From Insight to Evidence: Strengthening the Role of Patient Data in Regulatory Decisions.

HMA EMA Annual Data Forum,
December 9th, 2025

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Patient expert at EMA
SMA Europe
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Rare, neuromuscular, life-threatening disease.

HISTORICAL DATA ON SMA

Natural history: Progressive disease with spectrum clinical manifestations.

Biomarker as severity predictor.

Data collection & measurement tools designed to register motor decline.

SoC linked to inevitable decline: benefit-burden balance

REGISTRIES IN SMA

BEFORE DMTs

- Genetic registries: Incidence information.
- Natural history: clinical purposes: underrepresentation of groups

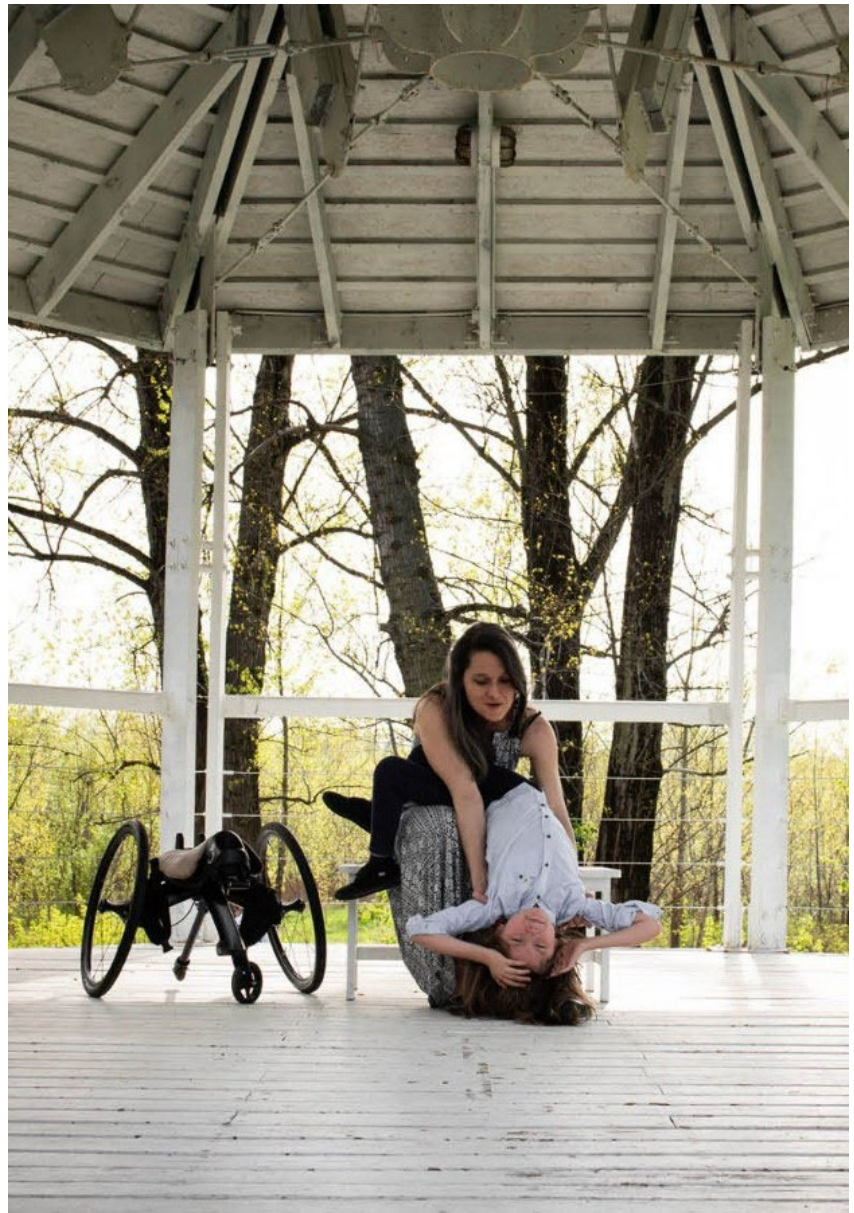
CLINICAL TRIAL ERA

- Registries to facilitate recruitment

CT DESIGN FOR FIRST SMA DMTs

- Urgent unmet need: asap.
- Natural history data for clinical purposes
- Measurement tools to register inevitable decline
- Under representation of patient subgroups.
- Endpoints: hard endpoints & pre-existing validated measurement tools.
- Exploratory: validated QoL tools.

3 DMTs: ASO (2017), GT (2020), SM (2021).



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SOME EXAMPLES IN WHICH RWE HAS CONTRIBUTE TO REGULATORY DECISIONS

ODD: significant benefit for patients

- Subgroups not accessing to DMT: spinal fusion
- Impact of expected effects on everyday life: fatigue

Input in Protocol and Scientific Advices:

- Patient preferences, disease & protocol burden.

