
# ‘Standardization and Interoperability in Rare Diseases, the Journey from Efficiency to Equity, the Duchenne Experience ”

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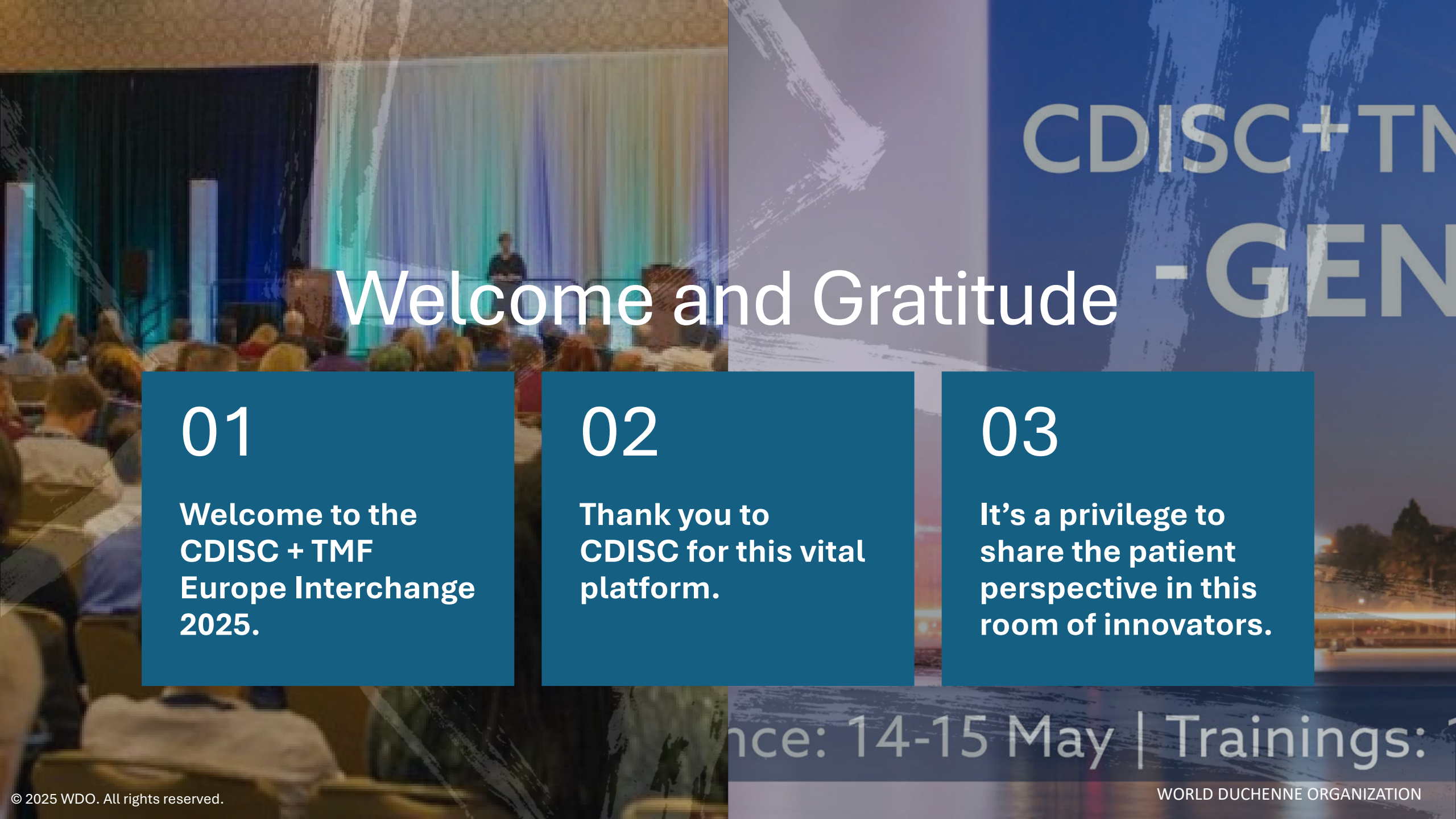


