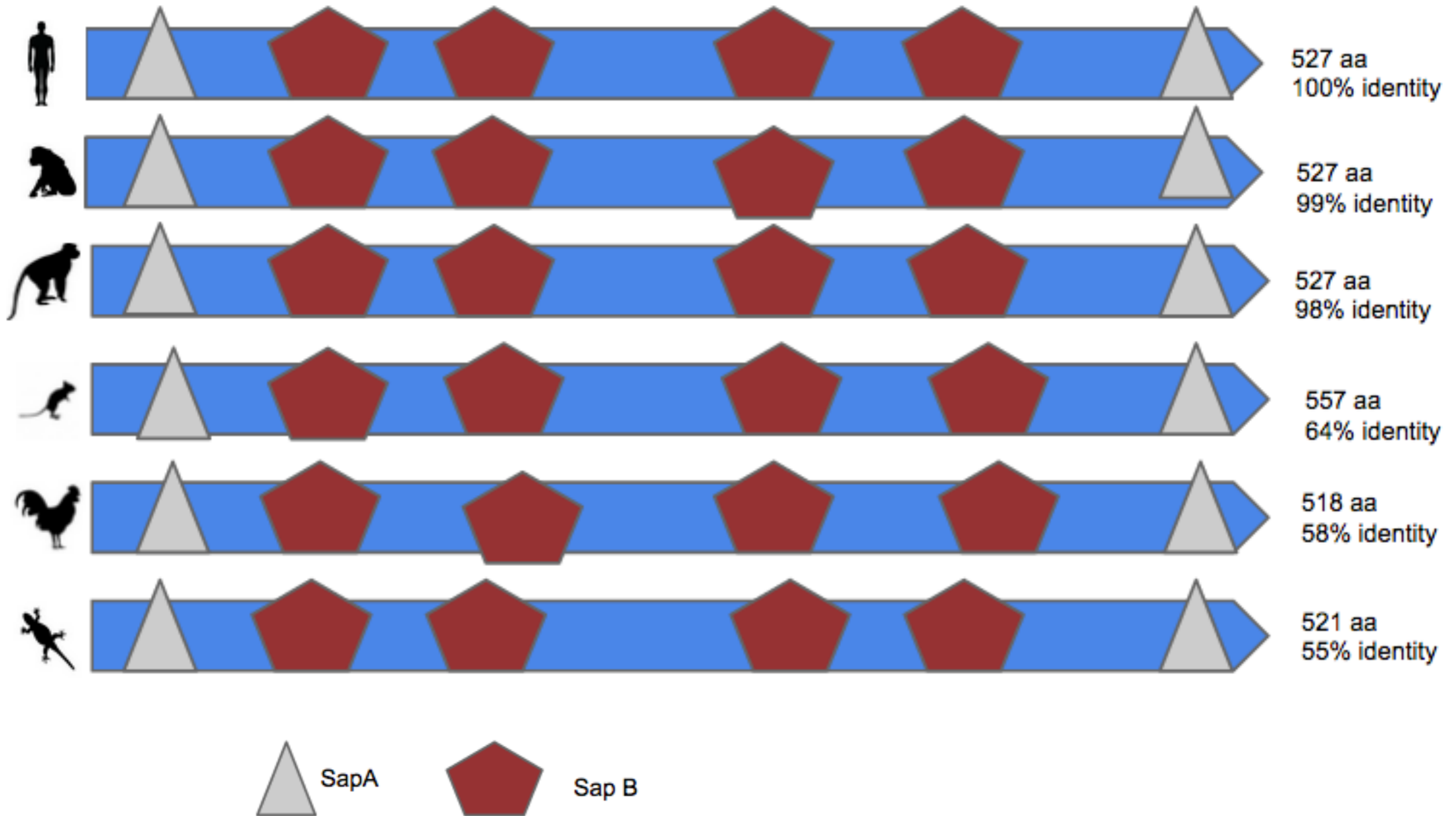
**B**



 Glycosylation site  
 Disulfide bridge

**Mutations occur in disulfide bridges**

# How well conserved is the protein?



