

INTERDISCIPLINARY AND MULTIPROFESSIONAL INTERVENTION PLAN IN CHILDREN WITH DUCHENNE MUSCULAR DYSTROPHY THE EXPERIENCE OF THE “ROBANESCU-PADURE” NATIONAL UNIVERSITY CENTER FOR CHILDREN NEUROREHABILITATION

*Madalina Leanca^{1 ■ 2}, Elena Neagu¹, Ileana Georgescu¹, Raluca
Petcu^{1 ■ 2}, Georgiana Nicolae^{1 ■ 2}, Andrada Mirea^{1 ■ 2}*

*¹ National University Center for Children Neurorehabilitation
“Robanescu-Padure”, 44 Dumitru Minca Street, 041408 Bucharest, Romania;
(NCHCN)*

*² University of Medicine and Pharmacy “Carol Davila”, 37 Dionisie Lupu
Street, 020021 Bucharest, Romania;*

Disclosure statement

I have no actual or potential conflict of interest in relation to this program/presentation.

✓ PERSONALIZED HEALTH SERVICES

Who we are?

✓ Subacute diseases

- ✓ Traumatic brain/spinal cord injury
- ✓ Burns
- ✓ Nervous System Infection
 - ✓ Meningitis
 - ✓ Encephalitis
 - ✓ Myelitis

PRECISION AND ETIOPATHOGENIC
DIAGNOSTIC

NEW CURRENT STRATEGY
USE AND EFFECTS OF HIGH EFFICACY TREATMENT

✓ Chronic diseases – various etiologies

- ✓ Genetic Rare Diseases
- ✓ Malformation – cerebral, orthopedic
- ✓ Metabolic/ Tumoral
- ✓ Autoimmune/ Inflammatory
- ✓ Vascular – stroke, cerebral thrombosis
- ✓ Perinatal events – prematurely, hypoxia
- ✓ Epilepsy

THE DEVELOPMENT OF SCIENTIFIC RESEARCH IN THE FIELD
OF CHILD RECOVERY

CHILDREN AND THE PANDEMIC

Who we are?

- ✓ **INTERDISCIPLINARY AND MULTIPROFFESIONALTEAM** doctors, nurses, physical therapist, psychologists, social workers
- ✓ Physical Medicine and Rehabilitation,
- ✓ Pediatric Neurology,
- ✓ Pediatric Orthopedy,
- ✓ Pediatria,
- ✓ Anesthesia and Intensive Care,
- ✓ Medical Genetics,
- ✓ Pediatric Psychiatry,
- ✓ Endocrinology,
- ✓ Epidemiology ,
- ✓ Laboratory Medicine,
- ✓ Radiology and Imaging.



DUCHENNE MUSCULAR DYSTROPHY (DMD)

- **DMD is a genetic muscle-wasting disorder caused by mutations in the DMD gene, which leads to little or no dystrophin protein. Without dystrophin, muscle cells are damaged easily and progressively weaken over time.**
- **It usually begins in early childhood, most often in boys, and mainly affects the skeletal muscles, then later the heart and breathing muscles. Common early signs include delayed walking, frequent falls, trouble climbing stairs, difficulty getting up from the floor, and enlarged calf muscles.**
- **DMD is X-linked, meaning the altered gene is on the X chromosome. It is one of the most common and most severe forms of muscular dystrophy.**



