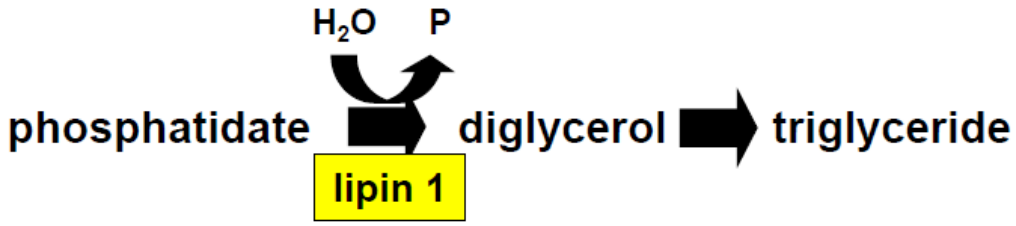
Skeletal muscle has abundant amounts of a protein called lipin 1 (LPIN1) that plays an important role in regulating metabolism. Lipin 1 is an enzyme that removes a phosphate molecule from a lipid called phosphatidate to make a new lipid called diacylglycerol. This is an important step in metabolism. For reasons that we don't fully understand yet, mutations in lipin 1 lead to episodes of severe muscle pain and damage (rhabdomyolysis) that can result in sudden death. This mutation affects mostly young children and the prevalence of this mutation, while rare, is increasingly being appreciated as a common cause of unexplained rhabdomyolysis in children.



The goal of my lab is to better understand the mechanisms by which loss of lipin 1 leads to muscle cell damage and by so doing, potentially develop new treatments that might help children diagnosed with this genetic disease. To do this, we study cells lacking lipin 1 in culture dishes and also use mice that that have been genetically-engineered to have mutations in lipin 1. We study how loss of lipin 1 affects their muscle and hope to better understand the basic biology of lipin 1 function. Much like people with lipin 1 mutations, we find that loss of lipin 1 in muscle of mice leads to damage of the muscle tissue. While we're making progress and believe that development of these mice will help in identifying a new treatment, there's still a great deal of work that needs to be done.

The research conducted by my lab is described on our website and you can find links to my email there. Please don't hesitate to contact us with any questions and we'll do our best to answer them.
Finck Lab website: https://gns.wustl.edu/about/faculty/brian-finck-phd/
Published research on the topic: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4484911/
Other resources:
http://neuromuscular.wustl.edu/msys/myoglob.html