# Welcome and Gratitude

**01**

**Welcome to the CDISC + TMF Europe Interchange 2025.**

**02**

**Thank you to CDISC for this vital platform.**

**03**

**It’s a privilege to share the patient perspective in this room of innovators.**

# Setting the Scene

**CDISC provides a space where patient voices are not only heard, but drive change.**

**This event unites brilliant minds across research, regulation, and advocacy.**

**Our shared goal: Transform data into meaningful action for patients.**

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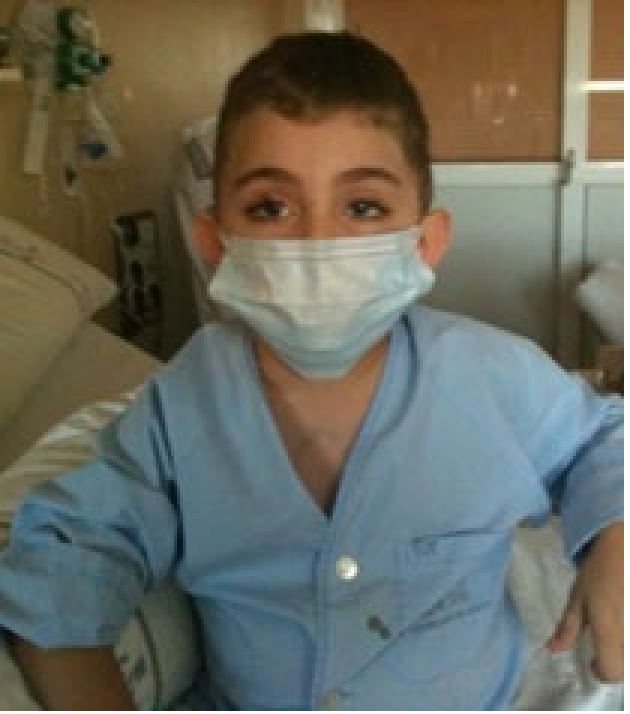
# Disclosures

**I have no real or apparent relevant financial relationships to disclose**

## DOI

www.ema.europa.eu/docs/en_GB/document_library/contacts/athanasioud_DI.pdf

# The Rare Disease Landscape

- 300 million people globally live with a rare disease.
- For most, there is no cure—and often, no treatment.
- **Data scarcity and fragmentation delay progress**.
- But these challenges also unite the community to act boldly.

# Rare Diseases High Unmet Need



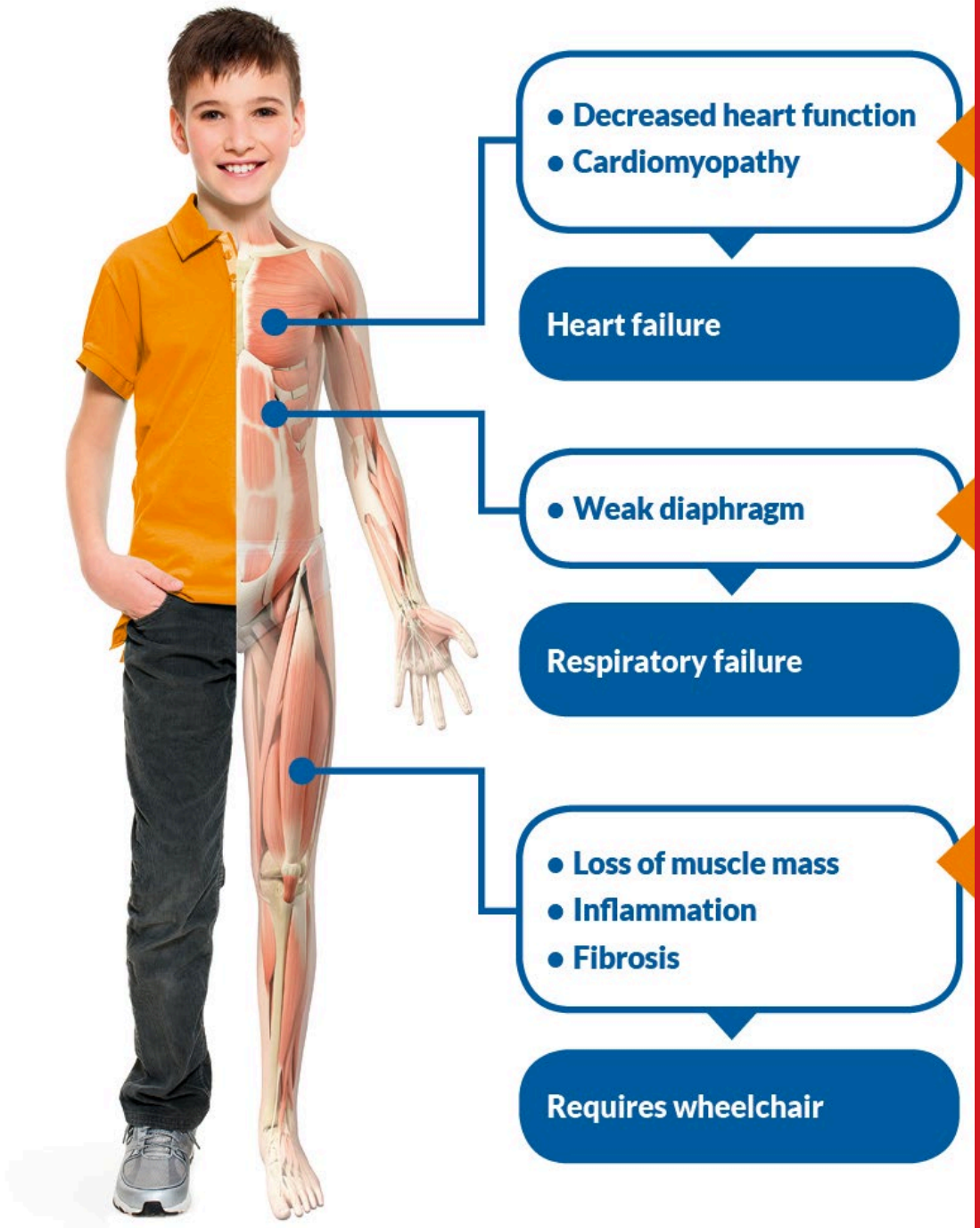
# The Rare Diseases

## 16th December 1999, European Parliament adopted Regulation (EC) No 141/2000 (Orphan Regulation)

As of May 2025, the European Commission has granted approximately **2,000 orphan medicinal product designations** since the implementation of the **EU Orphan Regulation in 2000**