DMT MA Assessment Procedures:

Harbouring four SMN2 gene copies does not guarantee a milder phenotype: SMA Registry in Spain M.G. Cattinari[1], M. De Lemus1[,2,3], E. Tizzano[4]

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EUPESMA-2019 – European Patient Experience Survey in SMA to understand patient expectations towards therapeutic development in spinal muscular atrophy

Nicole Gusset[1,2], Yasemin Erbas[1,3,4], Caroline Stalens[5], Eva Stumpe[1,6], Lori Klouvi[5], Alexandre Mejat[5], Marie-Christine Ouillade[1,5], Mencía de Lemus[1,7]

SMA PATIENT'S EFFORTS IN RWE

- Promote creation of patient registries to generate RWE
- Develop Patient Relevant Outcomes
- Establish minimally significant gains in different patient subgroups.
- Link validated measurements to everyday life gains
- Express expectations on the DMTs: surveys: Value of stabilisation.

Orphanet J Rare Dis. 2024 Feb 19;19(1):76. doi: 10.1186/s13023-024-03071-7.

RegistrAME: the Spanish self-reported patient registry of spinal muscular atrophy

Maria Grazia Cattinari [1], Mencía de Lemus [2 3 4], Eduardo Tizzano [5]

Understanding the relationship between the 32-item motor function measure and daily activities from an individual with spinal muscular atrophy and their caregivers' perspective: a two-part study

Tina Duong [1], Jessica Braid [2], Hannah Staunton [3], Aurelie Barriere [4], Fani Petridis [5], Johannes Reithinger [5], Rosangel Cruz [6], Jill Jarecki [6], Mencia De Lemus [7 8], Nicole Gusset [7 9], Ria Broekgaarden [7 10], Sharan Randhawa [11], Jessica Flynn [11], Rob Arbuckle [11], Sonia Reif [12], Lida Yang [12], Angela De Martini [12], Carole Vuillerot [4 13]

Neurol Ther. 2021 Jun;10(1):361-373. doi: 10.1007/s40120-020-00229-w. Epub 2021 Jan 9.

Design of a Non-Interventional Study to Validate a Set of Patient- and Caregiver-Oriented Measurements to Assess Health Outcomes in Spinal Muscular Atrophy (SMA-TOOL Study)

Marcos Madruga-Garrido [1], Juan F Vázquez-Costa [2 3 4 5], Julita Medina-Cantillo [6], María Brañas [7], María G Cattinari [8], Mencía de Lemus [8 9], Paola Díaz-Abós [7], Victoria Sánchez-Menéndez [7], Ángeles Terrancle [7], Pablo Rebollo [10], Jorge Maurino [11]

RWE on SMA

Post MARKETING AUTHORISATION ERA

- Security and efficacy
- New phenotypes
- Pat relevance

COMMON PROBLEMS IN PATIENT DATA USE

- FFP
- Trust: effective use
- Data today still not addressing the questions patients need answered:
  - Trajectories: which drug works better for my son?
  - Biomarkers: is he receiving enough stable SMN protein?

CAN WE DO BETTER?

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European Medicines Regulatory Network (EMRN) strategy to 2025

"By 2025 the use of Real-World Evidence will have been enabled and the value will have been established across the spectrum of regulatory use cases."

HAVE WE ACHIEVED THIS GOAL?

REGULATORY EFFORTS TOWARDS USE OF PATIENT DATA

- DARWIN
- Reflection papers
- Guides, reports
- Workshops
- Multistakeholder meetings
- Registry Qualification
- ...

DARWIN EU

Reflection paper on patient experience data

Optimizing Patient Registries for Regulatory Decision Making - Key Learnings From an HMA/EMA Multistakeholder Workshop

Kelly Plueschke[1,*], Carla Jonker[1,2], Hanna Kankanen[1], Thorsten Vetter[1], Bruno Sepodes[3], Lutz Naehrlich[4,5], Jan Hillert[6], Gracy Crane[7], Sabine Straus[2], Paolo Foggi[8], Simona Martin[9], Christina Kyriakopoulou[10], Peter Mol[2,11], Francesca Day[1], Kieran Breen[12], Neil Bennett[13], Mencia de Lemus Belmonte[14], Simon Bennett[15], Patrice Verpillat[1], Kit C. B. Roes[16], Ana Cochino[1], Franz Schaefer[17], Jesús María Hernández-Rivas[18], Patricia McGettigan[19] and Peter Arlett[1,20]

WHAT WOULD I LIKE TO SEE IN THE COMING YEARS?

- Early & sustained engagement with registry holders.
- Maintain/establish ambitious goals on the data use.
- Use technologies to reduce burden.
- Take bold steps towards the use of patient input.

- THANK YOU!

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