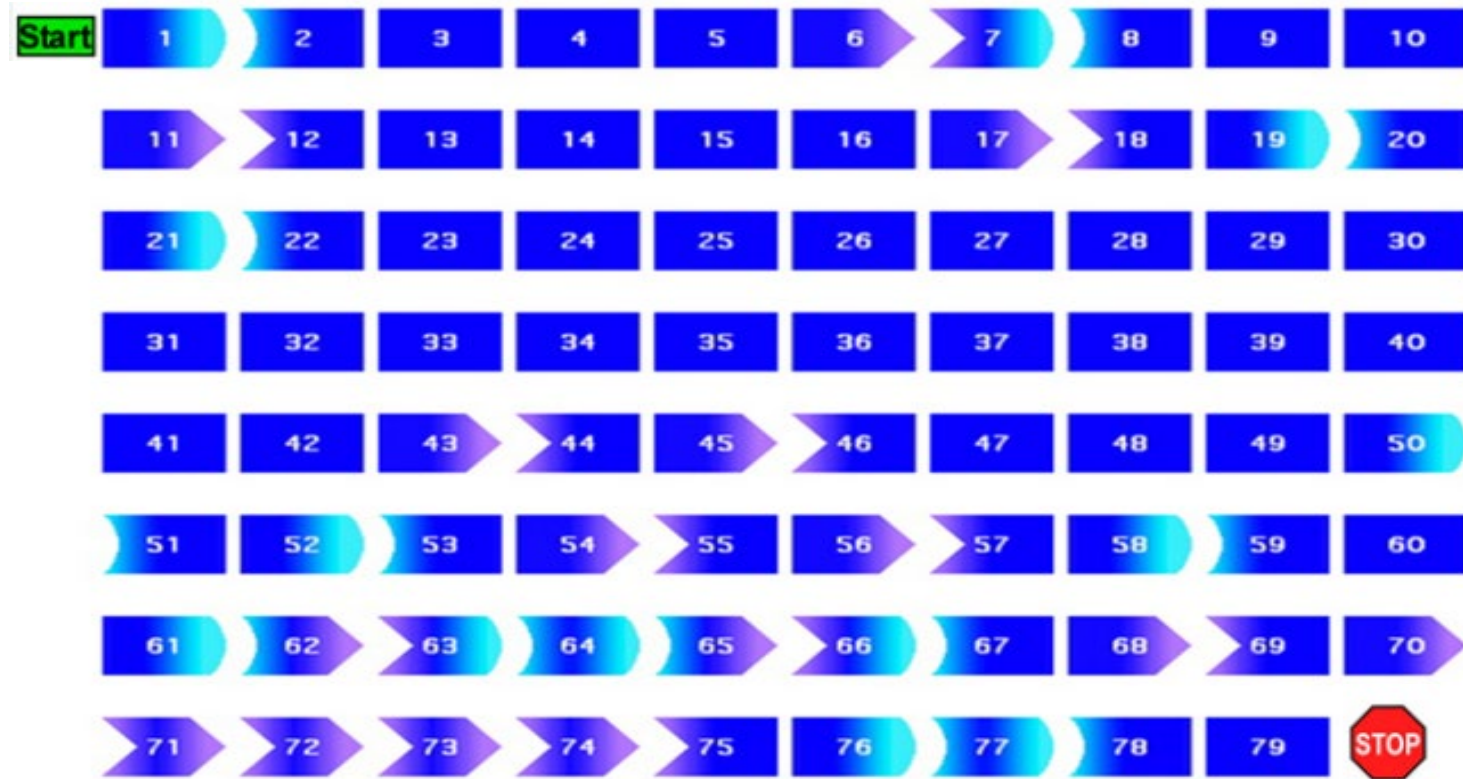
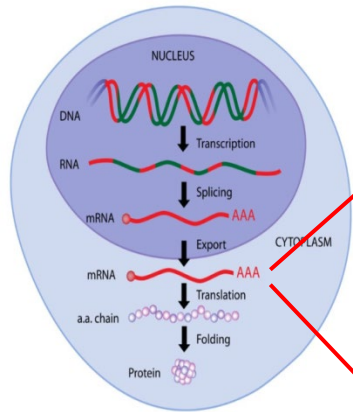
DUCHENNE MUSCULAR DYSTROPHY (DMD) (2)

DIAGNOSIS:

- **blood tests such as creatine kinase**
- **genetic testing MLPA Multiplex Ligation-dependent Probe Amplification DMD gene for deletion or duplication**

TREATMENT: focuses on slowing progression and managing complications

- **corticosteroids,**
- **physical therapy,**
- **cardiac and respiratory care and**
- **mutation-specific therapies when appropriate.**



Mutations — affecting a single nucleotide, a group of nucleotides, one exon, multiple exons, or the entire gene

Deletion (absence)
Duplication (addition)
Substitution (replacement)

Course of untreated DMD

Onset under 5 yrs of age

- Muscle weakness
- Contractures
- Loss of walking- 9.5 yrs
- Scoliosis
- Respiratory failure
- Cardiomyopathy
- Death in late teens