It's important to note that not all designated orphan medicines reach the market. To date, over **260 orphan medicines** have been authorized for use in the EU

# The Duchenne Experience




- DMD gene: one of the longest
- MUTATIONS +++
- Evolving Natural History


**A Personal Reflection:**
Raising a boy with Duchenne

- Living with a boy with DMD is a daily journey.
- It brings uncertainty, but also resilience and perseverance
- Families step into roles as advocates, experts, and collaborators.

# The Community

- **1860s,** was first described by the French neurologist Guillaume Benjamin Amand Duchenne
- **1950s** the first NMD Patient Organizations were founded both in Europe and US
- **1980s** Duchenne and Becker Patient Organizations have been created all over the world
- **2000s they have evolved to a vibrant global network of advocacy groups that shaped a lot of the rare disease advocacy into the new millennium**

**At the dawn of 2020s** the Duchenne Community is not any more the grassroots collective of desperate parents that were trying to save their children.

Although it keeps its **strong drive, can-do mentality and passion** it more closely resembles a **well-oiled advocacy machine** that finances and shapes medical innovation and ATMs, co designs and influences Regulation and Policy in a global level.

Credit : Chris Barbalis@cbarbalis

# 47 MEMBER PATIENT ORGANIZATIONS FROM 39 COUNTRIES

# 47 MEMBER PATIENT ORGANIZATIONS IN ALL CONTINENTS

# WORLD DUCHENNE ORGANIZATION

UPPMD



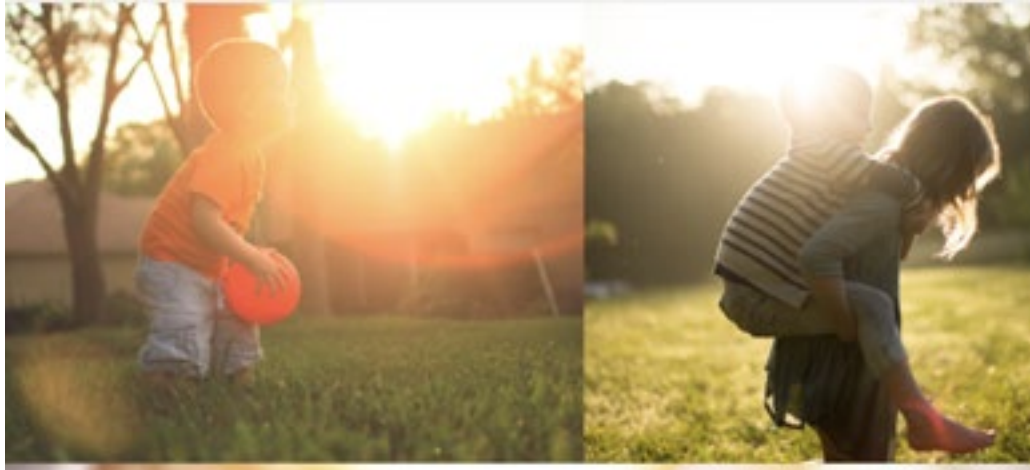
**Founded in 2005 with a strong focus on:**

- Harmonizing Research Funding & Knowledge
- Advocacy
- Regulatory work
- Policy

**WDO is Global network of member patient organizations**

- WDO is member and contributed to board positions in EURORDIS and EPF but also part of RDI.
- WDO's Board Members are involved in FDAs and EMA's EMA Committees, WPs and activities
- Eligible member of the European Medicines Agency

# DUCHENNE DATA FOUNDATION



**Founded in 2015 with a strong focus on:**

Patient-led foundation focusing on data collection and harmonization to facilitate and optimize DMD/BMD research & development.

DDF provides the framework and structure enabling research, knowledge exchange and education in the patient community and with other stakeholders.

