### 1 Intro



### 2 Coping with COVID-19





# [partecipanti, persone che salutano, si sistemamano]

#### TESTO:

**40 experts** from almost all **Italian centers** involved in the care of patients with **inherited neuromuscular diseases** met during the Covid-19 pandemics to understand how to progress the research to improve treatment for such patients.

#### TESTO:

The meeting was an opportunity to discuss the strategies adopted by each center on how to ensure continuity of care in these unprecedented times. Indeed, during the lock-down all centers were able to assist their patients, also by quickly adopting **online support to patients and telemedicine**.

[voce di Gualandi che dice: "ovviamente questo ha accelerato..."]

#### TESTO:

As the clinical work continued, **also the research continued**...



#### TESTO:

Professor Alessandra Ferlini from the University of Ferrara reported that the DMD Italian Network published in the March issue of Frontiers in Genetics. The paper showed the result of a 10-year collaborative effort to map DMD/BMD genotype landscapes and mutation frequency in the Italian population affected by dystrophinopathies.







#### TESTO:

Among various interesting findings, the study found that 63% of Italian patients with out-of-frame deletions are eligible for exon skipping therapy.

The findings of the DMD Italian Network have important implications for clinical research and for the design of clinical trials.

[sovrascritta: 63% of Italian patients eligible for exon skipping therapy]

#### TESTO:

As Professor Eugenio Mercuri from Policlinico Gemelli in Rome illustrated, so far, no study evaluated systematically the roles genes as disease modifiers. Indeed, in DMD, the genotype/phenotype correlation has an important impact on the disease progression and has implications for treatment.

### 7 Prof Mercuri 2



#### TESTO:

An older study by Pane and colleagues found that boys with duplications had better results than those with the other types of mutations, while **boys eligible for skipping of the exon 44 had better baseline results and less drastic changes than those eligible for skipping exon 45 or 53**.

#### TESTO:



Prof Mercuri 3

 The impact of genotype on disease progression was even more evident in longterm follow-up studies such as the Telethon supported longitudinal multicentric cohort involving 12 tertiary neuromuscular centers in Italy, 1 center in Belgium and one in UK followed patients with deletions in the DMD gene amenable for skipping exons 44,45, 51 and 53 for at least three years.

Brogna et al, su



#### TESTO:

The study allowed to identify how the variation among groups increased with the duration of follow-up and an analysis of individual data showed that variability within groups depended on **age at baseline**, **baseline score**, or **loss of walking ability** when the patient was 3 years of age.

[three-year follow-up]

### 10 Prof Mercuri 2



### TESTO:

Professor Mercuri highlighted how long follow-up is crucial to understand the progression if the disease and the efficacy of therapeutic options.

[qui si può mettere anche la voce di Mercuri che dice la frase sopra e le persone che dibattono]

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### Prof Comi 1



#### TESTO:

In conclusion, Professor Giacomo Comi from Milan provided and overview of genetic studies on **limb-girdle muscular dystrophy**, a genetically inherited condition that primarily affects skeletal muscle leading to progressive, predominantly proximal muscle weakness at presentation caused by a loss of muscle fibers.





#### TESTO:

In the Italian centers this condition is wellknown as is its molecular characterization, and, in the future, a **long-term genotype phenotype follow-up** study will improve the understanding of disease progression for the benefit of these patients.

## 13 Conclusions 2



#### TESTO:

The nearly three-hour web meeting was characterized by high interaction among participants and showed how the Italian community is actively involved in research and care of dystrophinopathies.



### **Conclusions 2**



#### TESTO:

And that research continues at a distance... with the promise to meet soon, in person.

[persone che salutano]