DMD care considerations 2018

Birnkrant et al. Lancet Neurol 2018; 17: 251–67, 347–61, 445–55

Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management



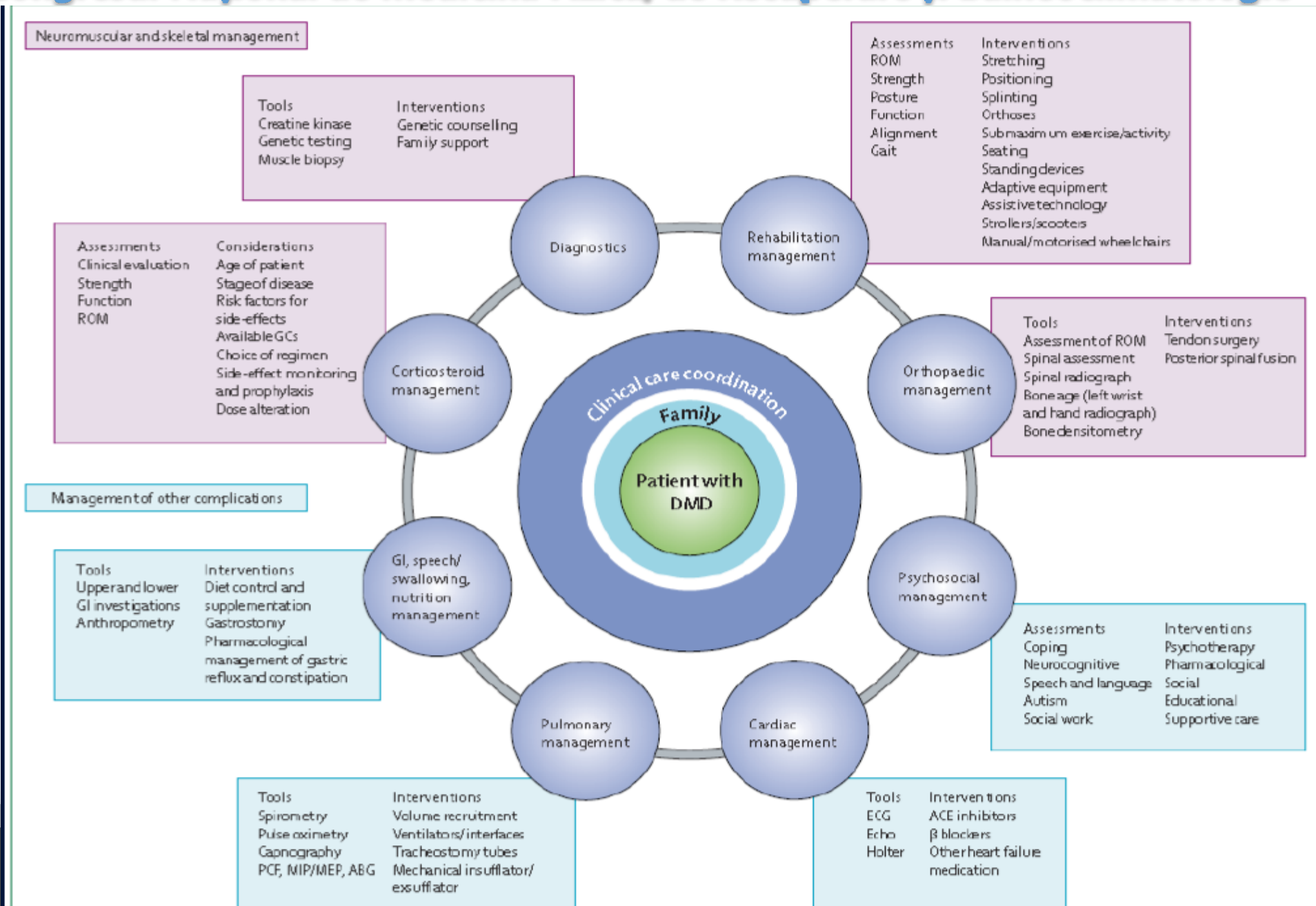
David J Birnkrant, Katharine Bushby, Carla M Bann, Susan D Apkon, Angela Blackwell, David Brumbaugh, Laura E Case, Paula R Clemens, Stasia Hadjiyannakis, Shree Pandya, Natalie Street, Jean Tomezsko, Kathryn R Wagner, Leanne M Ward, David R Weber, for the DMD Care Considerations Working Group*

Since the publication of the Duchenne muscular dystrophy (DMD) care considerations in 2010, multidisciplinary care *Lancet Neurol 2018; 17: 251–67*

Diagnosis and management of Duchenne muscular dystrophy, part 2: respiratory, cardiac, bone health, and orthopaedic management

Diagnosis and management of Duchenne muscular dystrophy, part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan

Congresul Național de Medicină Fizică, de Recuperare și Balneoclimatologie



Functional decline in Duchenne Muscular Dystrophy (DMD)