**The proteins domains are highly conserved**

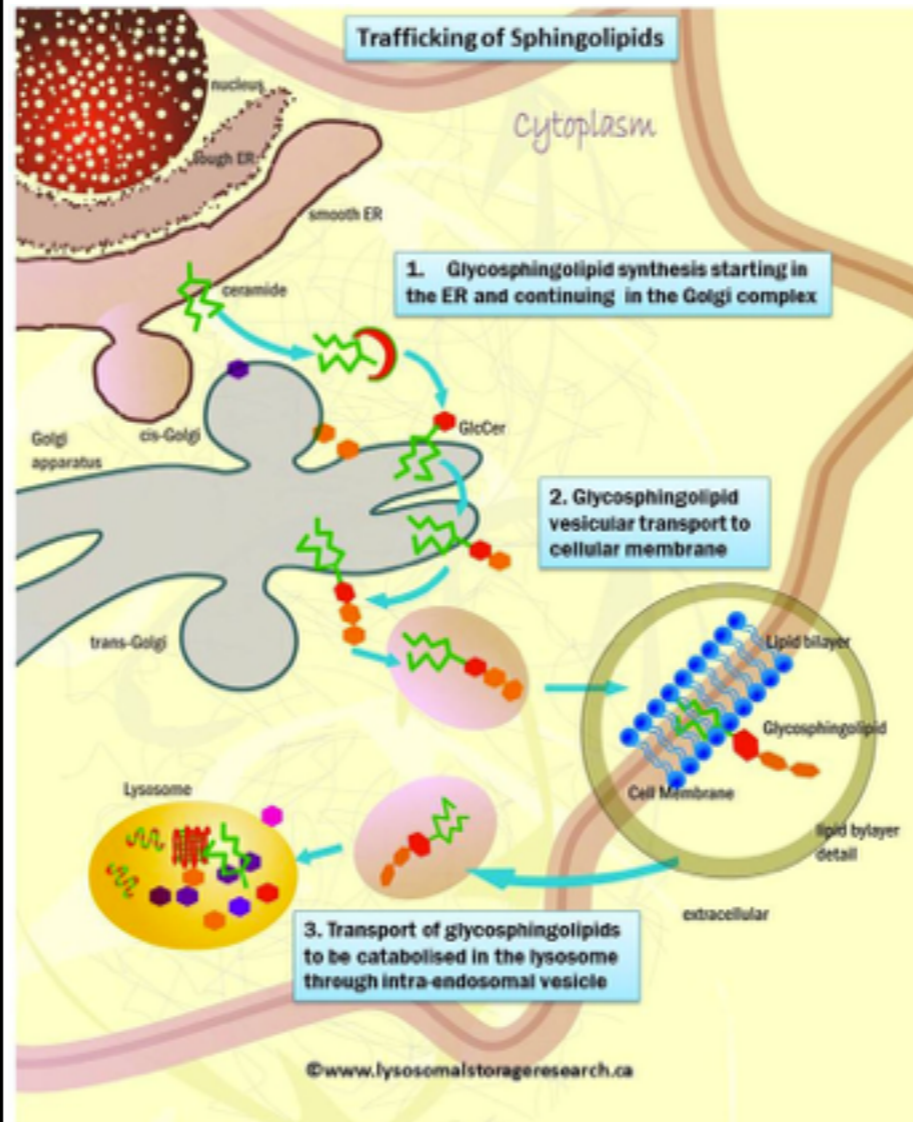
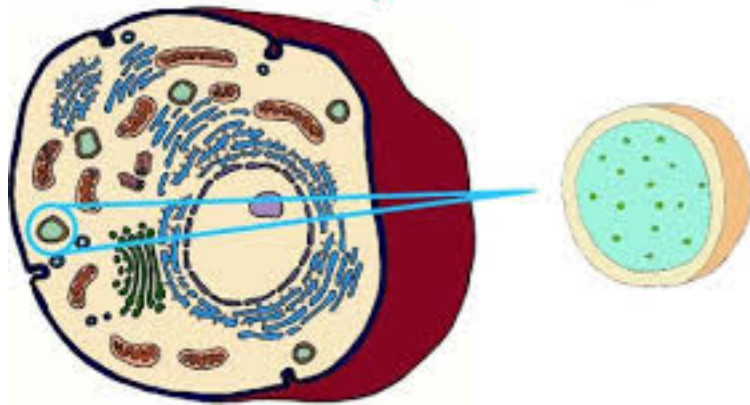
# Where and how does PSAP function?

