



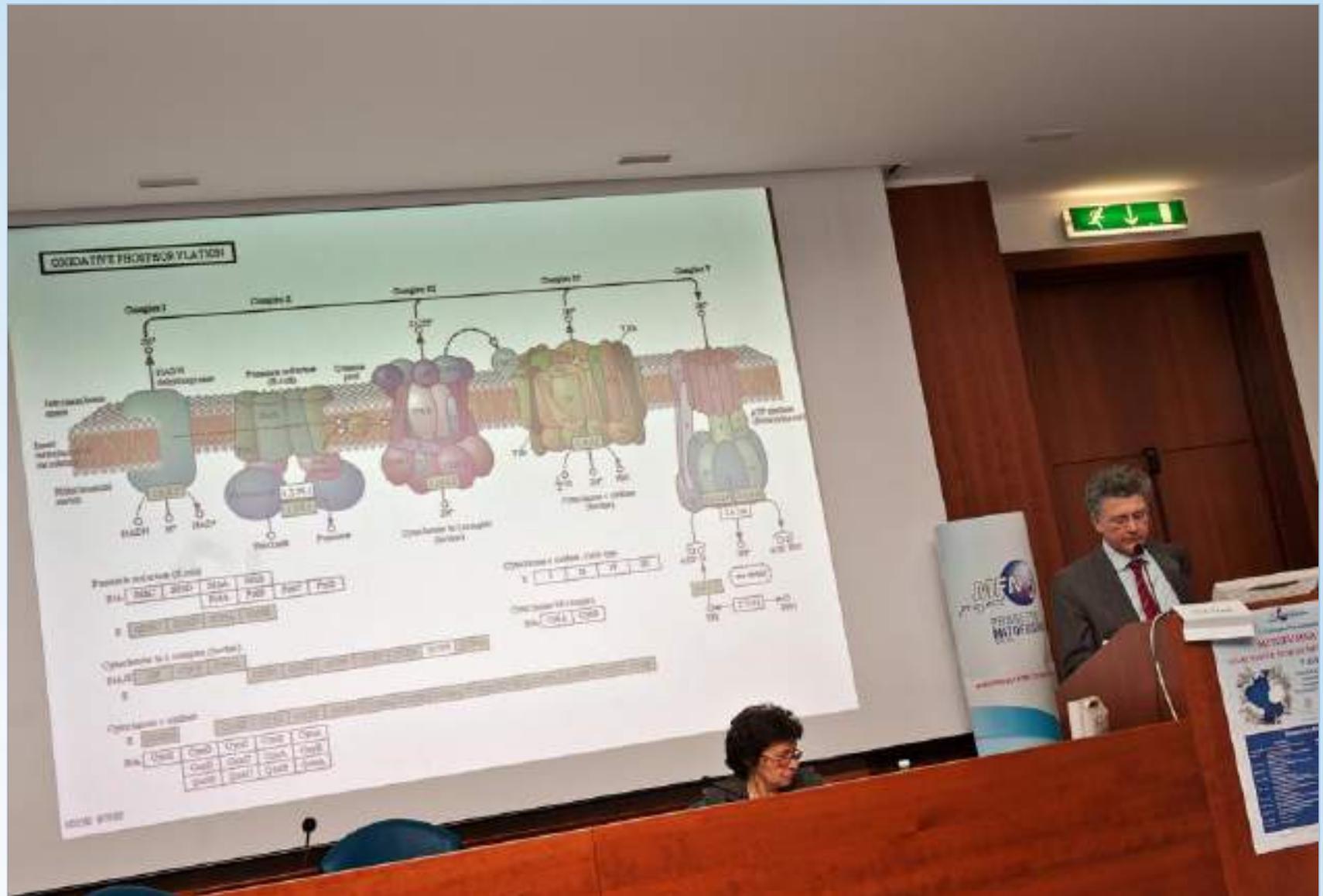
*The audience in the Lecture Hall*



*Professor Nereo Bresolin*



*Professor Giacomo P Comi*



*...during his speech about “Mitofusin 2 , fusion e mitochondrial dysfunction”*

# CMT2A and MFN2

Michael E Shy MD  
Professor of Neurology, Pediatrics and Neurophysiology  
Carver College of Medicine  
University of Iowa  
University Clinical Center University of Iowa