Congresul Național de Medicină Fizică, de Recuperare și Balneoclimatologie

Covasna, 23 – 25 Aprilie 2026



Multifactorial
movement analysis



Postural Assesment
T-BED



Dexa Scan

COMPLEX ASSESSMENT OF DMD PATIENT



Plantar pressure
assesment



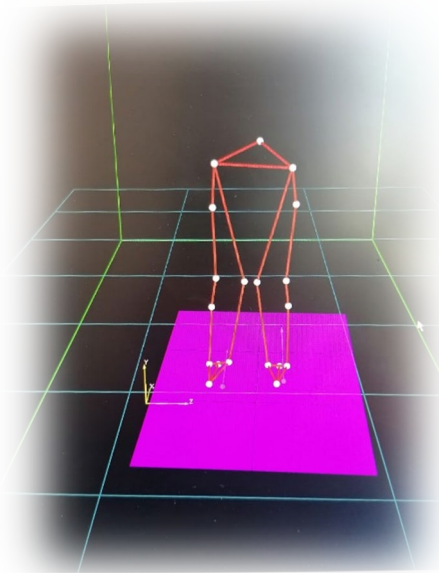
Spirometry



CARDIAC MRI

Multifactorial movement analysis

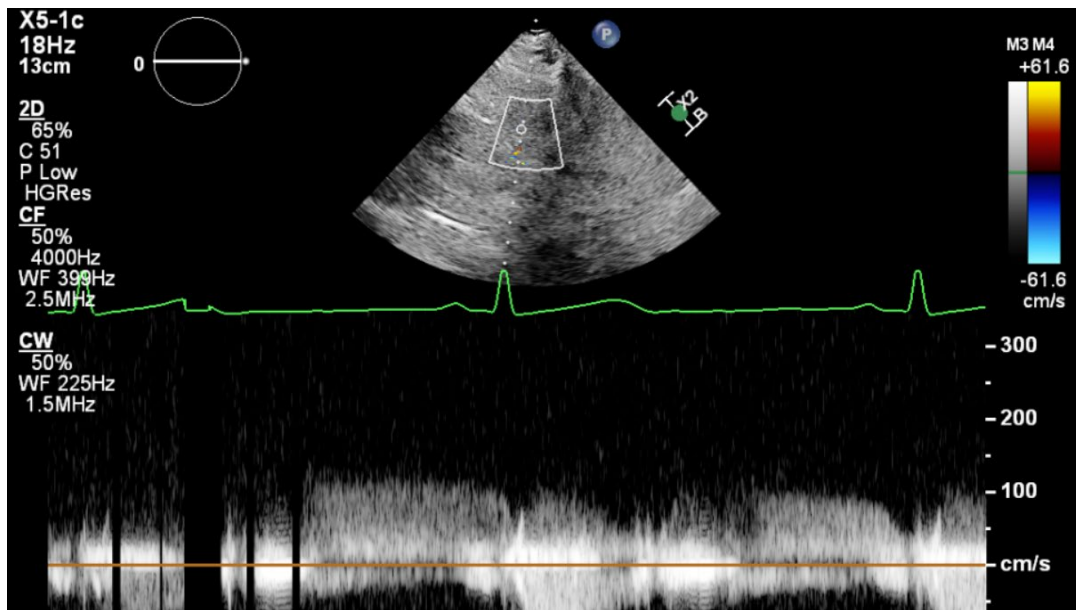
Ortesis



Patients should be regularly followed by a **pediatrician/pulmonologist** experienced in caring for patients with neuromuscular diseases



