4TH INTERNATIONAL SYMPOSIUM ON ARTHROGRYPOSIS



From the Womb to Adulthood

Post-symposium Booklet

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4TH INTERNATIONAL SYMPOSIUM
ON ARTHROGRYPOSIS

Thank you



We extend our heartfelt thanks to all those who attended the 4th International Symposium for Arthrogryposis in Montreal. Over 180 participants from across 14 countries participated, including more than 40 people living with AMC and family members, 100 medical professionals and clinicians, and trainees. Your presence, engagement, and shared expertise helped make this event a truly meaningful and inspiring gathering. We were honored to welcome clinicians, researchers, individuals living with arthrogryposis, families, and advocates from around the world, all united by a common goal of advancing understanding, care, and support for those affected by this condition.

Your contributions to the discussions, presentations, and workshops enriched the symposium and reinforced the importance of collaboration across disciplines and communities. We are especially grateful to those who shared personal experiences, offering valuable insight and perspective that continue to shape the direction of research and clinical care. Dr. Judith Hall's distinguished keynote provided deep insights, clinical wisdom, and decades of pioneering work, bringing both clarity and inspiration to the field. Our Chairman of the Board of Governors of the Shriners Hospital, Mr. Gary McKeown welcomed everyone to the start of a knowledge and fun-packed event. Our keynote speakers across early detection, genetics, orthopedics, rehabilitation, neurology, care in diverse resource settings, lived experience, and research provided us all with the latest evidence and knowledge in arthrogryposis, and an opportunity to share. Thank you to the members of the scientific committee, who ensured the program was high quality, inclusive and represented diverse perspectives. We acknowledge the important contribution of the members of the organizing committee and of our host institution for this event, the Shriners Hospital for Children in Montreal, for their unwavering support, as well as our generous donors Shriners' Children, MUSCO, the Canadian Association of Shrine Temples, J.E. Hangers, Mary Stillman and anonymous donors.

Thank you for your dedication, your passion, and for making the 4th International Symposium a memorable and impactful event. We look forward to continuing this journey together and sharing more knowledge, connections and networking at the next International Symposium for Arthrogryposis, to take place in Stockholm, Sweden in 2028!

Warm regards from Montreal, Canada

Noémi Dahan-Oliel, PhD **Reggie Hamdy, MD**

4th International Symposium for Arthrogryposis Co-chairs

4TH INTERNATIONAL SYMPOSIUM ON ARTHROGRYPOSIS

PRE-CONFERENCE WORKSHOPS

PRENATAL DETECTION OF AMC: CHALLENGES AND OPPORTUNITIES.

This 2-hour workshop showcased experts in the fields of obstetrics, gynecology, perinatology, clinical genetics, diagnostic medical sonography and lived experience in the Netherlands and the USA. The presenters included Johanna I.P. de Vries, MD, PhD (Netherlands), Jill Tjon, MD, PhD (Netherlands), Maria B. Tan-Sindhunata, MD (Netherlands), A. Arduç, MD (Netherlands), Ingeborg H. Linskens, MD (Netherlands), Remco Jansen (Netherlands), and Sara Lemin, MD (USA). The main challenges in prenatal detection of AMC, in three following areas were presented:

1. Cause, genetic with and without inheritance risk, infection, environmental or unknown.
2. Onset of AMC, during first, second or third trimester of pregnancy.
3. Severity: variation in expression of AMC differs classified into groups, from mild to more severe in Groups 1 (Amyoplasia) and 2 (Distal Arthrogryposis) to mainly severe in Group 3 (CNS involvement) including Fetal Akinesia Deformation Sequence (FADS).

Opportunities to improve prenatal detection from three perspectives were discussed: Amsterdam, Netherlands (The Amsterdam University Medical Center Expertise Center on FADS and AMC team); Ohio, United States (Sara Lemin, Take Time for AMC); and Basel, Switzerland (Isabel Filges, clinical geneticist).

JOINT EFFORTS OF PATIENT SUPPORT GROUPS.

This 1.5 hour workshop was presented by the leaders of three international support groups: Ani Samargian (AMCSI, USA), Victoria Castillo (Artrogryposis, Spain) and Remco Jansen (Spierziekten, Netherlands).

In March 2024, coordinators of the patient support groups met to discuss how to strengthen patient support groups worldwide concerning own organization and value for the AMC community. It was highlighted that collaboration with and between patient support group organizations will lead to a stronger international AMC network. With this purpose in mind, experiences, lessons learnt and suggestions regarding the organization, pride and concerns with patient support groups were shared.

REHABILITATION IN A LIFETIME CONTINUUM - THE 5 W´S FOR AMC

This 2.5 hour workshop was produced by experts in occupational therapy, physical therapy, physical rehabilitation medicine, orthotics and lived experience from five countries: Canada (Noémi Dahan-Oliel, OT PhD; Caroline Elfassy, OT MSc; Sarah Cachecho, OT MSc; Clarice Araujo, OT PhD; Francis Lacombe, Orthotist) France (Mona Hoy, OT; Claire Huzar, PT; Véronique Thellier, OT) Norway (Lena Lande Wekre, MD PhD; Kristin Schewe Loxley, OT) Poland (Alicja Fafara, PT PhD) and USA (Lisa Wagner OT DHS).

The AMC population presents with physical limitations in ROM and decreased muscle strength, which leads to challenges throughout the life span. The individual with AMC however often possesses a certain tenacity and “spunk” that assists them with adjusting to these limitations. The rehabilitation process seeks to gain function and maximize abilities to participate in all areas of life. Rehabilitation focuses on three levels of treatment: (a) body structure- the anatomical parts of the body, (b) activity- the execution of a task or action by an individual, and (c) participation- the involvement in a life situation.

This workshop addressed the rehabilitation journey from the infant stages continuing throughout the lifespan utilizing the important domains of the WHO’s International Classification of Functioning, Disability and Health. International experts shared assessments, interventions, orthotics, and adaptations within the framework of the developmental process addressing the 5 W`s: Who, What, Where, When and Why in the context of rehabilitation.

WHY IS IT IMPORTANT TO GET A PRECISE DIAGNOSIS OF AMC (AFTER BIRTH)?

This workshop was a collaboration of international professionals including: Klaus Dieterich, MD PhD (France), Philip Giampietro, MD PhD (USA), Frank Rauch, MD (Canada), Eva Ponten, MD PhD (Sweden), Andres Nascimento, MD PhD (Spain), Véronique Bourg MD (France), Sara Lemin, MD (USA), and Amanda Gilsin (France).

Arthrogryposis multiplex congenita, or AMC, is a clinical sign defined as congenital contractures of at least two joint levels. These joint contractures are always secondary to diminished fetal movement which can have numerous causes that affect any part of the anatomical structures implicated in movement: the central nervous system, the anterior horn cell, the nerve, the neuromuscular junction, the muscle, or the joint itself. Making a precise diagnosis of AMC is therefore challenging.

The aim of this workshop is to share the expertise of clinical geneticists, basic scientists, medical professionals in the fields of neurology, orthopedics and obstetrics, and people with lived experience to gain a better understanding of the use and diagnostic value of common examinations and analyses performed postnatally in patients affected by AMC.



ORAL PRESENTATIONS

Based On Submitted Abstracts

Growth Charts for Children with Arthrogryposis Multiplex Congenita

Lauren Hyer[1], Emily Shull, David Westberry

[1]Shriners Children's Greenville

Children with arthrogryposis multiplex congenita (AMC) often demonstrate growth differences compared with typically developing (TD) children. However, growth charts have not been developed for this population. The purpose of this study was to create AMC-specific growth charts and to compare these values to those of TD children. A retrospective review of height/length and weight for 206 children with AMC was performed. Growth charts were developed and stratified over seven percentiles; these were then compared with growth charts of TD children. Children with AMC tend to be smaller in stature and weight compared with TD children, particularly in the first 36 months of life. Thereafter, weight values trend toward the 50th percentile of TD children, but height/length values persist around the 5th percentile of TD children. The development of AMC-specific growth charts provides health care providers an objective tool to evaluate growth patterns of patients with AMC.