Cellular Components

Biological Processes

Molecular Function

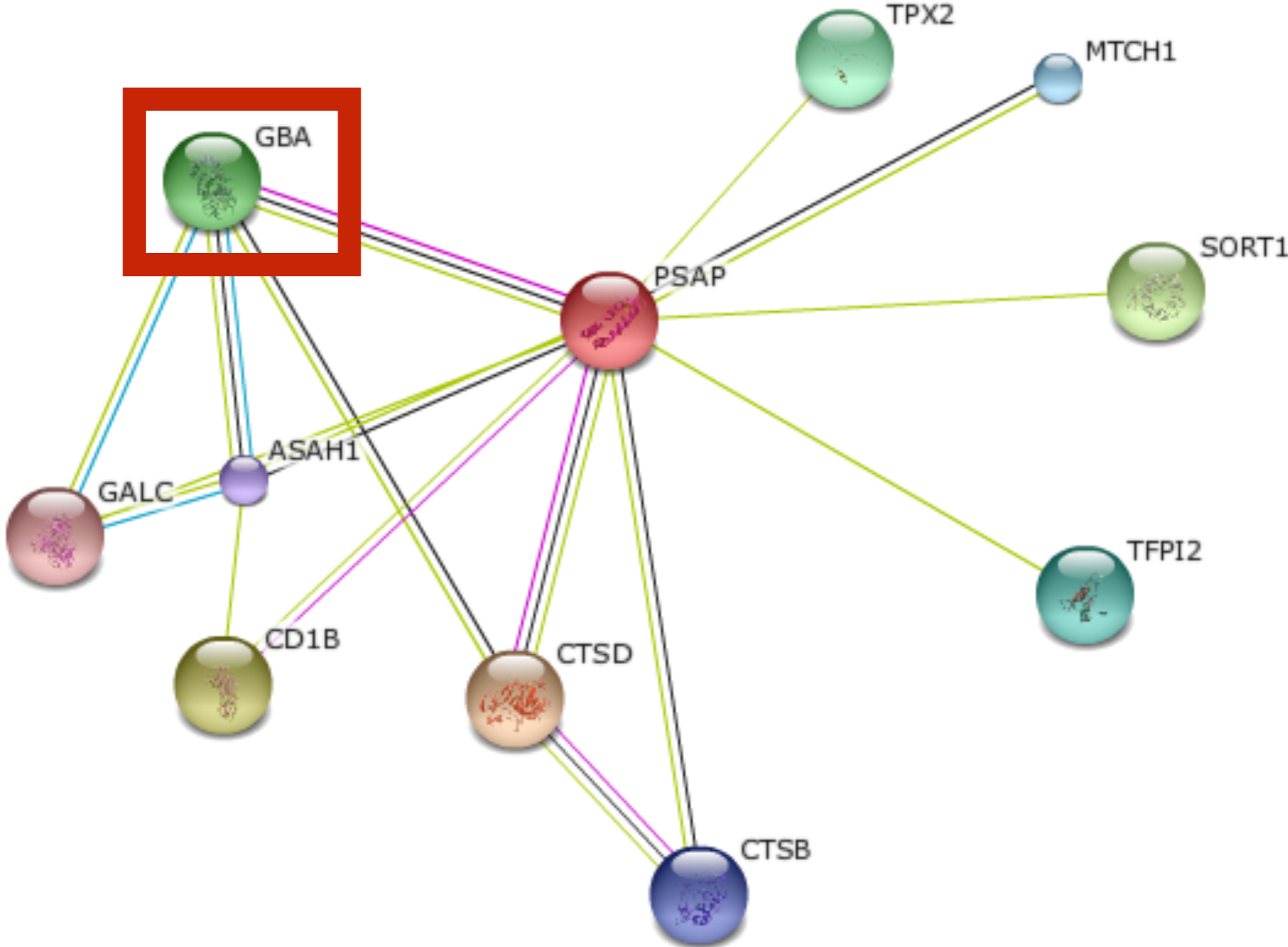
Lysosome



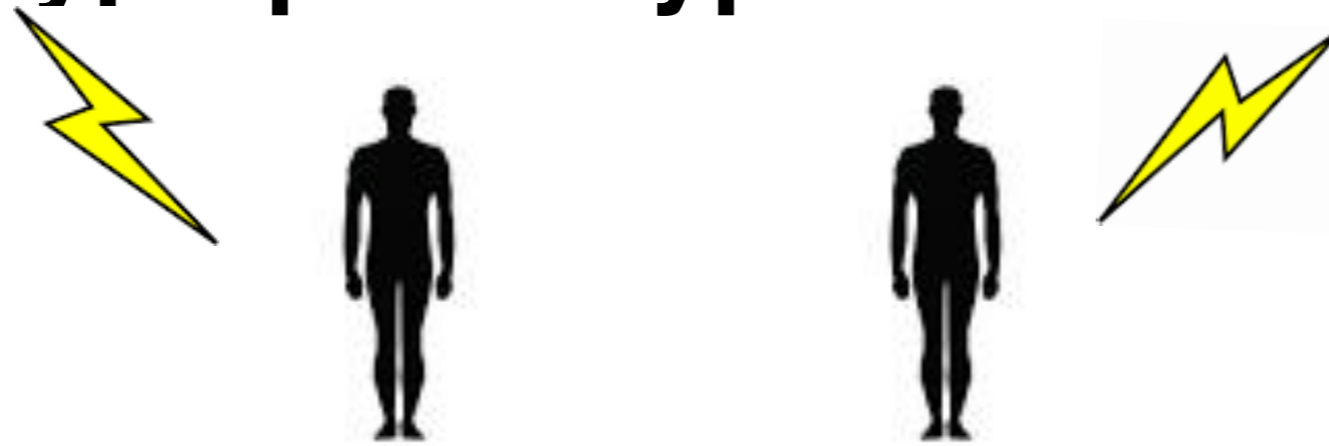
sphingolipid metabolic process




# PSAP and GBA are binding partners



# Genotype-phenotype association?



Exact same *GBA* mutation