CARDIAC MANAGEMENT

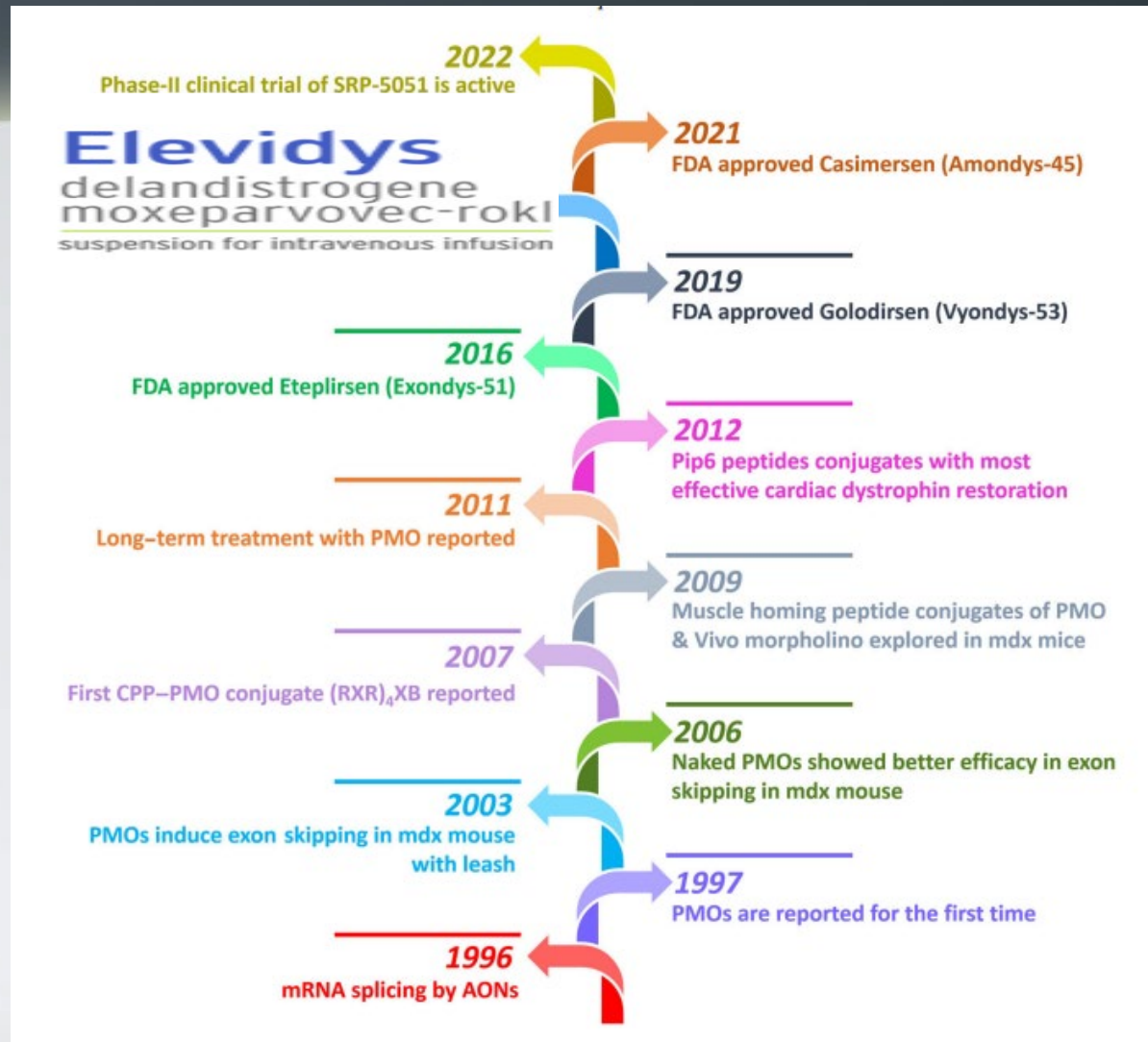


ULTRASOUND



CARDIAC MRI 3TESLA

THERAPEUTIC EMERGENCIES IN DMD

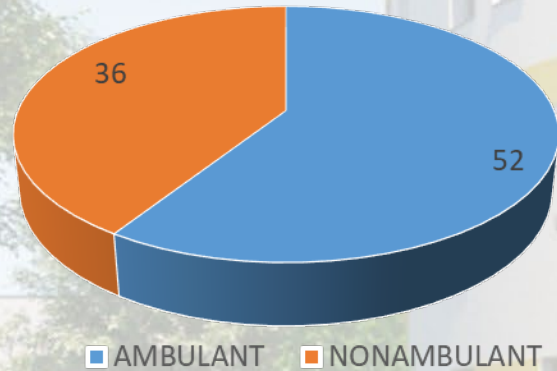


Gupta S, Sharma SN, Kundu J, Pattanayak S, Sinha S. Morpholino oligonucleotide-mediated exon skipping for DMD treatment: Past insights, present challenges and future perspectives. J Biosci. 2023;48:38. PMID: 37846020.