- **Data**
- **Research**
- **Training**

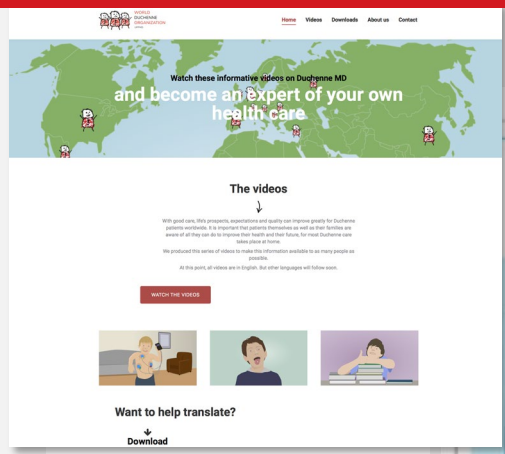
# The Community

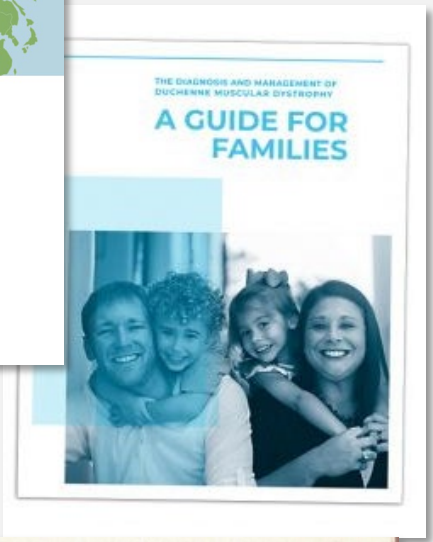
In Research :

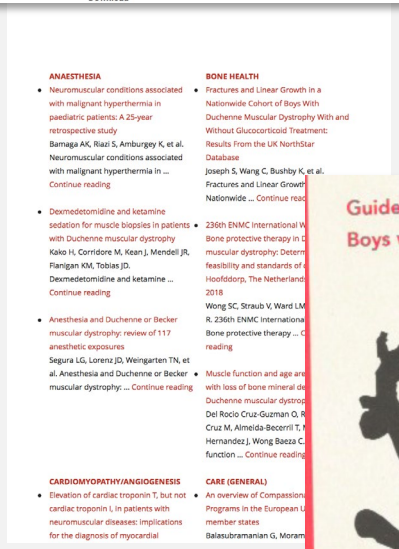
- Multi Millions € invested in Research and Care
- CT increase from 10 to 150+ clinical trails in the last decade
- Patient input to almost all the CTs in DMD via CAB
- Involvement in almost all DMD CTs in EMA, FDA, MHRA etc

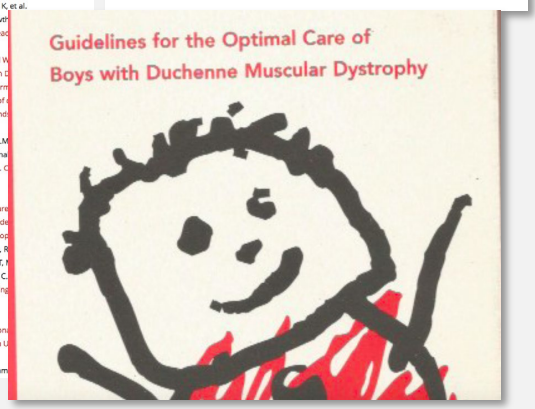
In Care and Education :

- International SoC development
- Accredited Duchenne Care Center Globally
- Increase in life expectancy by 7 years every decade
- Global Care Conference (+700 Part from 72 Countries)
- Health literacy, almost 100+ patient experts trained per year
- Education, materials reach out to more than 10,000 families




Credit : Chris Barbalis@cbarbalis

# The Community

The DMD community is supporting from basic Research to Care and Medicines Development :

- Community Advisory Board, Duchenne Patient Academy
- **In Silico Development A.I. and Machine learning**
- **Regulatory Guidelines for DMD Developers**
- **Clinical Trials Simulation Tools development**
- **Duchenne Platform Trials development**
- **Preclinical research support & Animal models**
- **Virus Development & ATM research like Gene and Cell Therapies,**
- **Gene editing and Exon skipping technologies**
- **Biotech and Spin-off seed funding**
- **Critical Path Institute (C-Path) and (CDISC) announce the open availability of a Duchenne Muscular Dystrophy Therapeutic Area User Guide (TAUG-DMD v1.0) (Pub.2017)**
  https://www.cdisc.org/standards/therapeutic-areas/duchenne-muscular-dystrophy
- …….and many other