Functional Independence of Children with Arthrogryposis

Lauren Hyer[1]

[1]Shriners Children's Greenvillle

Background: Arthrogryposis (AMC) is a descriptive term to characterize a child born with multiple joint contractures. Treatment aims to improve functional independence, yet the literature objectively describing functional independence in this population is scarce. This study aimed to describe the functional independence of children with AMC through the lens of the

Pediatric Evaluation of Disability Inventory-Computer Adaptive Test (PEDI-CAT) and observational activities of daily living (ADL) tasks. Methods: Patients with AMC between the ages of 3 and 12 years participated in this prospective study. Parents completed the PEDI-CAT while a trained occupational therapist observed children as they completed a checklist of functional ADL tasks. Patients were grouped according to developmental age groups: "preschoolers" (3 to 5 y), "early school-age" (6 to 9 y), and "late school-age" (10 to 12 y). Patient's PEDI-CAT normative scores were described, comparing the study population to typically developing children, and differences in each domain were examined between developmental age groups. The observed ADL tasks completed were also described, and differences in scores were examined between developmental age groups. Results: Forty-four patients (mean age of 7 ±2.86 y) were enrolled. The distribution between age groups was nearly even. Mean daily activities T-score for patients with AMC was 25.80 ±11.98 and the mean mobility T-score was 17.39 ±9.77. Late school-age children scored significantly lower than preschool-age children in both of these domains (P<0.01). Observed ADL tasks demonstrated a high level of required assistance for patients (range: 27.3% to 61.4%), although older school-age children did show greater independence with tested activities than preschool-age children (P=0.05). Conclusion: Children with AMC are significantly limited in functional independence, particularly regarding age-appropriate daily activities and mobility. Outcomes from this study provide a reference to help gauge the results of nonoperative and surgical treatment toward improving functional independence in this population.

Health-Related Quality of Life in Physical Disabilities of Childhood

Maureen Donohoe, Reid Nichols[1], Stephanie Butler, Nancy Lennon, Wade Shrader, Chris Church, Jose Salazar-Torres, Stuart Mackenzie, Sana Patil, Faithe Kalisperis, Freeman Miller

[1]Nemours Children's Health Delaware

Background: Use of patient reported outcomes is essential to best understand and manage health-related quality of life (HRQOL) in youth with lifelong disabilities. This study evaluated HRQOL in youth with physical disorders and examined its relationship with mobility. Methods: In this IRB approved retrospective study, the parent-reported Pediatric Outcomes Data Collection Instrument (PODCI), and the Gross Motor Function Measure (GMFM-D) were administered to ambulatory youth aged 2-18 with cerebral palsy (CP; GMFCS II; n=258), arthrogryposis (n=138), achondroplasia (n=102), and Morquio syndrome (n=52) during clinical visits to the gait laboratory. The PODCI has two validated versions, a child(age 2-10) and adolescent (age 11-18) that assess perceptions about mobility, happiness, and pain. Differences in HRQOL between diagnostic groups, between age groups, and compared to typically developing youth (TDY) were examined using non-parametric tests. The relationship between PODCI and GMFM-D scores was analyzed with Pearson correlations. Results: Both age cohorts (child and adolescent) within all diagnosis groups demonstrated lower mobility and higher pain compared to TDY (p<0.015). Happiness was lower for both the child and adolescent groups with CP and arthrogryposis, and for the child group with Morquio syndrome compared to TDY (p<0.002). Among diagnostic groups at both age spans, global function was higher (p<0.0001) for achondroplasia compared to other groups. Despite functional differences, there were no significant differences between the diagnostic groups in pain scores (p>0.10). Happiness was lower in the CP group compared to achondroplasia (p=0.01). GMFM-D was associated with the PODCI mobility domains for all diagnoses (r=0.31 to 0.79, p<0.03), but was not correlated with happiness for any group (r=-0.16 to 0.092; p>0.14), GMFM-D and PODCI pain scores were correlated only for the child group with achondroplasia (r=0.355; p<0.001). Conclusions: Significant limitations in HRQOL are present in youth with physical disabilities. Pain levels were higher than non-disabled peers, but pain was not related to lower motor function. Happiness was not associated with gross motor function suggesting the need to examine other contributors when mental health concerns exist in youth with disabilities.

Pediatric Pain & Function in Arthrogryposis: Prevalence, Principles, and Plans of Care

J. Megan Sions[1], Maureen Donohoe[2]

[1]University of Delaware, Department of Physical Therapy

[2]University of Delaware, Biomechanics & Movement Science Graduate Program

A lifespan approach to management of arthrogryposis is often focused on function. This is quite important, but evaluation and treatment of pain is rapidly evolving, with evidence supporting pain is a multi-dimensional, personal experience shaped by past experiences. Among children, pain is under-recognized and under-treated. Our collaborative research between Nemours Children's Health and the University of Delaware found that pain affects >70% of children with arthrogryposis and is associated with reduced functional mobility. Given arthrogryposis affects two or more body areas, unsurprisingly, multisite pain is common, affecting >50% of children. More pain complaints are found for the feet, knees, and spine, as compared to the upper extremities, which may be explained by repetitive, abnormal loading of the lower extremities with ambulation. The session will share reliable pain-related metrics and functional outcome measures that can be used in children with arthrogryposis to track pain intensity and extent, as well as associated functional decline over the lifespan. Content will include pediatric acute, chronic, post-operative, and procedural pain management and activities to enhance functional mobility. Functionally directed treatments that incorporate play, strengthening, falls mitigation, and energy conservation will be shared by a physical therapist, Dr. Maureen Donohoe, PT, DPT, who is a Board-Certified Pediatric Clinical Specialist with over 35-years of clinical experience treating children with arthrogryposis. Pain-related content will be provided by Dr. Megan Sions, PT, DPT, PhD, who is a Board-Certified Orthopaedic Clinical Specialist with 20 years of clinical practice experience, a pain researcher, and a mother of a child with arthrogryposis.

Prenatal diagnosis (or lack thereof) of arthrogryposis multiplex congenita (AMC) and its impact on the perinatal experience of parents: a retrospective survey

Sara Lemin[1], Harold van Bosse, Lauren Hutka, Shea Soberdash, Jay Patibandla

[1]Aultman Hospital/NEOMED

Objective: To examine parental experiences during pregnancies affected by Arthrogryposis Multiplex Congenita (AMC) identifying commonalities, risk factors, and areas for improvement in detection rates, care protocols, and patient experience. Study Participants & Setting: An online survey was distributed via AMC support groups on Facebook. A total of 301 responses from 23 countries met inclusion criteria of being the biological parent of a pregnancy in which the child had two or more joint contractures present at birth. The exact number of potential/eligible participants who viewed the online invitation to participate is not known. No external or commercial funding was obtained and ethical approval was waived by our institution as no personal identifying information was collected. Methods: The 57-question survey was hosted online at SurveyMonkey.com. Quantitative topics included demographics, risk factors, parental recall of sonographic findings, delivery characteristics and neonatal findings. Responses were analyzed with the Fisher exact test. Qualitative questions about parents' experiences were collected with free-text responses and analyzed with thematic analysis. Responses were reviewed collectively and then divided into antenatally detected cases (ADCs) and postnatally detected cases (PDCs). Results: The antenatal detection rate of arthrogryposis was only 37%. Fetal movement was described as decreased by 54% or absent by 12% of respondents. Early bleeding was reported by 21%. Sonographic findings in ADCs included club foot (83%), clenched hand (51%), decreased fetal movement (50%), elbow contracture (51%), and knee contracture (46%). Among ADCs, 29% delivered vaginally and 71% by cesarean vs PDCs (44% vaginal, 56% cesarean). Overall NICU admission rate was 63%. Bone fracture occurred in 9% (6% vaginal vs 11% cesarean; 11% ADCs vs 8% PDCs). Detection led to a planned change in delivery mode in 33% and location in 50%. Among ADCs, 17% felt their concerns were not adequately addressed versus 43% of PDCs. Conclusions/Significance: Antenatal detection of arthrogryposis was low. We propose enhanced screening criteria to aide prenatal diagnosis, raise awareness among healthcare providers, and promote utilization of more robust practice guidelines.

