Multistakeholder consensus for a pediatric core outcome set for arthrogryposis

Naomi Zukerman (M.Sc, OT, PhD student), V. Darsaklis, C. Araujo, S. Cachecho, C. Costa, K. Donlevie, A. Fąfara, U. Kasar, C. Krakie, F. Lacombe, V. Pacey, J. Megan Sions, Y. Zhang & N. Dahan-Oliel

Presentation supported by grants from Arthrogryposis Multiplex Congenita Support, Inc., McGill University Health Centre Research Institute and the Canadian Musculoskeletal Rehab Research Network

Overview & Primary Objective

- **Condition:** Arthrogryposis multiplex congenita (AMC) is a group of congenital conditions characterized by non-progressive joint contractures affecting 2+ body areas, with variable causes and functional impacts (Dahan-Oliel, 2019).

- **Current Discrepancies:**
  - Advocacy for holistic and family-centred care.
  - Gaps in the literature and a lack of guidelines, impacting services.
  - Need for outcome measure consensus in pediatric rehabilitation for AMC.

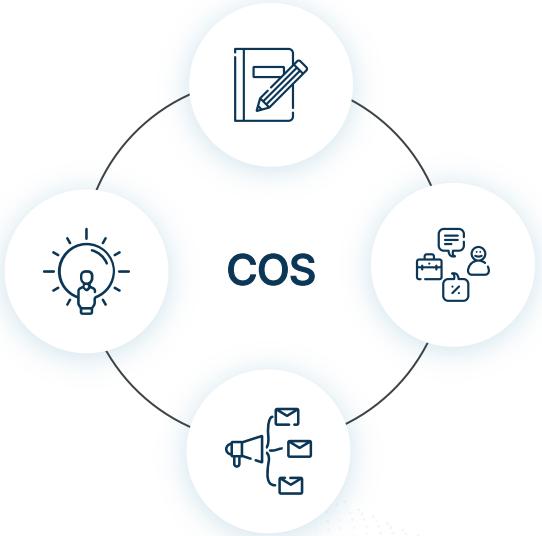
- **Primary Objective:** This study aims to develop a core outcome set for a group of rare musculoskeletal diseases (AMC), fostering standardized and inclusive care practices.

Core Outcome Sets (COS)

COS are lists of **consensus-determined outcomes** to be measured and reported in clinical research studies within a disease area (Vanderhout, 2021)

Choice of Valid Measures

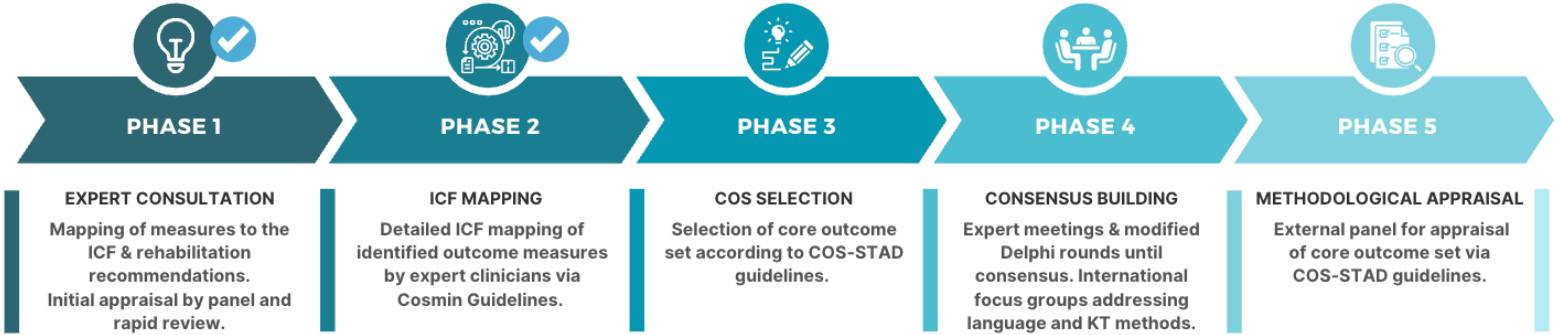
Optimized Quality of Care



Streamlining of Services

Improved Collaboration

Multi Phasic Mixed Methods Study



LEGEND

ICF = International Classification of Functioning, Disability and Health
Cosmin Guidelines = COnsensus-based Standards for the selection of health Measurement INstruments Guidelines
COS-STAD Guidelines = Core Outcome Set-STAndards for Development
KT = Knowledge translation

