



# UNIVERSITÀ DEGLI STUDI DI MILANO

## DIPARTIMENTO DI SCIENZE FARMACOLOGICHE E BIOMOLECOLARI

Milan 15-2-2018 International chromosome 15 day

A day when all organizations and families gather together and raise awareness about Angelman

INTERNATIONAL  
**ANGELMAN**  
DAY FEB 15



*"Rare diseases are rare, but rare disease patients are numerous"*



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### Il cromosoma 15

- I due cromosomi 15 rappresentano quasi il 3,5% del DNA totale nelle cellule umane.
- Il cromosoma 15 contiene oltre **800** geni ed il numero di polimorfismi a singolo nucleotide (SNP) è di oltre **325.000**

Alcune patologie legate a problemi del cromosoma 15:

Acidemia isovalerica  
Cancro alla mammella  
Malattia di Tay Sachs  
**Sindrome di Angelman**  
Sindrome di Marfan  
Sindrome di Prader Will  
Sordità sindromica



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**Neuronal Death and Neuroprotection Lab**



### **Angelman Syndrome**

"happy puppet syndrome"

Angelman syndrome affects an estimated 1 in 12,000 to 20,000 people, a rare disease.

Angelman syndrome is a genetic disorder due to the loss of function of a gene called UBE3A, with characteristic features that include severe speech impairment, developmental delay, intellectual disability, microcephaly, epilepsy and ataxia (problems with movement and balance).

Angelman is a complex genetic disorder that primarily affects the nervous system interestingly both copies of UBE3A are active in all of the body's tissues, in the brain, only the copy inherited by **the mother** is active (according to a phenomenon called genomic imprinting).

The life expectancy for individuals with Angelman syndrome is normal and there is no specific therapy for Angelman syndrome. Medical therapy for seizures is usually necessary.

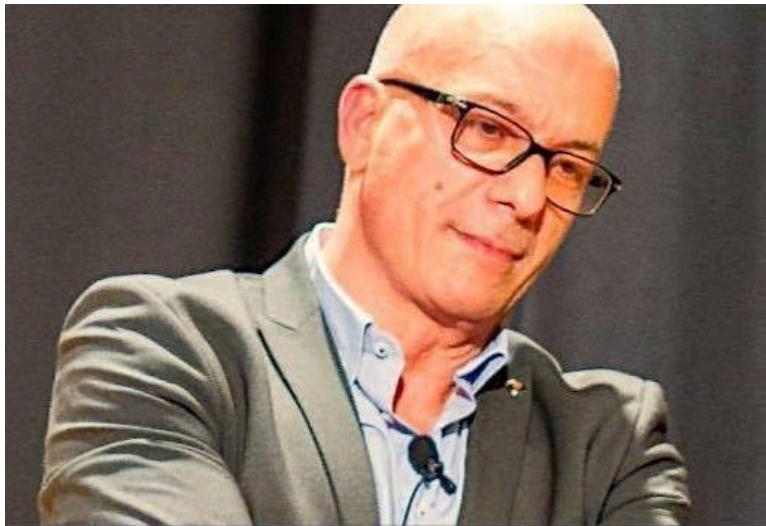


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**Dr. Tommaso Prisco**

Presidente dell'ORSA



GIORNATA INTERNAZIONALE  
SINDROME DI ANGELMAN  
15 FEBBRAIO

**Professor Ype Elgersma**

Department of Neuroscience and Director of the  
*Expertise Center for Neuro-developmental Disorders* -  
NETHERLANDS