AMC and the importance of sports/ physical activity and pain management

Carolina Navalon-Martinez[1]

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Presentation to be based on the following topics:

- Revision of clinical trials showing the benefits of sport (physically & emotionally) for people with disabling disease (explain why talk about disabling disease vs disability
- Importance of the practice of sports since an early age (why, how)
- A healthy how to transition to adult age
- How pain can affect this transition and how to overcome barriers
- Presentation of the initial results of the 1st European Therapeutic SUP (Sant Up Paddle) project: initial medical assessment on the benefits of SUP on people with musculoskeletal disorders (1 pax with AMC)

(Project is sponsored through an Art Foundation based in Barcelona. Project medical assessment from a recognized Sports Medicine Unit from Barcelona representing two reference hospitals. Project being coordinated with the help of Carolina Navalon, representing patient's organisations, among them, Artrogriposis España)

Restoration of elbow active flexion in children with amyoplasia: what better to do and when to do?

Olga Agranovich[1], Ekaterina Petrova, Svetlana Trofimova , Evgenia Kochenova, Sergey Batkin, Dmitry Buklaev

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Background and Objective(s). The absence of active elbow flexion is the most common problem in children with amyoplasia, leading to a violation of daily living. Pectoralis major muscle (PM) and latissimus dorsii muscle (LD) are the most used muscles for this purpose. There are no information in the literature about what muscle better to use and what optimal age for these operations. Study Participants & Setting. The retrospective study involved 61 patients (90 upper limbs) with amyoplasia (30 (49%) girls and 31 (51%) boys) who were examined and treated from 2011 to 2020. In 46 cases (51,1%) we used PM and in 44 (48,9%) LD as donor muscles. Methods. The clinical examination of the patients was carried out before and after operation (6 months and more). Statistical data processing was performed using the software packages Statistica 10 and SAS JMP 11. Results. The age of patients at the time of surgery was from 1,5 to 15,5 years (6,24 ± 4,24 лет), the follow-up period after surgery was from 6 to 99 months (41.25 ± 30.19). After surgery all patients had elbow flexion contractures however, when was used as a donor muscle LD the degree of contracture was less then after PM transfer (15.19° ± 13.04 and 23.24° ± 15.37, respectively). In addition, after the LD transfer the strength of the forearm flexors was on average 1 point greater than after the PM transfer (2.85 ± 1.08 and 4.00 ± 0.62 points, respectively). After LD transfer the amplitude of elbow active flexion was bigger compared to the PM transfer (75.37 ° = 17.86 and 55.88° = 24.60, respectively). The greatest dynamics of such indicators as the strength of forearm flexor muscles, the active elbow flexion, the function of the elbow were noted in patients at the age from 1 to 3 years. Conclusions/Significance. The study demonstrated the effectiveness of using LD and PM for restoration of elbow flexion in children with amyoplasia however, if it is possible to choose a donor muscle, the preference should be given to LD. We recommend these operations at the age from 1 to 3 years.

Unravelling the Association between Developmental Milestones Attainment among Children with AMC and Contextual Factors: Multicentric Cross-Sectional Study

Ahlam Zidan[1,2], Verity Pacey, Laurie Snider, Reggie Hamdy, Frank Rauch, Lauren Hyer, Michelle James, Haluk Altiok, Ellen Raney, Cary Mielke, Sarah Nossov, Sena Tavukcu, Philip Giampietro, Noémi Dahan-Oliel

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Background and Objective(s): Developmental milestones serve as important signposts for evaluating infant growth during the critical postnatal phase. Arthrogryposis multiplex congenita (AMC), with its heterogeneous clinical presentation, can significantly impact a child's ability to attain developmental milestones such as sitting, standing, and walking. Such impairments can predict physical limitations and disability later in life. The developmental delay was investigated in children with other congenital conditions but has yet to be studied in the AMC population. It behooves researchers to better understand the early developmental trajectory of AMC children. This study aims to determine the contextual factors associated with attaining developmental milestones among children aged 0 to 5 years with AMC. Study Participants & Setting: A total of 101 participants aged 0-5 years were recruited between January 2019 and December 2022 through the AMC registry, which was implemented in 2019 across eight Shriners hospitals in Canada and the United States. Methods: In this multicenter cross-sectional study, data from the Ages & Stages Questionnaire (ASQ-3), a developmental screening tool, was completed by parents. ASQ-3 assesses communication, gross motor, fine motor, problem-solving, and personal social skills in children aged 1 to 60 months. Descriptive statistics (mean, median, standard deviation) were calculated using SPSS software. Univariate and multivariate logistics regression were used to characterize the association between children's ASQ-3 data, representing developmental milestones attainment, and the role of contextual factors (age, sex, gestational age, birth weight, days in neonatal intensive care unit, parental stress, contracture location and severity, AMC classification; hand and toe anomalies, spine anomalies, surgical intervention's location, also parent age, marital status, employment, education, and stress) in these associations. Results: Analysis is currently underway. This research will generate new knowledge and enhance our understanding of contextual factors' impact and their associations with reaching developmental milestones among young children with AMC. Conclusions/Significance: Understanding these associations can assist clinicians and researchers in making the appropriate decisions regarding therapy and monitoring the functional progress of a child with AMC. Our results can inform researchers' hypotheses and rationale for studying specific factors that might assist in predicting physical limitations and disability later in life.

Psychometric Evaluation of Four Mobility Measures in children and adolescents with Arthrogryposis Multiplex Congenita (AMC).

Ahlam Zidan[1, 2], Verity Pacey, Laurie Snider, Frank Rauch[2], Lauren Hyer, Ellen Raney, Reggie Hamdy, Michelle James, Philip Giampietro, Cary Mielke, Sarah Nossov, Sena Tavukcu, Haluk Altiok, Noémi Dahan-Oliel

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Background/objective(s): Children with arthrogryposis multiplex congenita (AMC) show limitations in daily function and mobility. Due to the heterogeneous clinical presentation of AMC, clinicians should offer tailored assessments to monitor functional progress. Clinicians and researchers employ measures not formally validated for the AMC population. Validated measures can underpin the reliable interpretation of mobility status and the appropriate allocation of resources in line with patient needs. This study aimed to estimate the internal consistency reliability and construct validity of four commonly used mobility measures. Participants/Setting: In total, 234 individuals aged 5-21 were recruited between January 2019 and December 2022 through the AMC registry, implemented in 2019 across eight Shriners hospitals in Canada and the United States. Methods: Internal consistency of the Functional Mobility Scale, Gillette Functional Assessment Questionnaire, Functional Independence Measure for Children, and Patient-Reported Outcomes Measurement Information System was evaluated with Cronbach's alpha to verify whether all items within each of the four mobility tools measured the same construct (a≥0.7 considered acceptable). The convergent and divergent validity of these four measures was assessed using Spearman or Pearson correlation analysis, including estimates of the correlation coefficient (r) (95% CIs). The Independent Samples t-Test was used to assess known-group validity. Results: Analysis is currently underway. We hypothesize that the scores of the four mobility measures will show high reliability and have strong to very strong positive correlations for convergent validity (r≥0.4). For divergent validity, the scores of these measures will have a weak correlation with the upper extremity domain of PROMIS. Finally, our hypothesis for known-group validity is that mobility scores would be better in children and youths with only upper limb involvement as compared to children with only lower limb involvement, both limbs involvement, and those with central nervous involvement. Conclusions/Significance: These findings can help clinicians and researchers make wise decisions regarding which outcome measurement is more relevant to their patient's needs, and they can trust the validity of these measures' interpretation of a child's mobility status. Additionally, valid measures can be employed in research to understand the associations between different contextual factors and mobility functions in children and adolescents with AMC.