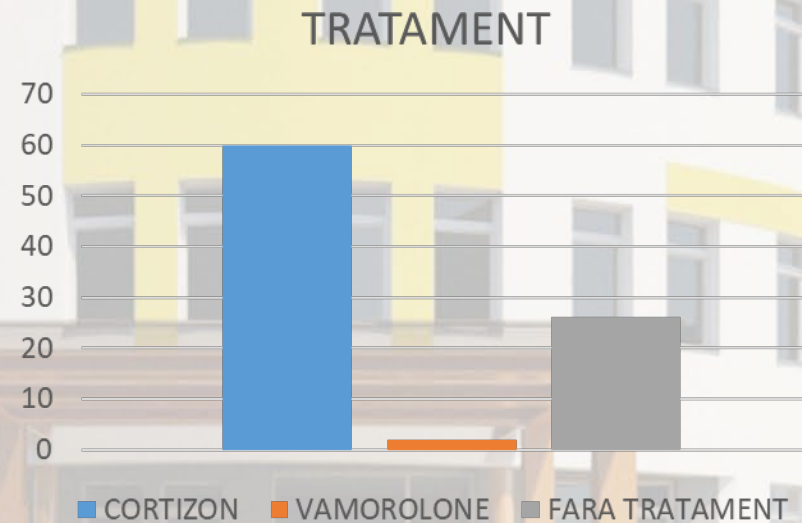
Congresul Național de Medicină Fizică, de Recuperare și Balneoclimatologie

Covasna, 23 – 25 Aprilie 2026

88 CHILDREN WITH DMD

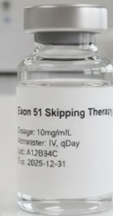


CORTISON TREATMENT 60
VAMOROLONE TREATMENT 2
WITHOUT TREATMENT 24



EXON SKIPPING

3 BOYS EXON SKIPPING 51
2 BOYS EXON SKIPPING 45

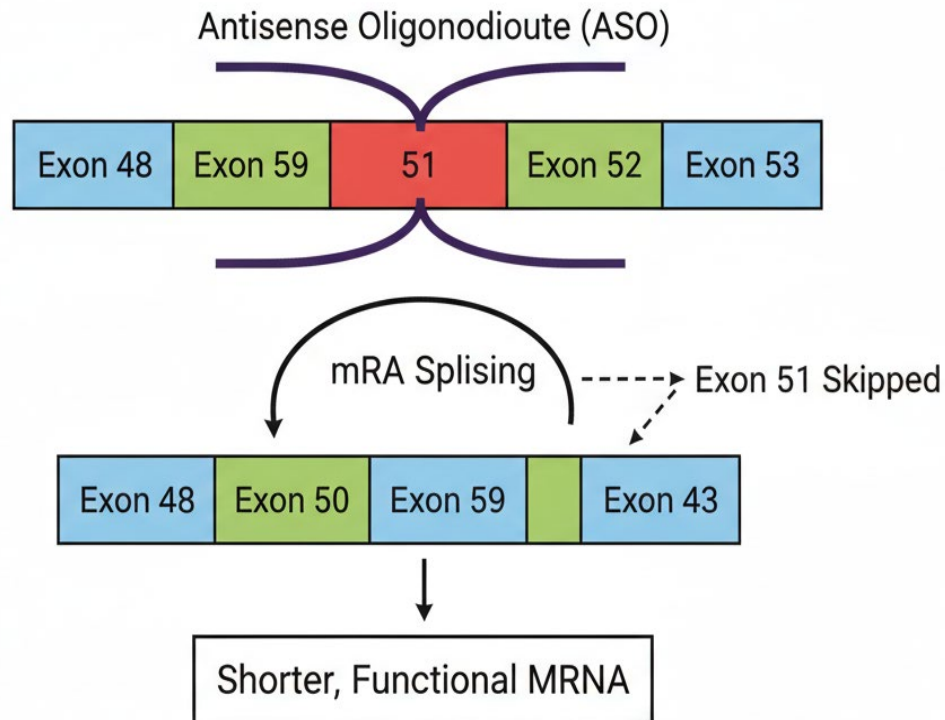


“1-stop” clinics - all work up on the same day

- Height, weight, blood pressure
- Urine sample collection for dipsticks
- Spirometry
- Physiotherapy assessment
- Blood tests,
- Echocardiogram,
- Spine Xray
- Dexa scan
- Doctors assessments
- ? CNS training
- Additional therapy sessions (SLT, dietetics family therapy)
- (? CRF visit)
- **How do you improve the patient / family experience?**

Activate Windows
Go to Settings to activate Windows.