Preliminary Steps

International Scope
Canada, USA, Poland, Australia and India

Varied Stakeholders
Creation of panel representing clinicians (OT, PT, MD, MSW, Technical Aids) **and** individuals with lived experience

Foundational Knowledge
Based on 5 multidisciplinary research activities (2017-2024)

Pooling of Outcomes
Candidate outcomes were compiled and 31 outcome measures were identified

Phase 1a: Expert Consultation

1) Extraction of ICF domains in 31 measures
2) Analysis according to 16 consensus-based rehabilitation recommendations (CBRR) for children with AMC (Dahan-Oliel, 2025)
3) **5 measures matched at least 3 ICF domains as well as 8/16 or more (over 50%) of the CBRR met inclusion criteria.**
4) Panel provided feedback on the 5 measures and suggested additional measures

Preliminary Outcome Measures	Comments
Pediatric Outcomes Data Collection Instrument (PODCI)	• Validated for AMC population • Considers fluctuating pain/ function, while accounting for assistive devices
Ages & Stages Questionnaires, Third Edition (ASQ-3)	• Comprehensive screening for infants/ young children • Integrates activities for parents
Patient-Reported Outcomes Measurement Information System (PROMIS)	• Covers a wide range of outcomes • Includes specific pain and fatigue short forms
Euro-QoL-5D (EQ-5D)	• Provides a simple and direct overview of health status • Available in 150+ languages
Assessment of Life Habits (LIFE-H)	• Spans several age groups • Touches on accomplishment, assistance and satisfaction

Phase 1b: Rapid Review

1

Search

Medline, Embase, CINAHL, Cochrane, Wiley and Global Index Medicus

2

Screening 1

Title/ abstract (NZ, YZ)

3

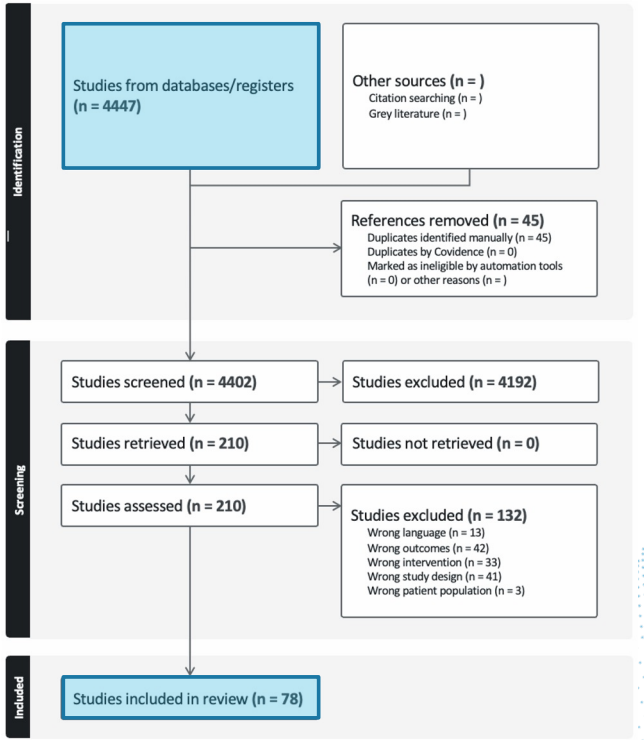
Screening 2

Full text review (NZ, YZ)

4

Extraction

Data was extracted from 78 articles (NZ, YZ)



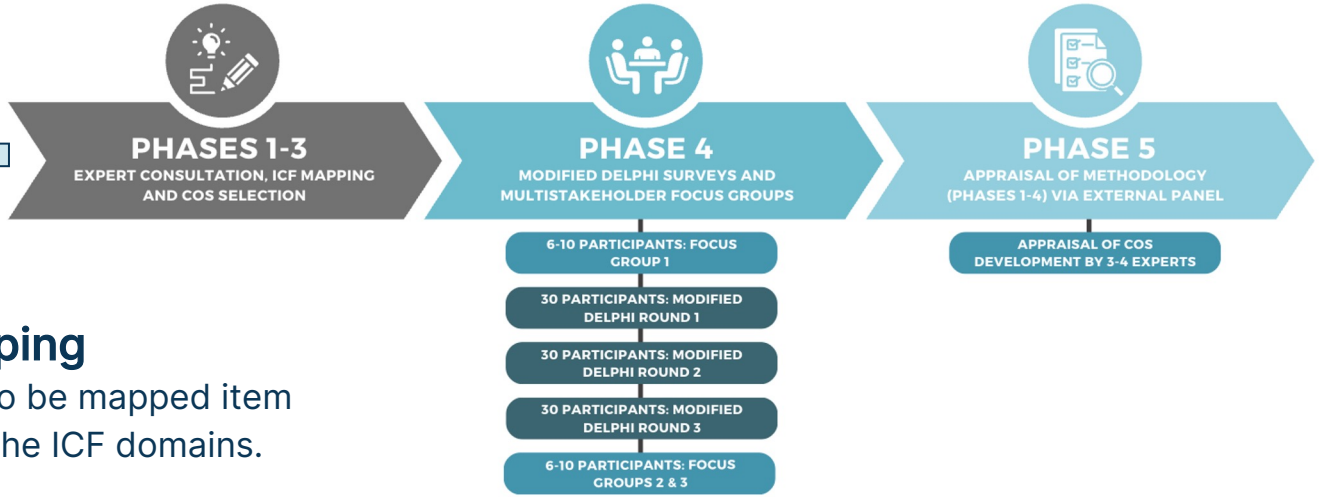
Rapid Review Results & “Pruning”

