Istituto Nazionale di Neuroscienze
Consorzio Interuniversitario di Neuroscienze

UNIVERSITÀ DEGLI STUDI DI TORINO

Dipartimento di Neuroscienze 'Rita Levi Montalcini'

NICO Neuroscience Institute Cavalieri Ottolenghi

INN Open Neuroscience Forum

May 10, 2018 h 2:00 p.m.
Seminar Room – Neuroscience Institute Cavalieri Ottolenghi
Regione Gonzole 10, Orbassano (TO)

TTC3: A DOWN SYNDROME GENE INVOLVED IN NEURONAL MIGRATION

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Abstract

Down syndrome (DS) is caused by trisomy of Human Chromosome 21 (HSA21). The clinical manifestations of DS vary in both penetrance and intensity among affected individuals, but the only hallmark common to all patients is intellectual disability (ID).

DS-associated ID is the result of developmental brain abnormalities, leading to altered neuronal migration and connections within the cortex. One likely candidate for these phenotypes is TTC3, a HSA21 protein, whose expression is increased in cells derived from DS experimental models and from DS patients. Moreover, up-regulation of TTC3 inhibits neuronal differentiation by modulating actin cytoskeleton. In this last years, we are studying the effect of TTC3 over-dosage in cortical development, up-regulating the protein using *in utero electroporation* and we demonstrated that TTC3 levels are critical for correct neuronal positioning within the cortex. These data support the hypothesis that DS-cortical phenotypes can be ascribed to the increased dosage of TTC3.

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