Outline

- What is AMC?
- What do we know about rehab in AMC?
- What is patient-oriented research (POR)?
- Patient-oriented research in AMC
- Lessons learned & the future
What is AMC?

Arthrogryposis multiplex congenita (AMC):

- 1 in 3000 live births
- Joint contractures ≥2 body areas
- Impairments in other systems
What is AMC?
Rehabilitation in AMC

• Lower levels in self-care domain using the PEDI during preschool and school-age (Joubert & Franzsen, 2016)

• Lower scores in all 6 domains using the PODCI (Amor et al, 2011)


Rehabilitation in AMC

- Early, comprehensive and multidisciplinary (Binkiewicz-Glinska, 2013)
- Achieve and maintain maximal range of motion and strength
  - Promote self-care, mobility and participation

Rehabilitation in AMC

- Manipulation, casting, taping, orthosis
  - Improve ROM and maintain gains
- Promote floor mobility and weight bearing
- Exoskeleton
  - Use of upper body for fine motor skills (Babik et al, 2016)

Rehabilitation in AMC

What we know:
- Complex cases
- Research is single-site and fragmented
- Collaborative research is beneficial

What we don’t know:
- Evidence-based interventions
“interactive, bi-directional exchange between stakeholders and researchers” in order to develop research that is most important to patients and to improve the quality and relevance of research. (Curtis et al., 2012) http://www.cihr-irsc.gc.ca/e/documents/spor_framework-en.pdf
Patient-oriented research

[Diagram showing levels of public involvement and influence, progressing from inform or educate to partner]

- Support
- Mutual Respect
- Co-Build
- Inclusiveness

Patient-oriented research in AMC

Setting research priorities

Planning meeting, May 22nd 2016
Patient-oriented research in AMC

Setting research priorities

Patient-oriented research in AMC

Setting research priorities

– Genetics inheritance: early diagnosis & screening
– Identification of outcome measures to evaluate interventions
– Effectiveness of treatment
– Physiotherapy/exercise program
– Longitudinal studies to document adult outcomes

Patient engagement framework
# Patient-oriented research in AMC

## 1 Registry

<table>
<thead>
<tr>
<th><strong>CHILD</strong></th>
<th><strong>MOTHER</strong></th>
<th><strong>FATHER</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Demographics</td>
<td>Sociocultural background</td>
<td>Sociocultural background</td>
</tr>
<tr>
<td>Race and ethnicity</td>
<td>Lifestyle habits</td>
<td>Lifestyle habits</td>
</tr>
<tr>
<td>Medical history</td>
<td>Medical history</td>
<td>Medical history</td>
</tr>
<tr>
<td>- Measurements at birth</td>
<td>- Labour and Delivery</td>
<td>- Labour and Delivery</td>
</tr>
<tr>
<td>- Apgar scores</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Complications at birth</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Interventions at birth</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Fractures at birth</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>Clinical features</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Facial abnormalities</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Neurological abnormalities</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Internal abnormalities</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Skin or soft tissue abnormalities</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Feeding</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Metabolic disease</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Contractures at birth</td>
<td>-</td>
<td></td>
</tr>
<tr>
<td>- Interventions to correct bony deformities and/or joint contractures</td>
<td>-</td>
<td></td>
</tr>
</tbody>
</table>

**Open access**

**BMJ Open** Development of a research platform for children with arthrogryposis multiplex congenita: study protocol for a pilot registry

Noémi Dahan-Oliel, Tanya Bedard, Vasiliki Betty Darsaklis, Judith Goelin Hall, Harold J P van Bosse, Reggie C Hamdy

Patient-oriented research in AMC

2. Rehabilitation Guidelines

Priorities:
1. Daily activities
2. Pain management
3. Mobility
4. Muscle and joint function
5. Participation in the community

Elfassy et al. Rehabilitation needs of youth with arthrogryposis multiplex congenita: Perspectives from key stakeholders. (2019) In press, Disability & Rehabilitation
# Patient-oriented research in AMC

## 2. Rehabilitation Guidelines

<table>
<thead>
<tr>
<th>Phase 1</th>
<th>Phase 2</th>
<th>Phase 3</th>
<th>Phase 4</th>
<th>Phase 5</th>
<th>Phase 6</th>
</tr>
</thead>
<tbody>
<tr>
<td>Identification of rehabilitation needs</td>
<td>Literature review</td>
<td>Expert opinion</td>
<td>Integration of evidence &amp; expert consensus</td>
<td>Knowledge translation</td>
<td>Implementation and evaluation</td>
</tr>
</tbody>
</table>

### Data collected from:
- Individual interviews with 6 youth with AMC, 10 caregivers, and 10 OT/PT
- Five focus groups with youth with AMC, caregivers and OT/PT
- A semi-structured survey questionnaire completed by 24 youth & family members

### Phase 1: Identification of rehabilitation needs
- A scoping review for each of the 5 priorities using the framework by Arksey and O’Malley to systematically select and summarize existing literature
- Searches conducted with keywords and MeSH terms from the earliest date available to March 2018
- Findings will be summarized to describe the current standard care

### Phase 2: Literature review
- A survey to identify the treatment methods used and associated outcome measures will be sent to OT/PT working with children living with AMC

### Phase 3: Expert opinion
- Information gathered from the scoping reviews and survey will be gathered and practice recommendations for each priority will be put forward
- These recommendations will be shared and refined with an advisory board comprised of youth and adults with AMC and parents

### Phase 4: Integration of evidence & expert consensus
- Practice recommendations will be translated into tools (infographic, annotated, flowchart, manuals) which will be:
  - Comprehensive, clear, and easy for OT/PT to use
  - Engaging for youth and families

### Phase 5: Knowledge translation
- Evaluation of implementation in various clinical settings to:
  - Tailor the practice recommendations to the different settings
  - Evaluate the impact of these recommendations on clinical practice, client-centeredness and satisfaction

---

**Hôpitaux Shriners pour enfants**
**Shriners Hospitals for Children**
**Canada**
Patient-oriented research in AMC

3. Animated video

“AMC and ME” content development
Translation to French and Spanish
Patient-oriented research in AMC

Other Studies

• Telerehabilitation to develop a home exercise program for youth
• PhotoVoice to empower youth and guide clinical practice
• Development of an AMC-specific outcome measure for the upper extremity
Lessons learned & the future

• POR is worth exploring
• Not all projects readily amenable to POR
• Funding for research partners
• Link with clinical departments and support groups
Funding Support
THANK YOU

Collaborations:
Dr. Reggie Hamdy
Dr. Frank Rauch
Dr. Harold van Bosse
Lisa Wagner
Dr. Judith Hall
Dr. Bonnie Sawatzky
Tanya Bédard
Dr. Laurie Snider
Dr. Cynthia Gagnon

Administrative support:
Carla Evangeliste

Research staff:
Sarah Cachecho
Jessica Collins
Vasiliki Darsaklis
Kathleen Montpetit

Students:
Caroline Elfassy
Marianne Gagnon
Alexa Cirillo
Kevin Caporuscio
Sabrina Shemie
Delphine Jaumard
Koorosh Semzar-Kazerooni
Qi Yue He

Youth and families

noemi.dahan@mcgill.ca