Credit : Chris Barbalis@cbarbalis

VISION-DMD

BIND
Brain Involvement
in Dystrophinopathies

Share4Rare

EURO-NMD
Registry Hub
European Reference Network
for neuromuscular diseases

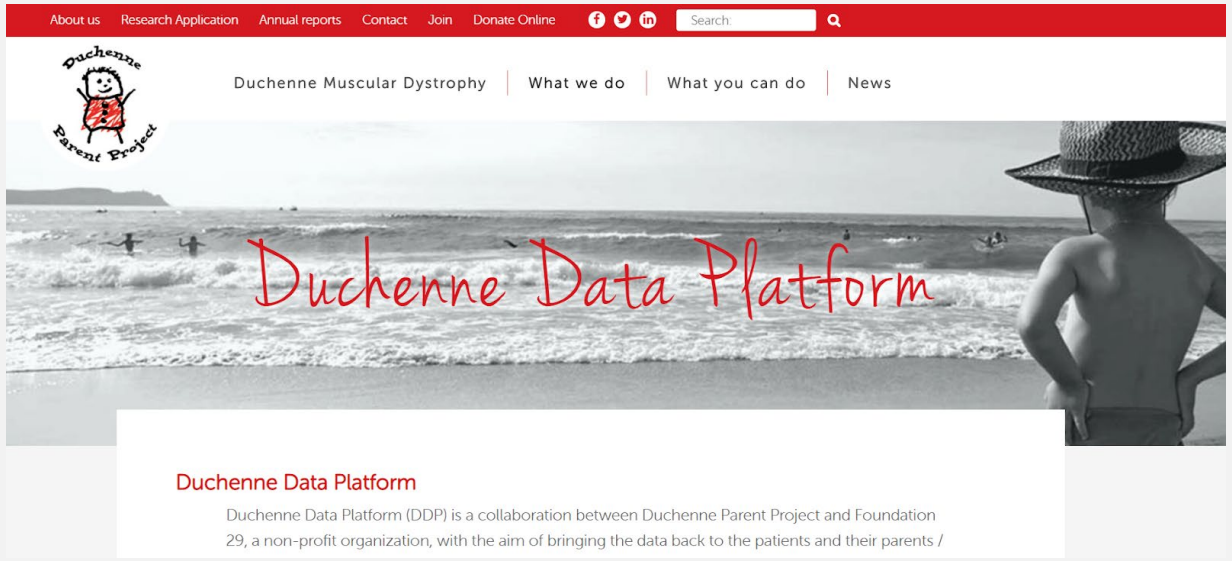
TRIALS
@HOME

DUCHENNE
COMMUNITY ADVISORY BOARD

DUCHENNE
DATA REPOSITORY

DUCHENNE
PATIENT ACADEMY

DUCHENNE MAP



# EURO-NMD Registry Hub

European Reference Network for neuromuscular diseases

## Key Facts

EURO-NMD Registry Hub
Start date: 01-05-2020
End date: 01-05-2023
www.registry.ern-euro-nmd.eu
info@registry.ern-euro-nmd.eu

## Objective

Building a registry hub for all neuromuscular diseases, including undiagnosed patients, and connect with existing ones.

## Partners

- Assistance Publique Hopitaux de Paris
- Universitaetsklinikum Freiburg
- Stichting Katholieke Universiteit
- World Duchenne Organization
- Duchenne Data Foundation
- Association Institut de Myologie
- Association Francaise Contre Les Myopathies

## Patient centered and interoperable registry hub for Rare Neuromuscular Diseases

EURO-NMD, ERN for Rare Neuromuscular Diseases, spans 14 European countries, with 61 reference centres that oversee more than 100,000 patients. Core objectives are the implementation of clinical practice guidelines and the definition and monitoring of core indicators of guideline conforming management, treatment quality and patient health outcomes.

Patient registries are key instruments for the ERN to be able to deliver its objectives. A recognised challenge for rare diseases is the heterogeneity of legacy data sets and the multiplicity of existing registries. EURO-NMD health care providers and patient organizations are currently active in more than 120, mostly disease specific and patient run registries. While the existing registries are collecting important information, none of them is used by all EURO-NMD centres and there is no unified NMD or NMD Disease Specific Registry in EU.

The general objective is to build a registry hub for all neuromuscular diseases, including undiagnosed patients, and connect with the existing ones. The EURO-NMD Registry Hub will use internationally agreed, state of the art concepts such as being built with a system that will collect standardized common data elements, defined by the Joint Research Center (JRC).

It will be registered in the JRC's meta-registry platform ERDRI, will allow for the generation of a Privacy Preservation Record Link (PPLR) through the EUPID system, it will use internationally accepted ontologies (HPO) and ORPHA codes for codification of the diseases. Development of a registry hub that will allow linking and extraction of data from different sources. Thus, the data collected through the registry hub will be Findable, Accessible, Interoperable, and Reusable (FAIR).

The registry hub will offer the unique opportunity to the fragmented NMD communities and their HCPs, Centers, Patients and Patient Organizations to be able to communicate between them and exchange knowledge, experience and news.

This project has received funding from the 3rd EU Health Programme under grant agreement No 947598