ETEPLIRSEN – EXON SKIPPING 51



ETEPLIRSEN – EXON SKIPPING 51

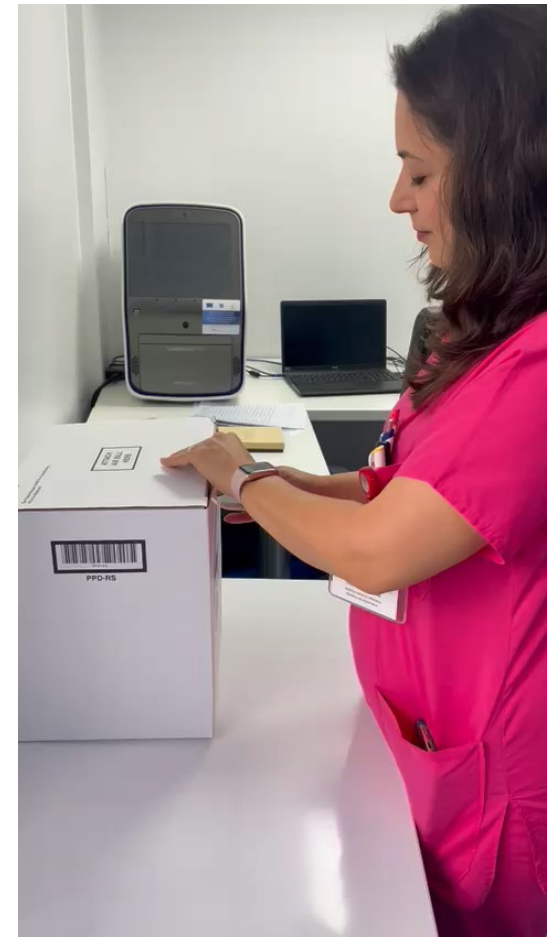
- **Route:** Intravenous infusion.
- **Frequency:** Usually once weekly, because antisense oligonucleotides are short-acting and do not persist long in muscle tissue.
- **Dose:** Weight-based dosing to maintain consistent drug exposure as the child grows.
- **Duration:** Long-term, ongoing therapy. Skipping stops if infusions stop.
- **Setting:** Infusion centers, hospitals, or trained home-infusion services depending on the region and patient stability.
- **Monitoring:** Regular checks of kidney function, infusion reactions, and clinical progression; occasional muscle biopsies to confirm dystrophin expression.
- **Onset:** Dystrophin restoration builds slowly over months; clinical effects may take longer.
- **Limitations:** Cannot fix DNA mutations—only modifies mRNA transiently, so continuous administration is required.

A Study to Compare Safety and Efficacy of a High Dose of **Eteplirsen EXONDYS 51** in Participants With **Duchenne Muscular Dystrophy** (MIS51ON)



The MIS51ON trial (high-dose eteplirsen) does not yet have published efficacy or final outcome data.