A black arrow pointing downwards from the text "Exact same *GBA* mutation" towards the two photographs below.

What is responsible for the differences in clinical manifestations?

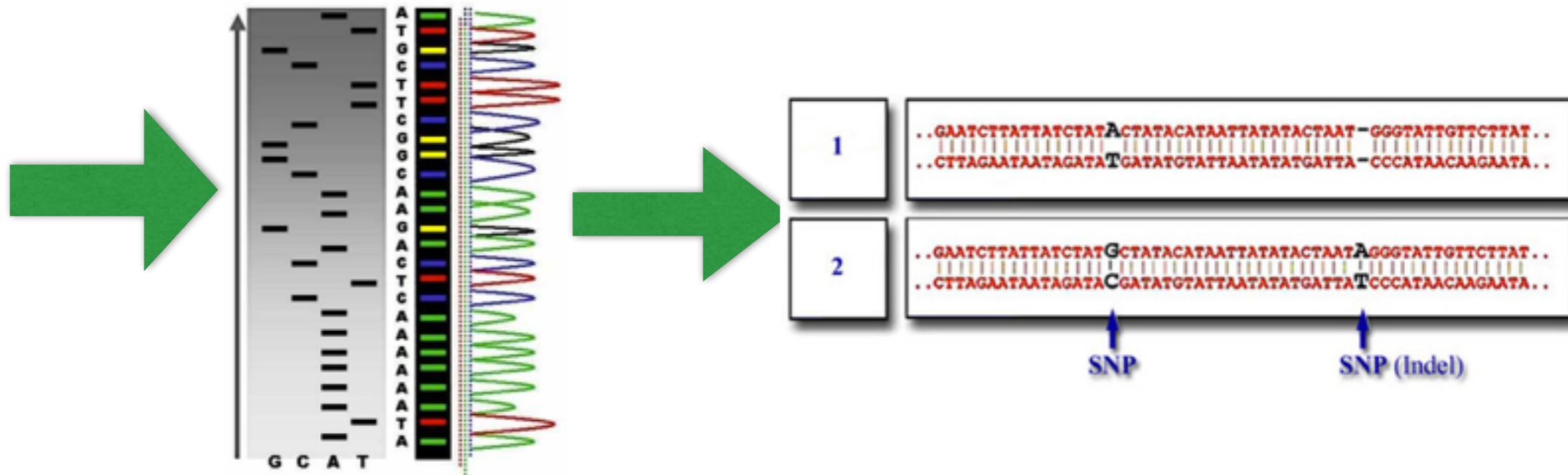
**Hypothesize that variations in *PSAP* contribute to the phenotypic variability in GD patients with same *GBA* mutations**