Exploring the impacts of COVID-19 on individuals with Arthrogryposis Multiplex Congenita

Megan MacGillivray[1], Sophie Bezanson, Bonita Sawatzky

[1]St. Francis Xavier University, Canada

Background: The COVID-19 pandemic has disproportionately impacted people with disabilities. Many policies intended to slow the spread of the virus have been associated with reduced access to health care services and adverse social impacts for people with disabilities. Our study aims to explore the health and social-related impacts of the pandemic on people with Arthrogryposis Multiplex Congenita (AMC). AMC is a rare condition characterized by multiple joint contractures and muscle weakness. We aim to better understand the lived experiences during the pandemic to inform future public health policies and ensure people with disabilities are included in public health responses. Methods: We invited participants enrolled in the AMC Adult Registry (Sawatzky, 2019) to take part in the COVID-19 study. The Qualtrics survey included 49 questions related to the health and social impacts of the pandemic. Data was descriptively analyzed. Results and Analysis: Respondents (n=27) were from the US, Canada, Australia, Norway, and the UK. Respondents who received a COVID-19 vaccine were administered in their arm (89%) and thigh (11%). 42% received the COVID-19 vaccine earlier than their peers due to perceived health risk. 59% of participants reported contracting COVID-19 at least once, with 25% of these individuals identifying that they had long COVID. In terms of care experience, 33% of respondents required assistance during the pandemic. Of these respondents, 11% experienced difficulty accessing care aids and 33% experienced increased anxiety with care aids entering their homes. Overall, 44% of respondents required assistance and adaptations for masking. Family caregivers or care aids who followed public health protocols created positive care experiences. Conclusion: People with AMC have challenges adhering to certain COVID-19 guidelines due to physical disability. We need to consider different abilities and needs in designing public health policies to ensure that such guidelines are accessible and identify areas where supports can promote health equity and dignity.

Phases of development of the Shriners Hospitals Arthrogryposis Pediatric Evaluation - UPper Extremity (SHAPE-UP) and initial psychometric evaluation

Caroline Elfassy[1], Lisa Wagner[2], Laurie Snider, Sarah Cachecho, Rose Elekanachi, Tessah Dunn, Clarice Araujo, Reggie Hamdy, Johanne Higgins, Kathleen Montpetit, Chantal Janelle, Lauren Hyer, Felicity Fishman, Krister Freese, Michelle James, Dan Zlotolow, Noemi Dahan-Oliel

[1]Shriners Canada

[2]Shriners Greenville

Background: Approximately 56% of patients with AMC have upper and lower extremity involvement, while 17% have upper alone. Upper extremity (UE) function plays a crucial role in the overall quality of life and autonomy of individuals with AMC. Currently, there does not exist any specific outcome measure for children with AMC. Generic measures do not give information on how the task is performed, which joint or muscle is limiting function, and cannot be used to compare to normative data. Aim: This multi-phase project was achieved by 1) identifying which performance-based outcome measure are most frequently reported to evaluate UE function in pediatric rehabilitation with a link between constructs of the ICF and meaningful concepts extracted, 2) determining the most clinically useful items for an outcome measure of UE function for children with AMC as defined by caregivers and clinicians, and 3) evaluating the psychometric properties of the SHAPE-UP Task Completion scale. Methods: Aims 1) a scoping review of the literature in addition to the Linking Rules was performed, 2) for the caregiver engagement, a nominal group technique was administered, while a three-round survey was carried out for the clinician group, 3) a cross-sectional study was employed for the initial psychometric evaluation of the SHAPE-UP. Results: 1) 32 outcome measures published between 1948-2013 were included and 538 meaningful concepts were extracted and linked to the ICF, 2) 11 individuals participated and according to the voting system, the most important and essential items identified were the following: Picking up an object, Eating, Reaching mouth, Getting out of bed 3) Based on the results of the previous phases, the rough draft of the SHAPE-UP was created. The SHAPE-UP consisted of 3 descriptive questions and 11 performance-based video-recorded tasks that were administered by an OT or PT. The measure was designed to focus on two scales: Task Completion and Analysis of Joint Motion and Position. SHAPE-UP was administered to 92 participants and a Rasch analysis was performed. The results indicated a need to condense Task Completion scores and revise Joint Analysis. The developers incorporated these results into the final version of the SHAPE UP.

Is There a Genetic Basis in Amyoplasia ?

Philip Giampietro[1], Frank Rauch, Reggie Hamdy, Ellen Raney, Krister Freese, Sarah Nossov, Haluk Altiok, Sena Tavukcu, Noemi Dahan-Oliel

[1]University of Illinois-Chicago

Amyoplasia refers to absence of muscle in upper and lower extremities and accounts for approximately half of arthrogryposis multiplex congenita (AMC). Patients present with bilateral symmetric involvement of upper and lower extremities in the absence of central nervous system and a positive family history. While no genetic marker for Amyoplasia has been identified, the current hypothesis is that Amyoplasia is vascular mediated and genetic testing has not been typically warranted. Shriners Hospital has established a Registry for phenotypic and genotypic description of AMC. Following consent, participating subjects provided a saliva sample, access to medical records and completed patient-reported outcomes. Photographs and clinical history corresponding to enrolled cases were subsequently reviewed by research study team members. Among a series of 247 enrolled cases in the Shriners AMC Registry, 116 (47%) cases had Amyoplasia, with 40 cases having clinical features consistent with classic Amyoplasia and 76 cases having features consistent with atypical Amyoplasia (e.g., asymmetrical involvement bilaterally and/or of upper and lower limbs, abnormal facies). Whole genome sequence analysis (WGS) was performed on saliva samples obtained from forty-five enrolled patients with Amyoplasia (32 atypical and 13 typical) with a yield of 16% of samples having a pathogenic mutation. Mutations were identified in ATL1, BICD2 (n=2), FHL, LMNA, TRPV4, and TTN. These findings support the incorporation of genetic consultation in conjunction with the implementation of whole genome sequence analysis to identify a molecular diagnosis in patients presenting with features of typical and atypical Amyoplasia. The benefits of obtaining a genetic diagnosis for these patients include a more precise diagnosis, which will facilitate optimal genetic counseling, treatment and help guide therapeutic strategies.

Using Telerehabilitation to Deliver a Home Exercise Program to Youths with Arthrogryposis

Marianne Gagnon[1, 2], Gabriela Marino Merlo, Rita Yap, Jessica Collins, Caroline Elfassy, Bonita Sawatzky, Jacquelyn Marsh, Reggie Hamdy, Louis-Nicolas Veilleux, Noémi Dahan-Oliel

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Background and Objectives: Exercises have the potential to maintain range of motion (ROM) and muscle strength in individuals with arthrogryposis multiplex congenita (AMC). However, access to specialized care may be limited because of geographical distance. This study evaluates the feasibility and explores the effectiveness of delivering a home exercise program (HEP) through telerehabilitation. Study Participants & Setting: Seven participants (5 male, median age: 16.9 years) with a diagnosis of AMC living in Canada completed the study. Methods: Pre- and post-intervention, participants completed the Physical Activity Questionnaire for Adolescents (PAQ-A) and the Pediatrics Outcomes Data Collection Instrument (PODCI) to evaluate pain, function and physical activity. The clinicians measured active ROM using a virtual goniometer and used the Goal Attainment Scale (GAS) to identify goals and develop a 12-week individualized HEP. To assess the feasibility, the satisfaction with the approach and the compliance with the HEP were collected. The interrater reliability of using a virtual goniometer was assessed using intraclass correlation coefficients (ICC). Nonparametric tests were used to evaluate feasibility and explore the effectiveness of the HEP. Results: The participants performed their HEP 2.04 times per week (95% CI 1.25-4.08) and reported good satisfaction with the approach. The inter-rater ICC ranged from 0.410 (95% CI:-0.392; 0.753) for forearm pronation to 0.998 (95% CI:0.996; 0.999) for elbow flexion measured with the virtual goniometer. Individualized goals were related to pain management; endurance in writing, standing, or walking; sports; and daily activities. In total, 12 of the 15 goals set with the participants were achieved. Statistically significant improvements were observed in the pain domain of the PODCI (pre-intervention: median 71; 95% CI 34-100; post-intervention: median 85; 95% CI 49-100; p=.08) and PAQ-A (pre-intervention: median 1.62; 95% CI 1.00-2.82; post-intervention: median 2.32; 95% CI 1.00-3.45; p=.046). Conclusions/Significance: Results demonstrate that telerehabilitation is a feasible approach to providing HEPs to youths of different functional levels across Canada. Promising results were found for the effectiveness of the HEP in helping youths with AMC to achieve their goals. The next step will be to assess the effectiveness of this exercise intervention in a randomized controlled trial.

