



*The audience in the Lecture Hall*

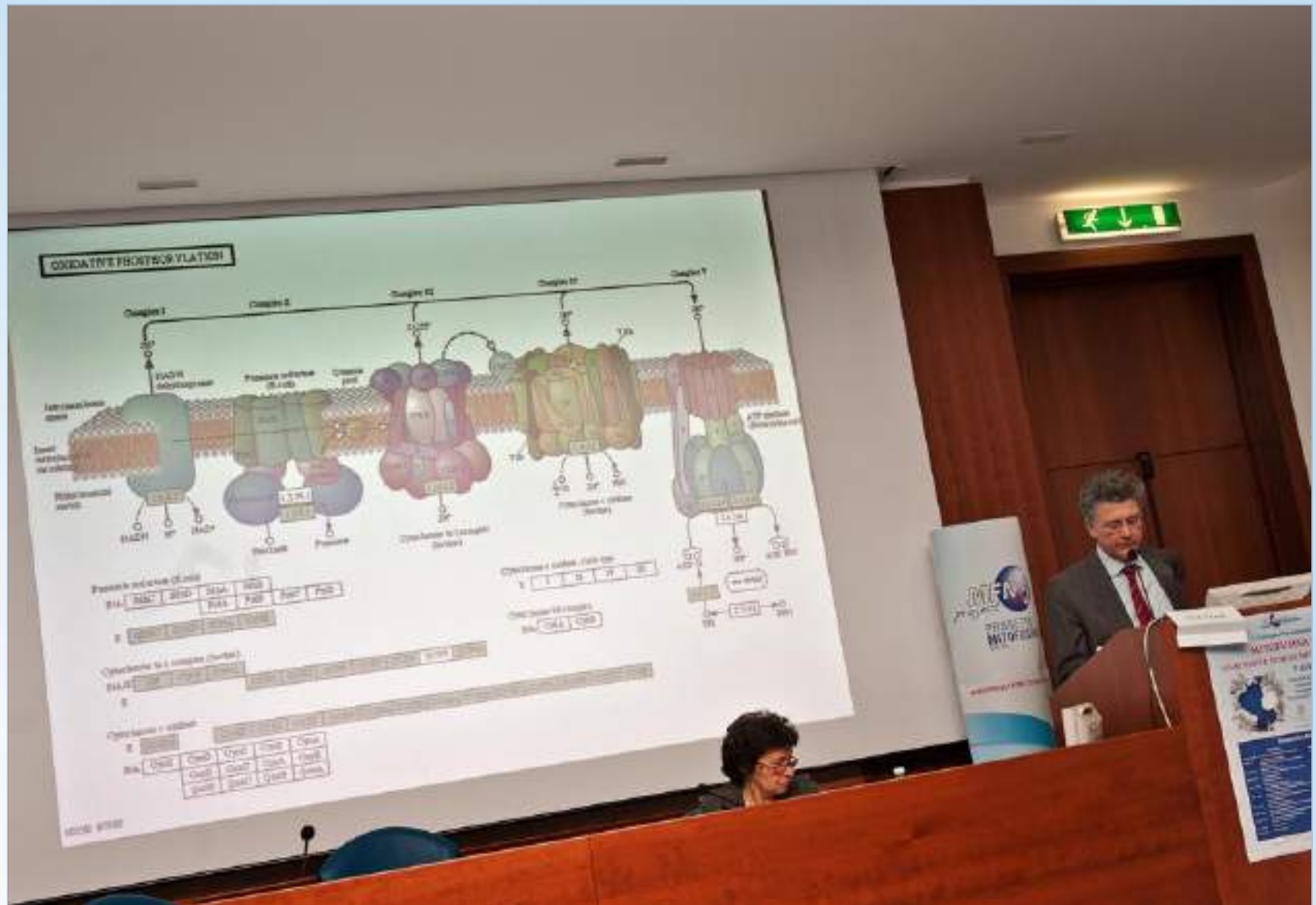


*Professor Nereo Bresolin*



***Professor Giacomo P Comi***





*...during his speech about “Mitofusin 2 ,fusion e mithocondrial 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***





***DrAngelo Quattrini***



*Professor Gian Maria Fabrizi...*



# MFN2 mutations

Exons

Variations

Alternating exons

Alternating exons

Residue overlap splice site

Coding sequence

Insertion

Deletion

Frameshift

In-frame deletion

Not found

MSLLFS CNSI TVKQNKHMAE NASPLKHVFTAKKKINGIFEDG N QP FLED  
 YRNELD TTE QVIDNHYLDMRG SEVDA RHM VFFG SN KS VINAM MDK  
 VL SGI ETTN FLEEGTDGHEAFLLE SEEEKR KTVWQL ALHQDKQLH GSLVS  
 MPNS CPLLKDE VLMDSP IDVTIELDSW KCLDAV VLV NSES LM KEGE  
 FHK SE SS IFELNNWDAAASE EYMEEL DNEETS LVDEL VV SS DR  
 IFFVSAKEVLARIQKAC MPEGC ALAEGFQVRMFFEFQNFERRAFELIQAVTSEFQ  
 ITVAKLIAEVLILDLHMA REGVYEE EEE QDL FIDKQLLLA DYKL IK  
 QI SEVERQVSTAMAGE IRRLSV VVLYQHEEPS VVLKVYKML HIKEGGLGNWD  
 RCSTAITSLQMDQ IDGLKPLLPTV SQIDLVV EFLVYDLNCKLCA QD  
 IEFHFS WTMLVNRFLGPKNSR ALMGYDQQR IPLPNTSMPPLPQGLT EEM  
 VMVT LA IL SRTSMGIL VGGVVR VGVRLIALSFG YLLY TERLTWTTKAGS  
 FKQQSVHASE LQLVISYTGNSHQQVQELSGTFEHLQQQV TLM EQSEHMMK  
 EVLDSLQSKAKL ENKALDSE VFTH YQPS

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...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...*





*Professor Bonneau*



***Dr Bergamin from London***



***Dr Stefania Corti***



# CHARCOT-MARIE-TOOTH TYPE 2A

## Definition:

classic axonal peripheral sensor-motor 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*