# Aim 1: Genome sequencing to find variable genomic regions



Same GBA

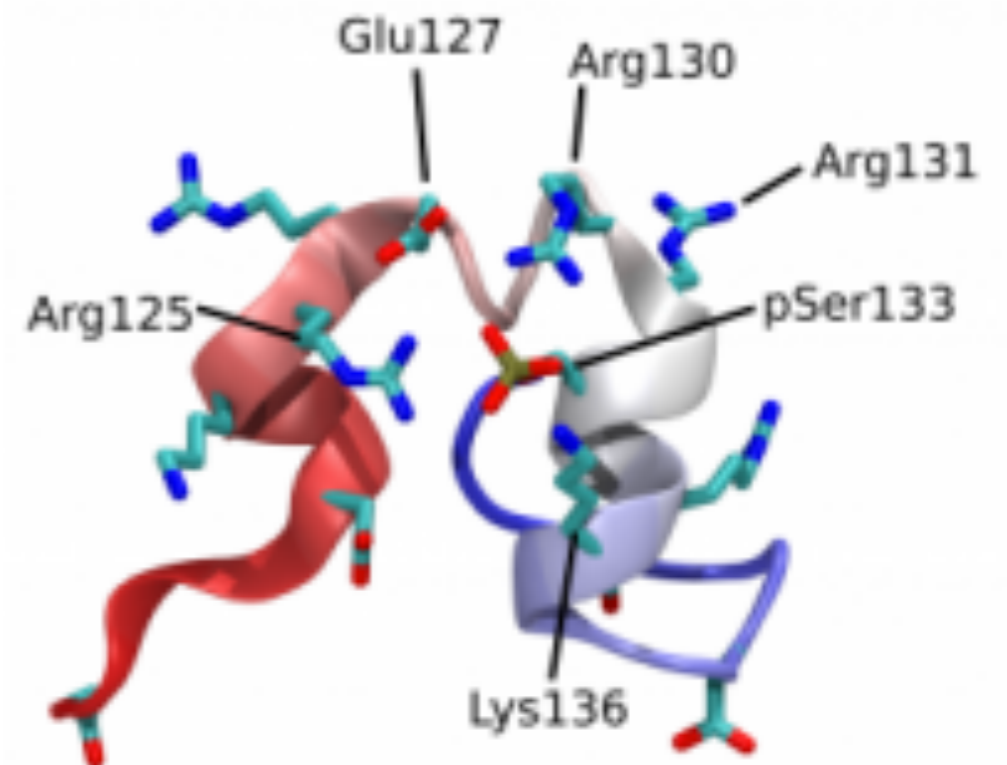


Hypothesis: If genomic differences are found, they may alter PSAP activity or expression

## Aim 2: Identify the phosphorylated amino acids in PSAP



**Same GBA**



Hypothesis: Differences in the phosphorylation of PSAP may result in changes of activity and expression