Posterior artrolysis of the elbow, as a method of correction extensor contracturing of the elbow in children with arthrogryposis.

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Aim. To evaluate the results of the correction of the extensor contractures of the elbow joints after posterior arthrolysis of the elbow joint with lengthening (Z-shaped, according to the V-Y technique) and without lengthening the triceps of the shoulder in children in different age groups. Materials and methods. From 2008 to 2023 was performed to increase passive flexion in the elbow joint in 109 patients with arthrogryposis with extensor contractures in the elbow joints (158 joints) posterior arthrolysis of the elbow joint. Results. All children were divided into groups depending on the age when the operation was performed and the method of surgical correction. The follow-up period in the postoperative period in the main group of patients in 64.3% of cases was 3-4 years. In groups 1 and 2 of children who did not lengthen the triceps, good treatment results were observed in 95.83% compared to groups 3, 4 and 6 in children who had good results in 85.56%. The range of passive movements after surgery increased in children under 1 year . However, extension was less limited in children who did not lengthen the triceps muscle. Over 3 years increased to satisfactory results. Discussion. This study showed that posterior arthrolysis of the elbow joint is best performed in children with arthrogryposis under the age of 1 year with extensor contractures in the elbow. The posterior release without lengthening the triceps muscle should be performed only in children whose flexion was no more than 120 ° . Flexion contractures in the elbow joints that required additional correction developed in children who had a release with lengthening of the triceps in 3-7 years, 45%, older than 7 years - 40% of cases. Conclusions. Received a passive range of motion in the elbow joint in children with arthrogryposis after posterior arthrolysis of the elbow joint allowed the child to use adaptive mechanisms for self-care. When we performed posterior arthrolysis of the elbow joint with lengthening of the triceps, the result of the treatment did not depend on the elongation technique, but was directly dependent on the age of the patient when the operation was performed.

Does Open Reduction of Arthrogrypotic Hips Cause Stiffness?

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Background: Concerns of potential hip stiffness following open reduction for congenitally dislocated hips in arthrogryposis multiplex congenita (AMC) persist, despite published accounts of positive outcomes. We hypothesized that the pre-existing limitation to hip motion was minimally affected by relocation and retrospectively compared preoperative and postoperative hip motion and evaluated the ambulatory abilities of patients who underwent the procedure. Methods: Over a ten-year period, 52 patients with arthrogryposis had 75 open reductions for congenitally dislocated hips with a minimum follow-up of 2 years. The mean patient age was 23 months. Hip range of motion (ROM) data was recorded pre-operatively, post-operatively, and at last follow-up, as was ambulatory ability. Radiographs were assessed for hip station and avascular necrosis (AVN). Results: Average follow-up was 68 months; 59% had concurrent femoral shortenings. Preoperatively, 34 hips had a mean flexion contracture of 33°, improving 22° at follow-up; 59 hips had <45° frogleg abduction and 39 hips had <30° abduction preoperatively, improving by 11° and 10°, respectively (all p-values ≤ 0.001). The flexion-extension total arc of motion (TAM) decreased by only 1° from preoperative to follow-up (p = 0.733). Patients with unilateral dislocation (n=29) had no significant difference in TAMs between the dislocated and non-dislocated hips at final follow up. Hips with <90° of flexion preoperatively showed no improvement following reduction. At a mean follow-up of 68 months

(24-152 months), 31 patients were independently ambulatory, 15 were walker-dependent but still progressing, and 6 remained non-ambulatory. AVN was seen in approximately half the hips but without affecting flexion-extension TAM at follow-up; the majority were transient changes (delayed ossification). Conclusions: Open reduction for arthrogrypotic hip dislocations maintains mobility without causing stiffness. While pre-existing hip motion limitations slightly worsened, lower limb positioning improved, especially in hip extension and abduction, enhancing ambulation. Most achieved independent ambulation.

Correction of mild to moderate arthrogrypotic knee flexion contractures with guided growth

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Methods: Medical records and radiographs of 57 consecutive pediatric patients (90 knees) with arthrogryposis and knee flexion contractures treated with FADHE were reviewed. Mean age at surgery was 7.1 years, and mean follow-up length was 4.2 years (17 months - 10 years). Posterior releases were performed concomitantly in 67 knees, including hamstrings lengthenings, and proximal gastrocnemius and posterior capsule releases; 6 had only percutaneous medial hamstring tenotomies; the remaining 17 had no releases. Post-operatively, the patients were wore a nighttime knee-ankle-foot orthosis (KAFO) in full extension until skeletal maturity. Results: Pre-operative mean knee flexion contracture, 35° (range 10°-60°) improved to 7° at hardware removal, which was done 17.5 months (range 4-41) after implantation, for an average correction rate of 7°/100 days. Contractures ≤ 25° that did not have a posterior release corrected well. Knees with 30° - 40° contractures corrected well with the addition of a posterior release, but >40° saw only 50% of knees correcting to ≤10°. The mean total arc of motion was 71° preoperatively and 61° degrees at followup. At latest follow-up, the mean contracture was 19°, and all knees had ligamentous stability, although 9 patients (14 knees) underwent repeat hemiepiphysiodesis for contracture recurrence at 39 months after plate removal. Nine patients who were previously nonambulatory became ambulatory, and 12 that ambulated with a walker/crutches became independent. Conclusions: Knee flexion contractures in growing children with arthrogryposis can be treated with anterior distal femoral hemiepiphysiodesis for contractures ≤25°, and relatively reliably for contractures ≤40° if a posterior knee releases is included. Greater than 40° contractures may respond in certain cases. Recurrence of the contracture is common due to growth, but did not impact on ambulatory gains made; repeat hemiepiphysiodesis effectively treated recurrences. A KAFO is essential to help stretch soft tissues during the correction as well as after the plates have been removed, to help prevent or slow recurrence.

Bridging Understanding: Fostering Informed Healthcare Provider Communication for Individuals with Arthrogryposis

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Background: Allied health professionals continue to report a lack of knowledge of Arthrogryposis Multiplex Congenita (AMC) and effective interventions. When surveyed about their experiences, adults with AMC expressed that even when their healthcare professionals demonstrated expertise and competence, the patients themselves knew more about their own condition and needs. This is due to a lack of knowledge translation of AMC. Knowledge translation is recognized as a means to bridge the gap between what is known about a condition and what is done in clinical practice. This project aims to directly address this gap in knowledge by creating an educational infographic and providing it to individuals with AMC, who can in turn use the infographic to supplement conversations with their healthcare providers about their specific condition and needs. Using a self-efficacy theoretical base, this project aims to empower individuals with AMC to advocate for their needs in the healthcare setting. Methods: Data collection will take place from May 2024-August 2024. A knowledge translation approach will be used. A set of five focus groups will allow adults with AMC to explore the barriers and supports they face when working with healthcare practitioners and identify key information they want healthcare professionals to know about AMC. Focus group data will be analyzed by a panel of AMC experts. This will inform the creation of a knowledge translation infographic outlining the takeaways from the focus groups. This infographic will be piloted at the AMCSI 19th Annual Conference in July of 2024 where individuals with AMC can provide feedback and suggest amendments. Plans for publication and dissemination of this project's findings and infographic will then take place, and be made available to individuals with AMC to use to enhance their interactions with their healthcare professionals. Discussion: This project will provide individuals with AMC a research-based tool to enhance their conversations with their healthcare practitioners regarding their medical needs. This project will utilize the tenants of self-efficacy theory and knowledge translation to contribute to a larger tool kit of resources for individuals with AMC and their associated care teams.