# EURO-NMD REGISTRY HUB

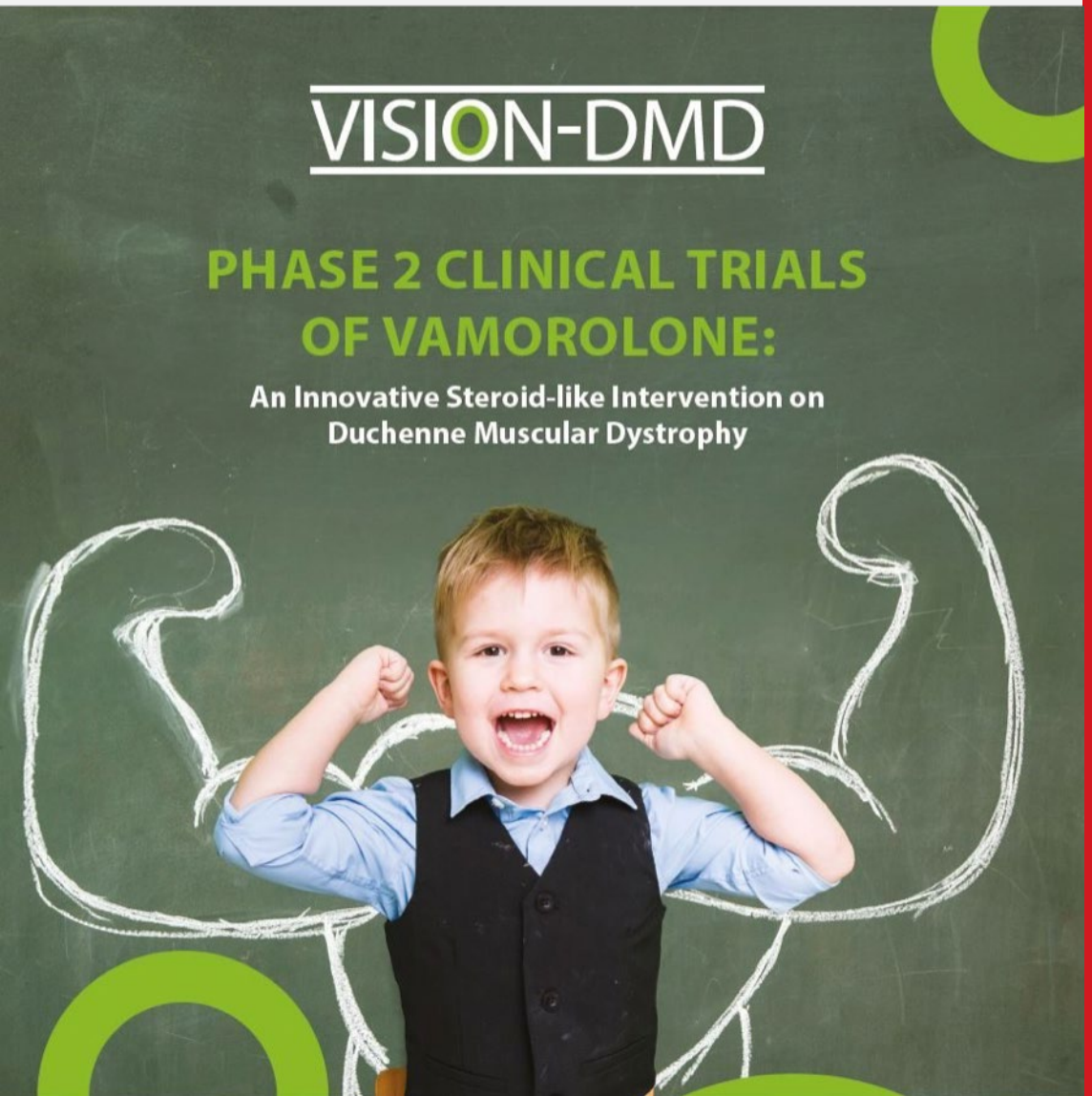
## RARE NMD DATA HUB

- ➔ Improving quality and equity of healthcare
- ➔ Enabling the exchange of knowledge through teaching and training
- ➔ Facilitating translational research
- ➔ Focus on strengthening the collaboration amongst the 61 HCPs so that, when new members are proposed and endorsed, their integration is swift with minimum impact on the ERN's activities.

https://registry.ern-euro-nmd.eu/

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement number 947598.

VISION-DMD




# Vision-DMD

## CLINICAL TRIAL INVESTIGATING VAMOLORONE

Aims to develop a safe, effective and affordable therapy for Duchenne Muscular Dystrophy (DMD) using venture philantrophy. This is done by investigating the safety and efficacy of Vamolorone (VBP15) in ambulant DMD boys and is currently undertaking Phase 2b clinical trial.

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement number 667078.

# BETWEEN HYPE AND HOPE

- Still the pathway is marked with successes and failures.
- The boys live longer, have a better a life, but still lose the fight with Duchenne.
- Balancing between Hype and Hope the community still fights to keep the boys alive while keeping DMD and the Rare Diseases in the centre of R&D, Regulatory and Policy discussions.

# The Challenges in DMD and other RDs remain

- Many unknows in patient management and care, transition etc
- Many unknows in disease trajectory and evolving SoC
- Clinical Trials Design (ex. Master Protocol, End Points, Biomarkers etc)
- Data Issues and in Silo attitudes (# of Registries)
- Collection of Reliable Data and Long Term Follow Up (DMD Post Marketing Registry)
- Pipeline issues , with many failing products but also Feasibility issues - # of drugs