ETEPLIRSEN EXONDYS 51 STUDY

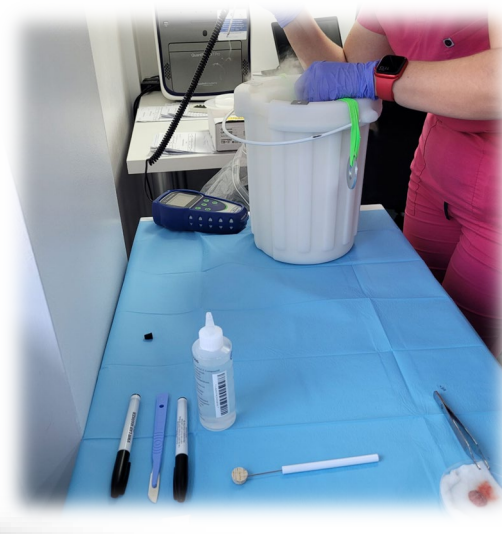
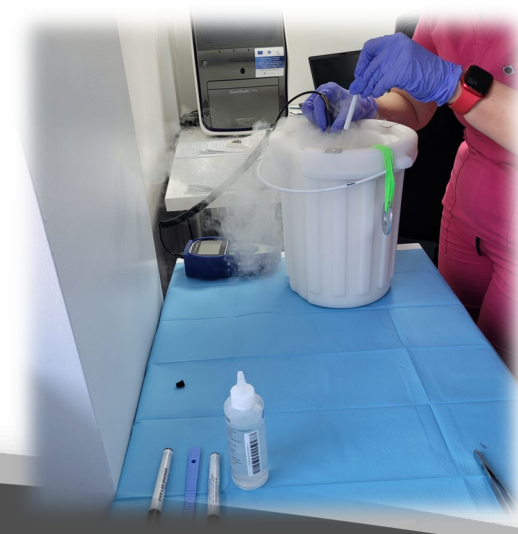
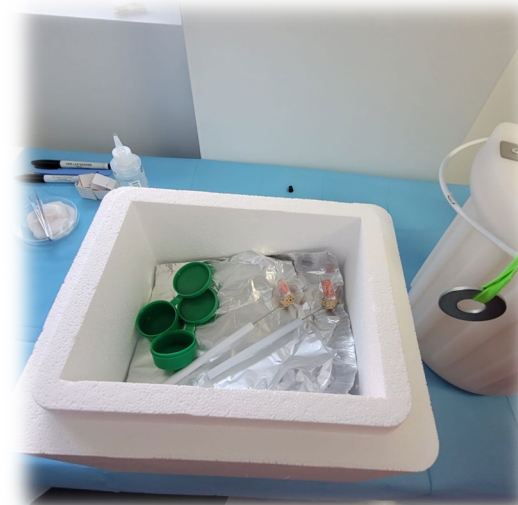
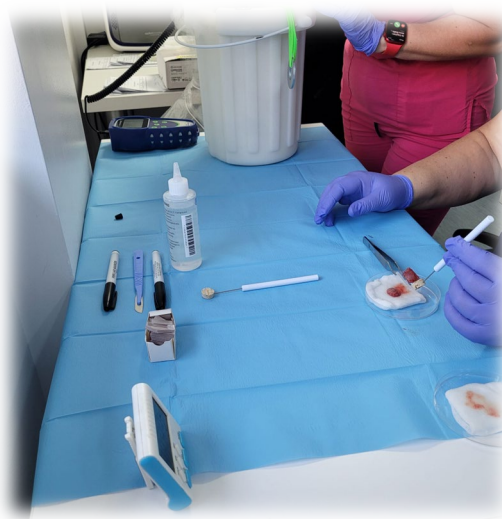
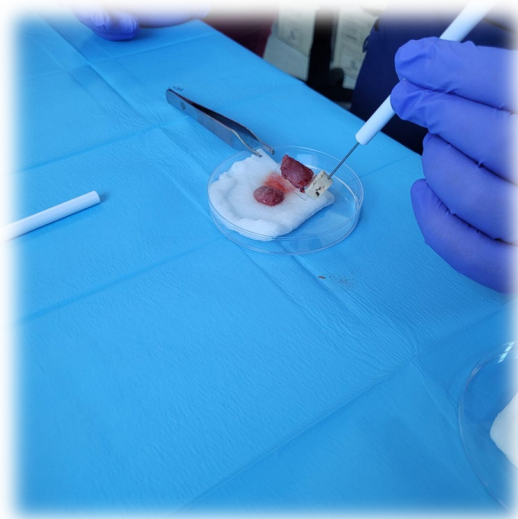
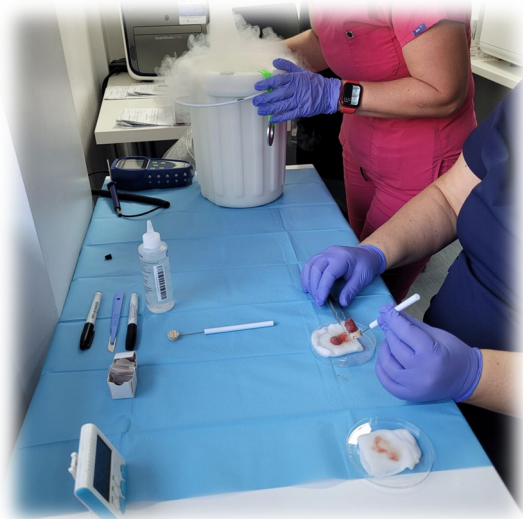


ETEPLIRSEN EXONDYS 51 STUDY

MUSCULAR BIOPSY



MUSCULAR BIOPSY (2)



CONCLUSION

DUCHENNE MUSCULAR DYSTROPHY IN THE NEW TREATMENT ERA

Targeted therapies like exon-skipping are shifting care toward molecular correction of the disease.

Next-generation AONs aim to increase functional dystrophin, with encouraging safety.

Earlier treatment is becoming critical, offering greater potential to preserve strength and delay progression.

The field is moving toward combination strategies (AONs, gene therapy, gene editing, muscle-enhancing agents).

Duchenne care is transitioning from primarily supportive management to personalized, mutation-specific therapy.