Health-Related Quality of Life in Physical Disabilities of Childhood

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Introduction: Health-related quality of life (HRQOL) is defined as wellbeing with respect to a person's health. This study evaluated HRQOL in children with common disabilities and its relationship with gross motor ability. Participants and Methods: In this IRB approved retrospective study, the parent-reported Pediatric Outcomes Data Collection Instrument (PODCI), and the Gross Motor Function Measure (GMFM-D) were administered to ambulatory children with Cerebral Palsy (CP; GMFCS II), Arthrogryposis, Achondroplasia, or Morquio syndrome during clinical visits to the Gait Laboratory. The PODCI assesses perceptions of mobility/happiness. PODCI results were compared to norms (TDY) using T-tests. The relationship between PODCI and GMFM-D scores was analyzed with Pearson correlations. Results: Children within all 4 diagnoses demonstrated limited mobility and higher pain compared to TDY ($p<0.015$), and happiness was lower in children with CP, Arthrogryposis, and Morquio Syndrome ($p<0.002$). GMFM-D was associated with the PODCI mobility domains for all diagnoses ($r=0.31$ to 0.79, $p<0.03$), but was not correlated with the happiness domain for any group ($r=-0.16$ to 0.092; $p>0.14$), and PODCI pain scores were correlated only in children with Achondroplasia ($r=0.355$; $p<0.001$). Conclusion: Significant limitations in HRQOL are seen in children with common physical disabilities of childhood. Pain is higher in disabled individuals and, like happiness, tends not to be related to low motor function. These findings suggest the need to examine other contributors when mental health concerns exist in children with physical disabilities. Patient reported outcomes must be used to assist in the management of HRQOL in children with lifelong physical disabilities.

Inferring the Cellular Origins of Arthrogryposis and Other Developmental Phenotypes: A Novel Computational Framework Using scRNA-seq Data

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Background/Objectives: Over 400 genes are associated with arthrogryposis multiplex congenita; yet we still don't understand how mutation of these genes produces fetal akinesia and results in congenital contractures. Questions remain about which cell types, at which developmental stages, initiate arthrogryposis when these genes are disrupted. Answering these questions is critical for understanding arthrogryposis, as well as thousands of other developmental phenotypes. Therefore, we are developing a computational framework to discover the cellular etiology of developmental phenotypes. Based on the well-supported observation that disorders with a shared phenotype arise from overlapping molecular processes, we hypothesize that these processes are often localized to a small set of cell types at specific developmental stages. Our two aims are to develop a tool to discover the cell types whose disrupted intrinsic (Aim 1) and extrinsic (Aim 2) processes putatively initiate arthrogryposis or any other developmental phenotype, as well as the impacted developmental stages. Study Participants/Setting: We are using publicly available scRNA-seq atlases. These data are collected from 61 wild-type whole mouse embryos from developmental stages E9.5 to E13.5, and from 15 healthy tissues collected from 28 human fetuses at 72 to 129 days post-conception. Methods: The premise of our approach is the identification of molecular pathways shared among disorders with a common phenotype, localized to implicated cell types. Using scRNA-seq data in combination with sets of phenotype-associated genes, we are inferring cell type-specific gene regulatory networks and cell-cell communication networks and searching them for modules of significantly connected phenotype-associated genes. We will evaluate our framework on a set of selected phenotypes with well-understood etiology. Results: Upon completion, we expect to see specific cell types at particular developmental stages associated with the phenotype of interest based on the presence of significantly connected phenotype modules. Conclusions/Significance: Current treatments of arthrogryposis, while beneficial, still have limited success. By uncovering the cellular origins of arthrogryposis and other developmental phenotypes, we gain insight into their root cause, informing future treatments. Our project takes a major step toward elucidating these etiological processes by developing a computational tool to infer which cell types, at which developmental stages, initiate each phenotype's pathogenesis.

Surgical Treatment of Children under the age of 3 years with Hip Dislocation in Amyoplasia-Type Arthrogryposis: A Rational Approach to Treatment Selection

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Background and Objective(s). Patients with amyoplasia-type arthrogryposis and hip dislocation have different variants of hip contractures and deformities, but there is no difference in the selection of the type of surgery. The study aim to justify and evaluate the effectiveness of the original algorithm of the rational selection of surgical approaches. Study Participants & Setting. 70 patients were examined: 21 children under the age of 1 year (main group) after 25 hip open reduction, 19 children aged from

1 1/2 years to 3 years (main group) after hip open reduction, Salter innominate osteotomy, proximal femoral osteotomy and 30 patients aged 3 to 7 years (control group) who had not previously received conservative and surgical treatment. All patients were divided into two subgroups depending on the variant of hip contracture: flexion-extension-abduction-external rotation (frog-like) (subgroup 1) and flexion-extension-adduction-external rotation (subgroup 2). Methods. Clinical, radiological, statistical methods. Results. In children of subgroup 1, after hip open reduction, good results were noted in 17% of cases, satisfactory in 50%, unsatisfactory in 33%, severe complications class III, IV according to the modified Clavien-Dindo-Sink classification were obtained in 83%. After hip open reduction, Salter innominate osteotomy, and femoral osteotomy performed in patients of subgroup 1 good results were noted in 50% of cases, satisfactory and unsatisfactory each in 25% with less severe complications (50%) (p=0.041). In children of subgroup 2, after hip open reduction, good results were obtained in 90% of cases, satisfactory in 10% with a rate of severe complications of 10%, and when this surgery was combined with Salter innominate osteotomy, femoral osteotomy, good results were noted in 75% of cases, satisfactory in 19% and unsatisfactory in 6%, with a rate of severe complications of 25% (p=0.05). Conclusion. A differentiated treatment approach of children with hip dislocation in amyoplasia will increase the effectiveness of the treatment methods.

Spinal involvement in a pediatric and adult cohort of patients with Arthrogryposis multiplex Congenita.

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Background and Objective(s): Arthrogryposis multiplex congenita (AMC) is classified into 3 subgroups (Amyoplasia, distal arthrogryposis (DA), other causes) according to their phenotype and underlying aetiologies. Our main objective was to analyse the type of spinal involvement within each AMC subgroup. We hypothesized that there was a difference in the prevalence of scoliosis between subgroups. Study Participants & Setting: This was a single-center retrospective observational study with national recruitment from October 2007 to March 2022 at the AMC clinic of the University Hospital Grenoble Alpes (CHUGA). Participants underwent a clinical spinal assessment and spine radiography. Methods: We carried out comparative statistical analyses between AMC subgroups within two separate series: children and adults. Our outcome measures were the presence of scoliosis, Cobb angles, and underlying spinal abnormalities. Results: In total, 203 participants were selected. We performed a statistical analysis of 146 individuals (101 children, 45 adults). We found an overall prevalence of scoliosis of 46% in children and 78% in adults and an overall prevalence of underlying spinal anomalies of 41% in children and 53% in adults. There was a difference in the prevalence of scoliosis between the three AMC subgroups in children (X^2=6.8, p=0.033, v=0.18). The prevalence of scoliosis was higher in the DA group (47% in children and 78% in adults) and in the group “other causes” (65% in children) compared to the Amyoplasia group (33% in children and 75% in adults). Cobb angles were greater and spinal abnormalities more prevalent in the DA group than in Amyoplasia and “other causes” groups. Conclusions/Significance: Our data confirm the significant difference in the prevalence of scoliosis between AMC subgroups in children. The high prevalence of scoliosis and spinal abnormalities in all AMC subgroups justifies screening by clinical examination and systematic frontal and sagittal spine X-rays during childhood.

Content Co-Development of the Gross Motor Functional Classification for Children and Youth with Arthrogryposis Multiplex Congenita.