# From Challenges to Advocacy

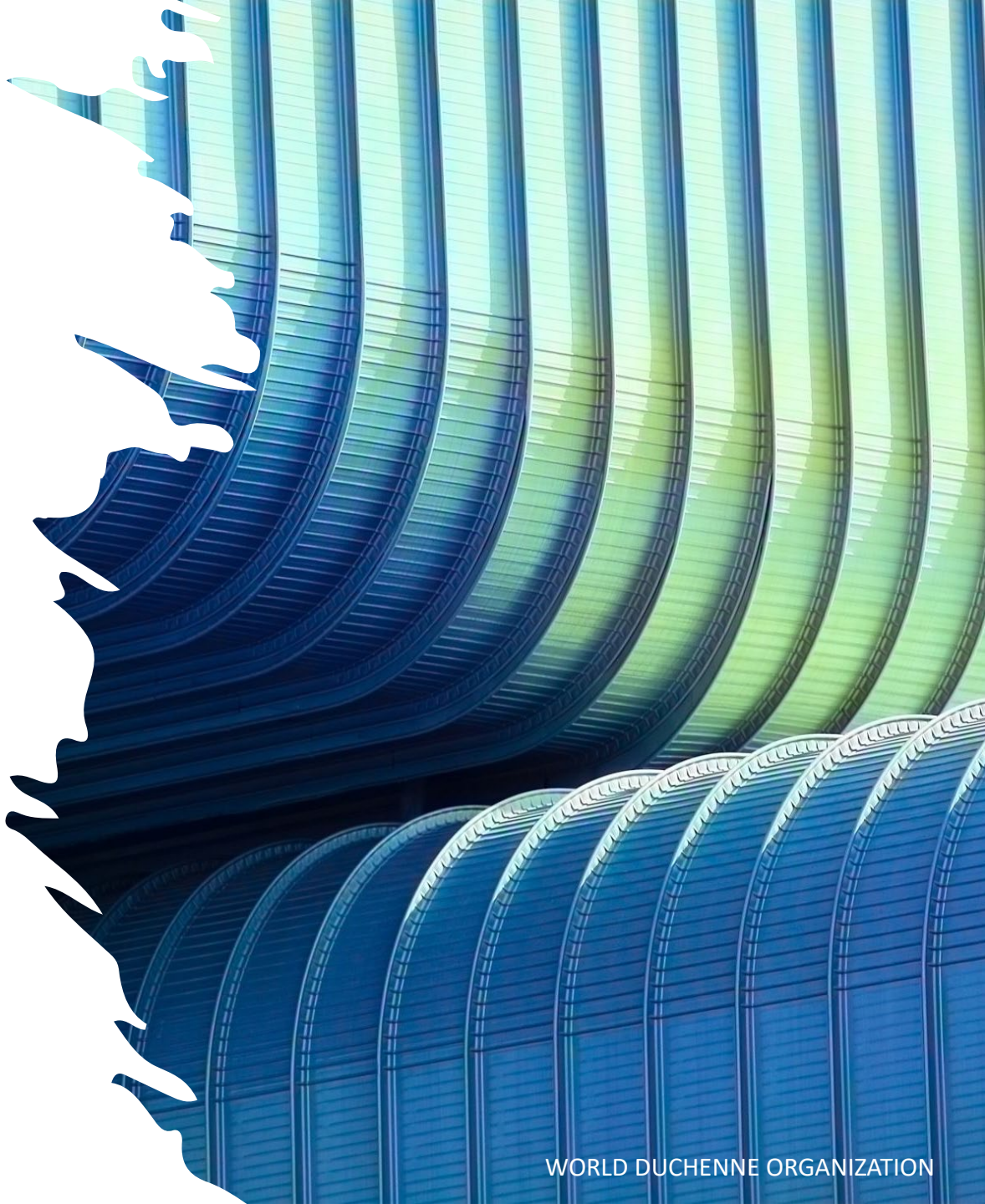
**Rare diseases demand more than clinical innovation—they demand systemic change.**

**Advocacy leads to:**

**- Better trial designs**

**- More inclusive endpoints**

**- More ethical and effective research**



# A Shift in Perspective

**The role of the patient has evolved.**

**We are not passive participants; we are partners in progress.**

**Insights from lived experience must shape:**

**- Study design**

**- Data interpretation**

**- Regulatory dialogue**

# The Power of Standardization

**Standards ensure consistency across:**

**-Trials**

**- Institutions**

**- Nations**

They enable global collaboration and accelerate learning.

It ensures continuity of care across geographies and lifespans.

# Interoperability Matters

**Interoperability = communication between systems, institutions, and disciplines.**

**For rare diseases:**

It’s the difference between isolated data and actionable knowledge.

It ensures continuity of care across geographies and lifespans.

# From Efficiency to Equity

**Efficiency helps us move faster.**

**Equity ensures we move together.**

**Equity means:**

Representing all populations

Designing accessible trials

Addressing real-world barriers

# Equity in Data

**We must ask: Whose data is missing?**

**Ensure inclusion across:**

- Gender
- Age
- Ethnicity
- Socioeconomic status

# Multi-Stakeholder Collaboration

Our progress is rooted in partnerships:

- Regulators
- Researchers
- Industry
- Patients

Co-creation leads to sustainability, relevance, credibility and builds **TRUST**

# Real-World Evidence and Data Reuse

Standardized real-world data unlocks:

- Long-term outcomes
- Post-marketing surveillance
- New therapeutic insights

Especially important in rare diseases with limited trial populations.