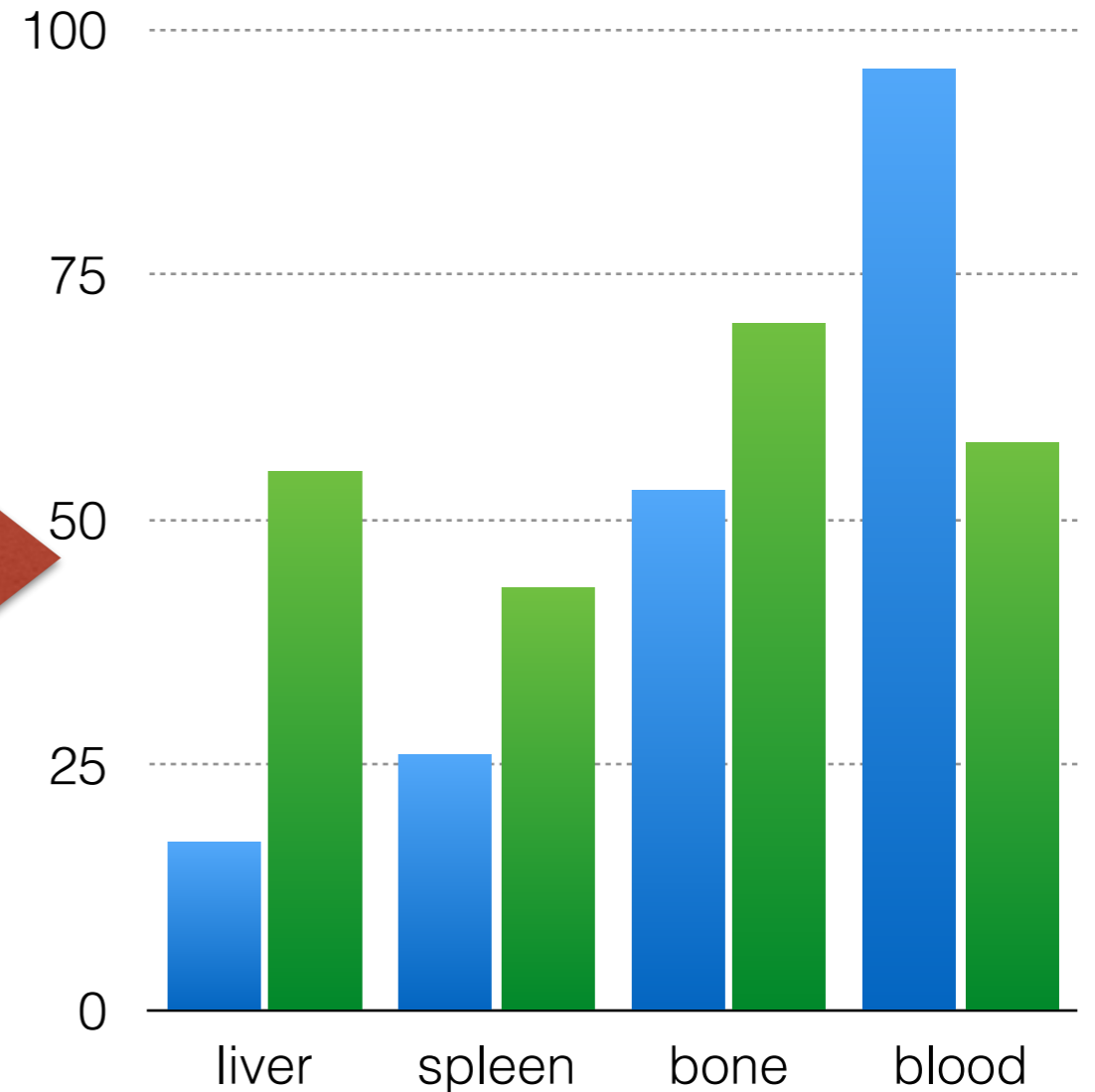
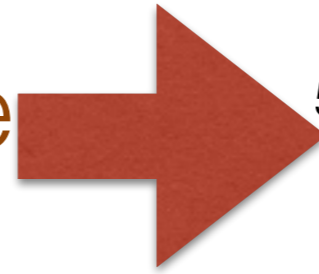
# Aim 3: Compare tissue specific levels of PSAP expression in GD patients



**Same GBA**



liver tissue



**Differences in expression?**

Hypothesis: Tissues that result in differences in symptoms will have irregular PSAP expression

# Payoff & Future?

**PSAP is a factor in disease symptoms**



# Questions?



<http://cliparts.co/people-animation>