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Functioning of children with arthrogryposis multiplex congenita (AMC). The Gross Motor Function Classification System for children with cerebral palsy is a widely used tool. However, its application to children with other congenital/orthopedic conditions has not been validated. This study aims to develop a Gross Motor Functional Classification in AMC (GMFC-AMC) for children and youth. Study Participants & Setting. The design includes 3 phases with key stakeholders from geographically diverse regions: clinicians, researchers, patient representatives (individuals with or parents of children with AMC). Inclusion Criteria for phases 1-3: clinicians with at least 2 years of experience providing care to the AMC population; individuals with AMC above 18 years and/or parents of children with AMC. Exclusion Criteria: do not communicate in English, French or Spanish. Methods: Phase 1: A development team (n= 14; 11 clinicians, 2 adults with AMC, 1 parent of children with AMC) drafted the initial version of the GMFC-AMC over nine remote meetings. Phase 2 (May-June 2024) will comprise up to three nominal groups with stakeholders (n=15) from different geographical regions to reach final consensus on content. Phase 3 (July-September 2024): Delphi surveys will be conducted with participants (n=40 international experts in AMC) to achieve consensus on content and format (clarity, relevance) by rating each statement on a 7-point Likert scale (1=strong disagreement, 7=strong agreement). We will consider 80% agreement level in three rounds. Results. The initial content of the GMFC-AMC for 2-18 years encompasses four age groups: 2-4, 4-6, 6-12, and 12-18, across five performance levels in various settings (home, school, and community). In addition to classifying overall mobility, GMFC-AMC incorporates qualifiers related to upper extremity involvement. This nuanced approach recognizes the diverse ways in which upper extremity functioning may support gross mobility in AMC. Conclusions/Significance. Acknowledging the rarity of AMC, GMFC-AMC addresses the unique challenges individuals might experience, fostering tailored interventions and strategies. It has great potential to be culturally adapted and used across countries, supporting clinicians to provide care in this group of rare conditions.

Clinical case of arthrogryposis combined with osteogenesis imperfecta

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Background and objective (s). Bruck syndrome is a rare disorder that features osteogenesis imperfecta, combined with arthrogryposis, severe congenital joint contractures often with pterygia, short stature, severe limb deformities and progressive scoliosis. Its two forms, Brooke syndrome types 1 and 2, are similar in clinical manifestations with autosomalrecessive inheritance are caused by pathogenic variants of nucleotide sequence in the FKBP10 and PLOD2 genes, respectively. The purpose is to demonstrate the phenotypic characteristics, radiological and laboratory difference of sibling with Bruck syndrome type 2. Study Participants and setting: In sibling with Brucksyndrome type II the phenotypic characteristics, radiological and laboratory data were compared. The description of the given clinical observation expands doctors’ knowledge about the variability of clinical manifestations of Bruck syndrome and help with differential diagnosis of Arthrogryposis. This clinical observation is free from commercial bias and Safety. Methods: A 10-year-old boy and a 13-year-old girl - are siblings with Bruck syndrome type 2, born to healthy parents in a consanguineous marriage were observed. The boy had congenital flexion contractures of the knee and elbow joints, a small number of fractures, and severe kyphoscoliosis. The girl had no congenital joint contractures; she had kyphoscoliosis, more severe osteoporosis, and a history of more fractures than her younger brother. Results: The cases we present demonstrate significant phenotypic intrafamilial variability of Bruck syndrome type 2, caused by a homozygous variant in the PLOD2 gene, which consists of varying degrees of osteoporosis, as well as the presence and severity of contractures. Conclusions/ Significance: The description of the given clinical observation was made with the aim of drawing attention to a rare pathology and expanding doctors’ knowledge about the variability of clinical manifestations of Bruck syndrome. Genetic diagnostic is necessary for timely diagnosis of Brucks syndrome, determining the prognosis and developing patient management tactics.

Caregivers’ perspective on the impact of Arthrogryposis Multiplex Congenita (AMC): A global mixed method study

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Background: In pediatric healthcare, rare diseases impacting the musculoskeletal (MSK) systems pose substantial challenges for families caring for their affected children. Arthrogryposis Multiplex Congenita (AMC), a rare, congenital musculoskeletal condition, has a substantial global health impact, placing significant psychological, financial, and emotional impact on caregivers. This impact encompasses direct, indirect, and psychological challenges resulting from the complex caregiving needs of children with AMC. Caregivers also grapple with financial strain, job adjustments, strained relationships, and emotional stress. Therefore, our study sought to delineate the multifaceted costs associated with caring for children with AMC and to explore the caregiver’s caregiving experience. Methods: The global mixed-methods triangulation design employed both quantitative and qualitative data collection. The quantitative arm of the design, included a validated cost questionnaire, to gauge caregivers’ self-perceived health-related quality of life and that of their child and explore participants’ experiences as caregivers. The qualitative arm of the study entailed 60mins semi-structured interviews with a purposively selected group of caregivers of children with AMC. The quantitative phase involved the recruitment of 158 caregivers of children aged 0-21 years with AMC. Subsequently, 13 of these same participants consented to engage in the qualitative aspect, these interviews elucidated the multifaceted impacts of AMC, encompassing direct, indirect, and psychosocial costs. Data analysis included concurrent triangulation, integrating both quantitative and qualitative findings. Descriptive statistics were applied to the direct, indirect, and psychosocial costs. Linear regression modeling and logistic regression were used to identify potential associations between the cost factors and the impact of care as experienced by caregivers. Results & Conclusion: The cost of caring for a child with AMC was determined to vary depending on the age of the child and the type of insurance that the family can access. Themes that described the experience of caregivers were impact of caregiving experience; cost of childcare; support systems for care; managing and navigating care; and supporting the child’s growth and development. Alongside these themes, were recommendations to include the need for support groups and support for youth to prepare for adolescence. These findings will inform resource allocation, policymaking, and support services for children with rare conditions.

Genetic yield in prenatal detected Arthrogryposis Multiplex Congenita, a cohort between 2007-2021

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Background and Objective: Arthrogryposis Multiplex Congenita (AMC) presents challenges for prenatal detection due to its heterogeneous etiology (genetic and non-genetic), onset, and expression. Improved genetic testing over time enhanced knowledge on genetic underlying disorders . This study describes the phenotype and genotype of fetuses prenatally suspected for AMC. Study participants & setting: The study included suspected AMC cases from 2007 to 2021 through targeted anomaly scans and genetic investigations at the Amsterdam UMC, a tertiary center for Fetal Medicine in the Netherlands with an expertise center on Fetal Akinesia Deformation Sequence (FADS) and AMC. In 2019, prenatal Whole Exome Sequencing (WES) was introduced in our center. Methods: Results of the performed genetic tests were evaluated and the genetic yield was calculated over the three 5-year periods (2007-2011, 2012-2016 and 2017-2021). Results: The cohort consisted of 65 cases with AMC. Genetic testing was performed in all, prenatally in 83% (54/65), postnatally in 48% (31/65), of which combined pre-and postnatally tested in 31% (20/65). Performed tests were karyotyping (n=26), rapid aneuploidy testing (n=47) , single gene test (n=23), chromosomal microarray (n=42), targeted panel (n=18), WES (n=28), and Whole Genome Sequencing (n=1). Genetic (monogenic or chromosomal) abnormalities were identified in 18 cases (27.7%), of whom 9 were identified by prenatal testing alone, 2 by postnatal alone, and of the combined testing 3 in prenatal material and 4 in postnatal material. An overview of the prenatal phenotype and genotype will be presented in detail. Targeted panel and WES had the highest yields (33.3% and 28.6 %). A increase was found over the three 5-year periods resulting in 48% yield in 2017-2021. Conclusion: In this study, the perinatal genetic yield was about a quarter , of which two third was identified prenatally between 2007 and 2021. Increasing genetic yield over time and the additional value of targeted panel and WES emphasizes the advice to include these genetic tests within the set of tests parents are counseled for, dependent on a hospital's test availability.