(Howie et al., 2021)

- Data was extracted from 78 studies
- Physical, psychosocial and functional outcomes were discussed across studies

Preliminary Measures Identified	Additional Measures From Panel (1a)	Measures Identified Post Rapid Review (1b)
PODCI, ASQ-3, **PROMIS**, EQ-5D and LIFE-H	**PEDI-CAT; Screening Tool for Pain;** Patient Specific Functional Scale; FMS	**PODCI** n= 18; **PROMIS** n=8; **PEDI-CAT** n=5; SF-36 n=4; WeeFIM n=4; **Numerical Pain Rating Scale** n=4

Next Phases



ICF Mapping

Measures to be mapped item by item to the ICF domains.

Key Takeaways and Implications

1

Individual Needs

Each rare disease is a unique entity, reflected by the inclusion of condition-specific outcome measures.

2

Patient-Centred

A COS for AMC is a natural progression to continue bridging gaps and offering patient-centered care.

3

OT Innovation

A COS for AMC is the first of its kind in pediatric rehabilitation, affirming the unique perspective of OTs in cutting-edge research.

Questions?
naomi.zukerman@mail.mcgill.ca

References

Dahan-Oliel, N., Cachecho, S., Barnes, D., Bedard, T., Davison, A. M., Dieterich, K., Donohoe, M., Fąfara, A., Hamdy, R., Hjartarson, H. T., Hoffman, N. S., Kimber, E., Komolkin, I., Lester, R., Pontén, E., Van Bosse, H. J. P., & Hall, J. G. (2019). International multidisciplinary collaboration toward an annotated definition of arthrogryposis multiplex congenita. *American Journal of Medical Genetics Part C Seminars in Medical Genetics, 181*(3), 288–299. https://doi.org/10.1002/ajmg.c.31721

Dahan-Oliel, N., Araujo, C., Fąfara, A., Lacombe, F., Samargian, A., Costa, C., Donohoe, M., Flanagan, A., Kowalczyk, B., Krakie, C., Wagner, L., Navalón, C., Pacey, V., Steen, U., Walker, M., Wong, T., Bussières, A. Consensus-based recommendations for the rehabilitation of children with arthrogryposis multiplex congenita: an integrated knowledge translation approach. *Orphanet Journal Rare Diseases 2025;20*(1):168. https://doi.org/10.1186/s13023-025-03671-x

Howie, A. H., Tingley, K., Inbar-Feigenberg, M., Mitchell, J. J., Butcher, N. J., Offringa, M., Smith, M., Angel, K., Gentle, J., Wyatt, A., Campeau, P. M., Chan, A., Chakraborty, P., Turk, F. E., Mamak, E., Mhanni, A., Skidmore, B., Sparkes, R., Stockler, S., & Potter, B. K. (2021). Establishing a core outcome set for mucopolysaccharidoses (MPS) in children: study protocol for a rapid literature review, candidate outcomes survey, and Delphi surveys. *Trials, 22*(1). https://doi.org/10.1186/s13063-021-05791-8

Vanderhout, S., Smith, M., Pallone, N., Tingley, K., Pugliese, M., Chakraborty, P., Stockler, S., Offringa, M., Butcher, N., Nicholls, S. G., & Potter, B. K. Patient and family engagement in the development of core outcome sets for two rare chronic diseases in children. *Research Involvement and Engagement 2021;7*(1) https://doi.org/10.1186/s40900-021-00304-y