

Fruit fly research may lead to better understanding of human heart disease

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Researchers at the Burnham Institute for Medical Research (Burnham) have shown in both fruit flies and humans that genes involved in embryonic heart development are also integral to adult heart function. The study, led by Rolf Bodmer, Ph.D., was published in *Proceedings of National Academy of Sciences*.

Dr. Bodmer's lab has discovered that in the fruit fly *Drosophila*, interactions between cardiac nmr genes (TBX20 in humans) and other transcription factors, are involved in regulating cardiac performance, rhythm and heart muscle structure. TBX20, along with other congenital heart disease genes, has been previously shown to be critical to the development of the embryonic heart first in flies and subsequently in mouse models.

However, this study is the first indication that nmr/TBX20 also plays a role in adult heart function. These genes are highly conserved from flies to humans and Bodmer's research showed that some human individuals with structural congenital heart abnormalities, as well as problems with heart function, including arrhythmias and heart failure, also exhibited TBX20 mutations.

"These studies demonstrate that *Drosophila* has potential as a model system for exploring the genetics underlying human heart disease and for identifying new candidate genes that potentially cause heart disease," said Dr. Bodmer.

To make the connection between human and *Drosophila* heart malfunction, human subjects with structural congenital heart disease, as well as heart muscle dysfunction were examined. In 96 human subjects with clinical evidence of dilated cardiomyopathy (this causes a weakened heart that cannot pump blood efficiently), DNA analysis identified three different variants of the gene TBX20, suggesting TBX20 may be involved in the development of cardiomyopathy. In addition, TBX20 variants were identified in four children with atrial septal defects.

Source: Burnham Institute

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