



# 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



### Professor Ype Elgersma

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



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