# CDISC's Role in Rare Diseases

**CDISC needs to set the gold standard for structured, reusable, reliable data.**

**It fosters and should be translated to:**

Faster approvals

Better data quality

Trust between stakeholders

AP Photo/Martin Meissner

# Ask Provocative Questions to Change the System

**Why is it that in 2025, patient registries are still treated as secondary sources of evidence?**

**We claim the TMF ensures quality — but how much of it is about protecting sponsors instead of patients?**

**Are we using CDISC to truly understand disease — or just to speak the language of regulators?**

**Why do we keep optimizing a system designed in the 20th century for problems we now face in the 21st?**

**Who really owns the data? And who owns the consequences of how it's used or ignored?**

# Call to Action

**Let’s bridge research and real-life experiences.**

**We can:**

- **Amplify patient perspectives**
- **Advance impactful, compassionate innovation**
- **Build a data ecosystem that serves everyone**

# The Future We Imagine

**is a world where:**

- Every patient’s data counts
- Standards reflect lived experiences for the benefit of the patients
- Data drives policy, practice, and personal impact

**If we truly believe in innovation, let’s stop polishing the status quo. Let’s disrupt it.**

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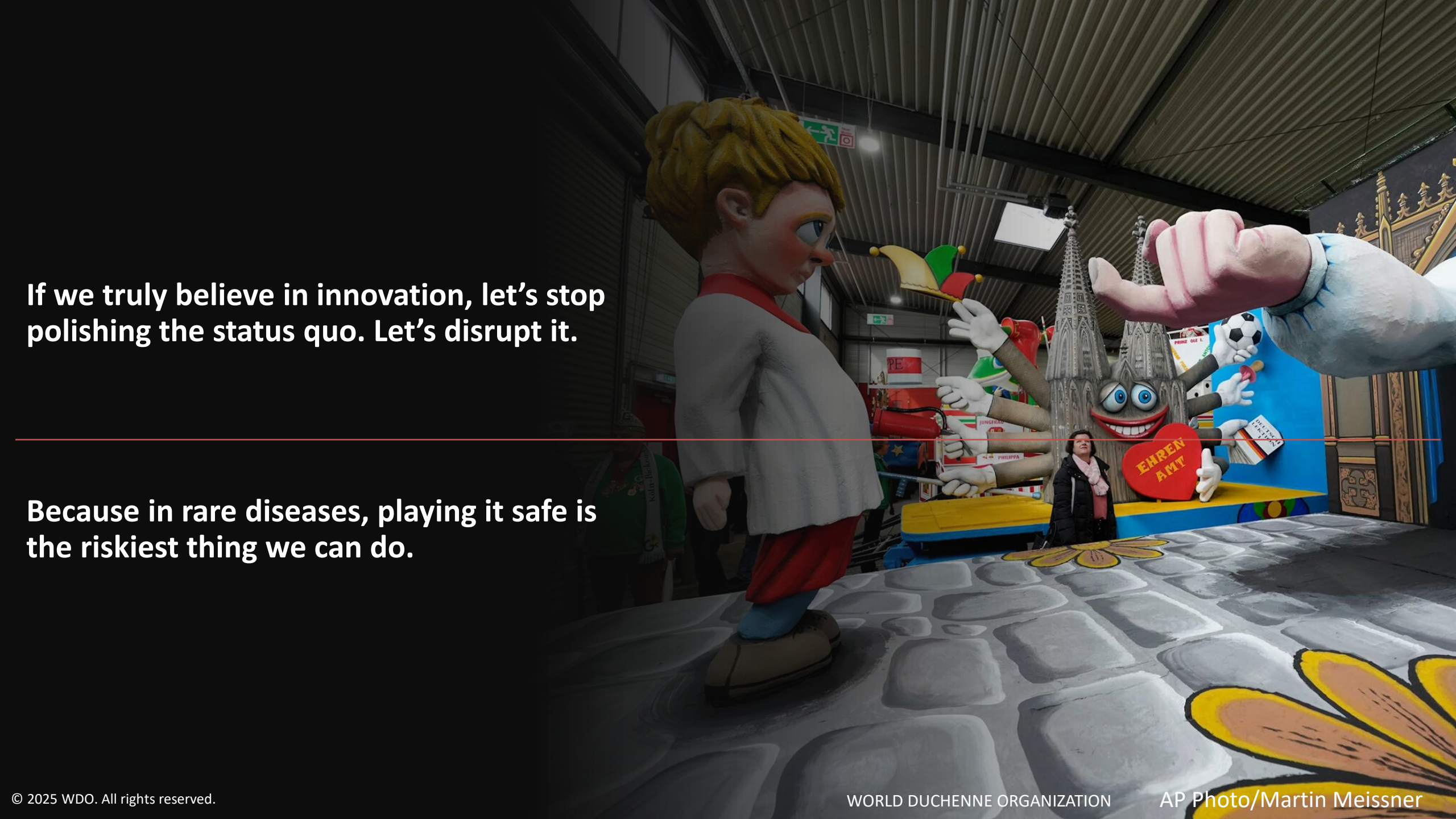
**Because in rare diseases, playing it safe is the riskiest thing we can do.**








AP Photo/Martin Meissner


# THANK YOU!

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