

Upsetting a fragile alliance triggers a deadly childhood disease

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Scientists at the University of Malta and the Institut de Génétique Moléculaire de Montpellier (CNRS/Université de Montpellier) have shown that fruit flies and brewer's yeast can reveal clues about the cause of Spinal Muscular Atrophy (SMA), the most common genetic killer of infants.

SMA is a devastating [neuromuscular disorder](#) that robs children of their ability to walk, eat, or breathe. Mostly caused by an inherited flaw in the Survival Motor Neuron (SMN) gene, SMA is presently without a cure. A key reason is the lack of detailed information on how the SMN protein works in living organisms.

Using extensive genetic manipulations, the research team found that SMN forms an alliance with a set of diverse proteins known as Gemins. So incredibly fragile is this alliance that it can be broken if the perfect balance in [protein levels](#) is upset. The consequences are catastrophic. They range from death in flies to muscles that are too weak to support flight. The breakthrough discovery, which was published in the journal *PLOS ONE*, strengthens the fight against SMA.

"Our study is the first to show that the special relationship between SMN and Gemins exists in a living model system," said the study's lead author Ruben Cauchi, PhD, a senior lecturer at the University of Malta Faculty of Medicine & Surgery. "Furthermore, what we see in Game of Thrones is intriguingly true in cells. Upsetting the delicate balance of power leads to grave repercussions," he added with a smile.

SMN in partnership with Gemins is thought to have a role in assembling the constituents of the gigantic machine that edits messenger RNA, the genetic mail carrier of instructions for building proteins. Whether a fault in this process is to blame for the neuromuscular problems experienced by patients is still an open question.

Cauchi's team recently showed that a selective deficiency of Gemins within the motor system leads to flies with similar problems as when SMN levels are reduced. These findings coupled with those in the present study, show that a collapse of the SMN-Gemins alliance is responsible for Spinal Muscular Atrophy.

Right now the researchers are hunting for friends and foes of the alliance to find a therapy. "Current treatments being developed are based on boosting SMN levels. Broadening the therapeutic targets is essential to develop an effective treatment. Model organisms, like flies and yeast, hold the key for the successful implementation of this strategy," remarked Rémy Bordonne, PhD, CNRS Principal Investigator and study co-author.

Provided by University of Malta

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