***Professor Bresolin introducing Professor Shy in teleconference***



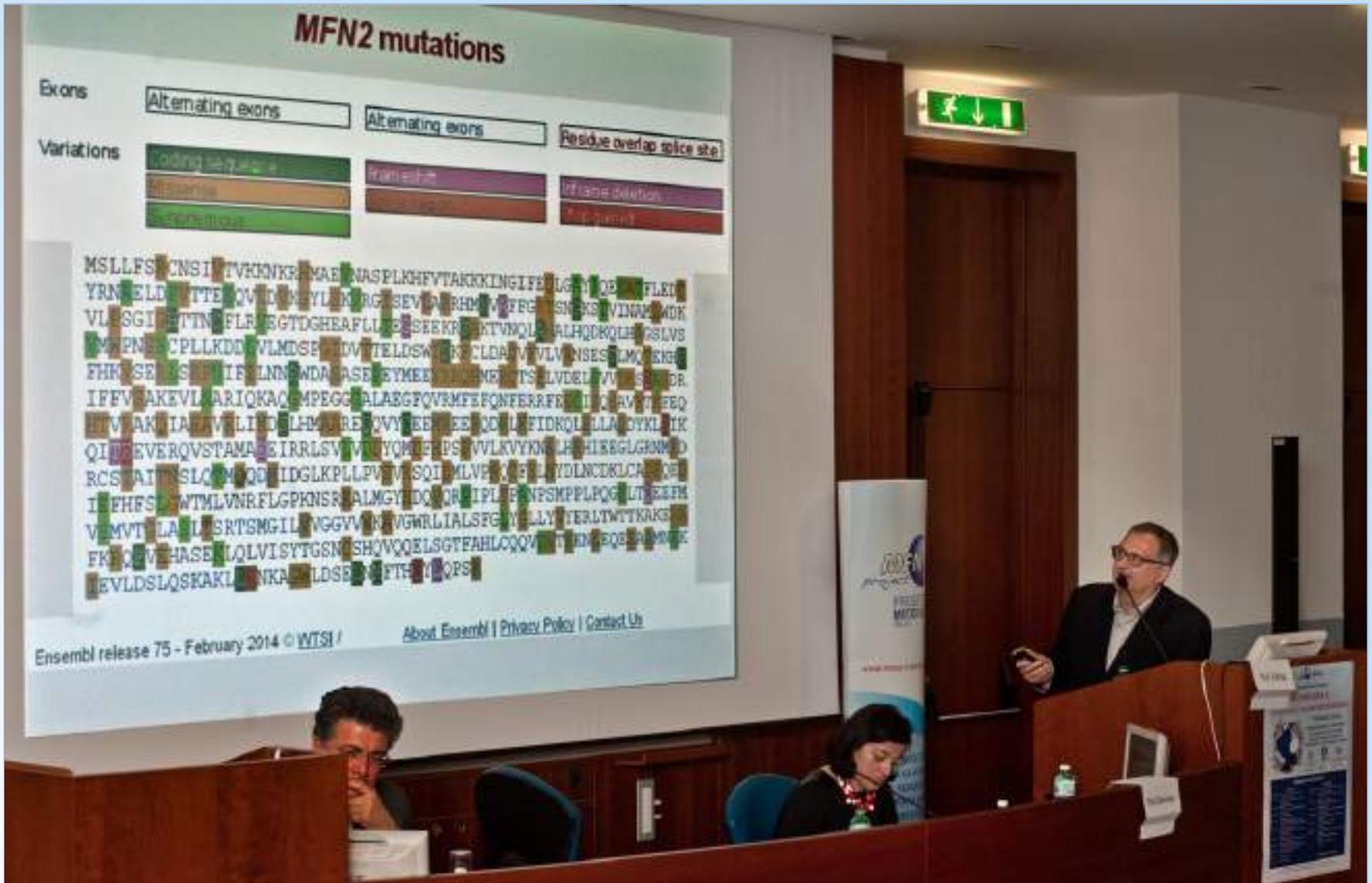
*Professor Maurizio Moggio*



***Dr Angelo Quattrini***



*Professor Gian Maria Fabrizi...*



*...during his speech “Pathogenic variants and polymorphisms in MFN2 gene”*



*Professor M. Luisa Mostacciolo...*

## General questions

- Is it possible to identify mutational hot spots in the MFN2 gene?
- Is it possible to make a direct genotype-phenotype correlation?
- Are variable expressivity and incomplete penetrance present in the same families with CMT2A?
- Does the age of onset correlate with disease severity?
- Is it sufficient to identify a mutation in one affected for family to consider this mutation as causative?

*...during her speech “MFN2 gene mutations”*



*Professors Comi, Bresolin e Bonneau chairpersons in the first Session*



*During the Lunch...*



*During the Lunch...*





*Dr Bergamin from London*



*Dr Stefania Corti*

# CHARCOT-MARIE-TOOTH TYPE 2A

**Definition:**  
classic axonal peripheral sensorimotor neuropathy

**Causative Gene:**  
Mitofusin 2 (MFN2) gene

**Onset:**  
first or second decade

**Inheritance:**  
autosomal dominant (most frequent cases)  
autosomal recessive (very rare)  
sex chromosome-related (10-15% of all cases)





*Dr D'Angelo*



*Audience*



*Audience*



*Dr Dell'Urso*



*Professor Braathen from Norway*



*Some charter members with some supporters of Mitofusin 2 Project*



*The French delegation*



*Our girls at reception*