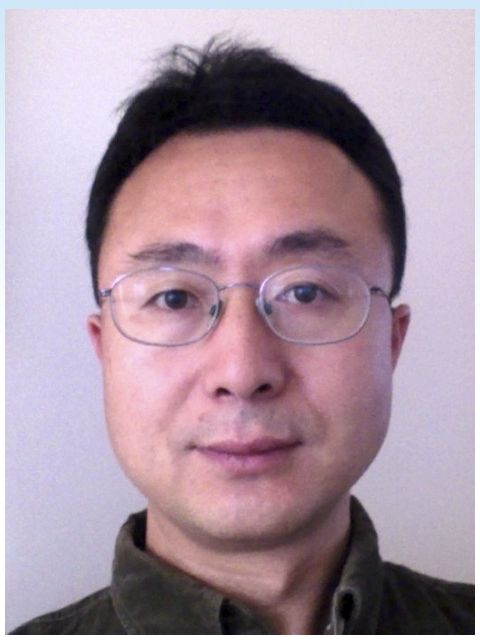
香港浸會大學
HONG KONG BAPTIST UNIVERSITY

School of 中醫藥學院
Chinese Medicine

Genetic analyses of essential splicing factor genes in C. elegans

(關鍵剪接因子功能的遺傳學分析)



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Facilitator：Dr. Li Min

Abstract

RNA splicing is a fundamental regulatory mechanism for eukaryotic gene expression. Mutations in essential RNA splicing factors could lead to severe diseases such as spinal muscular atrophy (SMA). It remains to be understood how these splicing factors interact *in vivo* and why mutations in these factors cause variable diseases. Recently we isolated conditional mutations affecting the U2AF large subunit (UAF-1), splicing factor 1 (SFA-1) and a novel splicing factor MFAP-1 in *C. elegans*. We found that UAF-1 and MFAP-1 interact to affect alternative splicing. Using an endogenous splicing reporter, we found that the *C. elegans* SMA-related gene *smn-1* is required for splicing at weak 3' splice sites. Surprisingly, we found that conditional mutations of UAF-1 could reverse the lifespan and behavioral defects of *smn-1* loss-of-function mutants, providing novel genetic evidence that RNA splicing defects underlie SMA. We hope our studies will provide new understanding about the functions of these factors and insights into the molecular mechanisms of related diseases.

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