Drosophila Mutant Holdup Arises from a Mutation in a Conserved Gene Required for Muscle Integrity

Tang, Lien

Past studies indicated that Drosophila (fruit flies) share 75% of the genes that cause diseases with humans. Thus by studying fruit fly genetics, one can learn about human genetics and possibly gain a better insight into diseases that affect both humans and fruit flies. Past projects revealed that CG6700 is the gene that contains a mutation known as “hold up” (hup), which was found to affect the fruit flies’ fertility and muscle structure. This year, the project focuses on confirming the identity of the hup gene through a genetic rescue, determining how the gene causes muscle defects utilizing cryosectioning, and finding out whether it is found in other species using a “BLAST search.” Observations of the genetic rescue showed a low yield of fruit fly embryos being produced and no transgenic flies in the offspring. As a result, further experiments on the genetic rescue will be required to validate the identity of the hup gene. Results from the cryosections revealed muscle defects in adult and pharate hup mutants, indicating that the defects begin during fruit fly development. The “BLAST search” revealed that the gene CG6700 is found in humans and other mammals such as mice. Furthermore, similarities at the end of the amino acid sequence between CG6700 and a human gene, LENG8, which is involved in RNA transport, were found, thus giving some insight into why the hup mutation causes muscle defects based on the amino acids located there. In addition, the sequence in CG6700 is conserved in a protein family called SAC3_GANP, which controls trafficking, secretion, and transport inside the cell. This may explain the muscle defects caused by hup, as the mutation may affect the transportation of proteins that are essential for muscle structure and development.

Awards Won: