Genetic variant impairs glycogen synthesis

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Glycogen is stored in skeletal muscles and liver and is of central importance as a first source of energy for muscle contractions, especially during high intensity exercise. Human genetic disorders primarily affecting skeletal muscle glycogen turnover are well-recognised, but rare.

In new research published in *PLoS Medicine*, Stephen O'Rahilly (University of Cambridge, UK) and colleagues describe the effect of a recently identified genetic variant – known as PPP1R3A FS – that affects glycogen turnover and other aspects of metabolism.

Confirming that the variant is common in the UK white population (in 1.46% of the 744 adults enrolled in the study), the findings identify PPP1R3A FS as the first prevalent mutation known to imp air glycogen synthesis and to decrease glycogen levels in human skeletal muscles.

In an accompanying perspective that discusses the study, Leif Groop and Marju Orho-Melander (both at Lund University, Sweden), who were not involved in the study, says that the study helps to “shed new light on the role of disturbed glycogen synthesis in disease pathogenesis.”

Citation: Savage DB, Zhai L, Ravikumar B, Choi CS, Snaar JE (2008) A prevalent variant in PPP1R3A impairs glycogen synthesis and reduces muscle glycogen content inhumans and mice. PLoS Med 5(1): e27.

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