Consiglio Nazionale delle Ricerche
Istituto di Genetica Molecolare

SIBBM

7th ARTURO FALASCHI LECTURE

Lunedì 22 Maggio 2017
ore 11:30

AULA 1
Nuovo Polo Didattico Cravino
Via Ferrata 9 - 27100 PAVIA

Prof. Adrian Krainer

Cold Spring Harbor Laboratory
Cold Spring Harbor – NY - USA

"Nusinersen (SPINRAZA): the first FDA-approved treatment for SMA"

Adrian Krainer's lab studies the mechanisms of RNA splicing, ways in which they go awry in disease, and the means by which faulty splicing can be corrected. In particular, they study splicing in spinal muscular atrophy (SMA), a neuromuscular disease that is the leading genetic cause of death in infants. In SMA, a gene called SMN2 is spliced incorrectly, making it only partially functional. The Krainer lab is able to correct this defect using a powerful therapeutic approach. It is possible to stimulate protein production by altering mRNA splicing through the introduction of chemically modified pieces of RNA called antisense oligonucleotides (ASOs) into the spinal cords of mice. Previously, using ASOs in mice carrying a transgene of human SMN2, they developed a model for SMA using a technique they called TSUNAMI (shorthand for targeting splicing using negative ASOs to model illness).
Recently, this method has been approved by the U.S. Food and Drug Administration to treat children and adults with spinal muscular atrophy (SMA).
The Krainer lab has also worked to shed light on the role of splicing proteins in cancer. They have found that the splicing factor SRSF1 functions as an oncogene stimulating the proliferation of immortal cells. They found that SRSF1 can actually stop cell growth by stabilizing a powerful tumor suppressor protein, called p53—suggesting that the cell is responding to the aberrant SRSF1 activity. This discovery offers insight into how tumors arise and the pathways that lead to transformation.

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