Optimizing (pre-)pregnancy counselling for women with Arthrogryposis Multiplex Congenita (AMC): insights and recommendations from a questionnaire study

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Background and Objective: (Pre-) pregnancy counselling is not implemented as standard care for women with Arthrogryposis Multiplex Congenita (AMC) despite reported challenges in the limited available literature. Our aim is to optimize counselling and guidance of women with AMC during (pre-)pregnancy, childbirth and parenthood, by taking into account their needs. We try to add to the overarching theme of this symposium, "From the Womb to Adulthood". Study Participants & Setting: Women with AMC were invited for an online and anonymous questionnaire developed by one of the five participating patient support groups (the Netherlands, United Kingdom, Spain, United States of America) and Canadian adult AMC register. Methods: The survey covered six pregnancy-related themes. Results: In total 71 women responded to the questionnaire, of those 53 were included who finished the entire questionnaire. Pregnancies were reported in 64.2% (33/53). 26/53 (49.1%) women delivered 45 children. A third (33%, 15/45) of the children was born vaginally. All children were born healthy and without AMC. Nearly all (95.7%, 45/47) would (have) preferred a pre-pregnancy counselling. As optimal circumstances they prefer at or above 18 years of age, by a gynecologist, at the outpatient clinic and for those with a relationship together with their partner. The importance of multidisciplinary team of physicians was emphasized as well as the need of information from peer support groups and websites. Women's opinions expressed concern about lack of knowledge on AMC in healthcare workers, discontinuity in care and not always correct attitude towards treatment and acknowledged the importance of this study. Conclusions/Significance: Women with AMC provided insights in their needs and preferences on pre-pregnancy counselling and they paved the way to educate healthcare providers on specific challenges and opportunities of pregnancy and parenthood. Besides tailored multidisciplinary pre-pregnancy counselling, care during and after pregnancy, also peer support from AMC patient support groups and available knowledge on websites is advocated. We invite you to share your experience concerning pregnancy and AMC during the 4th International Symposium on Arthrogryposis in Montreal.

Possibilities and challenges in pregnancies of women affected by Arthrogryposis Multiplex Congenita: a literature review

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Background: The rarity of Arthrogryposis Multiplex Congenita (AMC) results in healthcare providers with limited exposure to possibilities and challenges in pregnancies of women with AMC. Women with AMC have little access to share information from peers that were pregnant or had child wish. Within the theme of this symposium "From the Womb to Adulthood" we present a literature review to obtain more knowledge on pregnancy-related topics in women with AMC to enhance knowledge among healthcare providers and women with AMC. Study participants: A literature search was conducted on documented cases of pregnant women with AMC. Methods: Available literature was bundled and summarized in three themes: 1. Outcomes (maternal, fetal and neonatal) in reported cases; 2. Impact of pregnancy on AMC and vice versa; 3. Advice on multidisciplinary approach to pregnancies with AMC. Results: The study includes 23 publications reporting 36 women with 74 pregnancies. Details on pregnancy-related outcomes could only be depicted from 19 women with AMC with in total 22 pregnancies and 23 born infants (19 live-born, 2 neonatal and 2 immature death). The type of AMC was Distal Arthrogryposis in 16 of the 19 (84%) women. Pregnancies resulted in 37% (7 /22) preterm births, 73% (16/22) cesarean sections, 18% (4/22) small for gestational age infants. Twelve infants had AMC. Pregnancy had limited negative influence on daily functioning or AMC during pregnancy except for two cases (2/19, 10.5%) with prior lung function impairment. Four times introduction issues of epidural analgesia was reported. Conclusions: In this literature review we reported that about a quarter of the women delivered vaginally, a fifth a small for gestational age infant, and a third preterm. Pre-pregnancy counselling and care by a multidisciplinary team is recommended to ensure optimal preparation for the care around pregnancy since the reported pregnancy complications. Future research should prospectively register the influence of AMC on pregnancy and vice versa. After the presentation, there will be a Question & Answer session with AMC experts. They will be available to answer the audience's questions on pregnancy-related topic.

The use of serial advanced sonographic examination for structural and motor analysis to enhance prenatal detection of contractures

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Introduction The prenatal detection of Arthrogryposis Multiplex Congenita (AMC) including Fetal Akinesia Deformation Sequence (FADS) is challenging due to its varying onset, etiology and expression. Amsterdam UMC is a tertiary referral center for Fetal Medicine in the Netherlands with an expertise center on FADS and AMC. We developed a perinatal care pathway with a stepwise approach to of examinations, counselling and care by a permanent multidisciplinary team for all fetuses with suspected contractures at the Standard Anomaly Scan at 20 weeks gestation. The targeted ultrasound examination includes serial advanced sonographic examination for structural and systematic motor assessment with an preferred interval of two weeks.

Study participants and setting Cases with prenatal suspected isolated contractures, AMC or FADS that were seen in our center during the last decade were selected.

Methods The sonographic findings including motor assessment, genetic testing results, course of pregnancy and postnatal findings of the cases were collected. During an interactive session we update the audience on the yield of systematic motor assessment in distinction between isolated contractures, AMC and its most severe and often lethal form FADS.

Results Three cases will be presented with prenatal suspected and postnatal confirmed contractures.

Data and images of sonographic findings will be demonstrated supplemented with videoclips of motor assessment. The three cases will represent different outcomes: normal motor assessment in isolated contracture, normal motor assessment except for reduced participation in the contracted joints in AMC, and deterioration of motor assessment in FADS.

Conclusion This overview will help you to understand what the additional value is of serial advanced sonographic examination for structural and systematic motor assessment in fetuses with contractures and how to distinguish within the spectrum of AMC milder forms and the most severe form , FADS.

Evaluation of Maternal Experience of fetal movements from a child with AMC (MECA survey)

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Background: Prenatal detection of Arthrogryposis Multiplex Congenita (AMC) is low due to the heterogeneous etiology, onset, and expression. This might also be due to not routinely offered serial monitoring in case of isolated contractures. Therefore, deterioration of the contractures will not be observed into AMC. Many healthcare providers are under the impression that the perceived fetal movements are always abnormal in pregnancies with AMC. It is unknown if these mothers' experienced movements influences the indication for repeated ultrasound examinations. By reaching out to AMC patient support groups and registers of various countries the knowledge will be further extended. The outcome could enhance the awareness to plan follow-up ultrasound examinations in isolated contractures. Study participants: Mothers of a children with AMC are invited for an online and anonymous questionnaire by the AMC patient support groups (the Netherlands, United States of America, Canada, Spain, United Kingdom, and Germany) and the Spanish and Canadian AMC patient registers. Methods: The questionnaire was developed in collaboration with the patient support groups and AMC registers. The survey covers questions on maternal experience of fetal movements, onset of the first movements, number and quality of the movements in pregnancies of children with AMC. The control population will consist of the same mothers

who also had a child without AMC. Women are invited to participate in the period from half of May- half of July 2024. Results: The results of the survey will be presented during the 4th International Symposium on Arthrogryposis in Montreal. The findings of this study will be compared with the prior studies. Conclusion: Collaboration with the AMC patient support groups and registers facilitate to obtain knowledge of women who have experienced movements of their child with AMC and whether their findings (normal experienced movements do not rule out AMC) were implemented to motivate healthcare workers for follow-up ultrasound examinations in case of contractures and normal experienced fetal movements by the mother. Obtained knowledge will enable to develop future guidelines on prenatal detection of AMC.

Expanding the spectrum of clinical and genetic characteristics of X-linked arthrogryposis multiplex congenita with central nervous system involvement (Wieacker-Wolff syndrome)

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Wieacker-Wolff syndrome (WRWF) is a rare X-linked arthrogryposis multiplex congenita with central nervous system involvement. The zinc finger C4H2-type containing (ZC4H2) gene located on chromosome Xq11.2 was confirmed to be the pathogenic gene underlying WRWF. Objective of our study is to describe the clinical and genetic characteristics of Russian patients with WRWF caused by pathogenic variants in the ZC4H2 gene.

We present genetic and clinical findings of 10 Russian patients (6 females and 4 males from the age 2 to 35 years old). In 8 families, cases had de novo status, as confirmed by Sanger sequencing. And one case was familial, where a boy inherited the pathogenic variant from his mother. Five of the eight ZC4H2 variants identified are novel: three missense variants, designated as c.227T>A (p.Met76Lys), c.131G>C (p.Arg44Pro) and p.Val108Ala, one splice site variant c.561+1G>A and one deletion c.77_79del (p.Lys26del). Three variants were previously reported (Brunet et al. 2021, Frinst et al 2019, May M. et 2015, Bertoli-Avella et al. 2021): a deletion with frame shift c.22_23delAT (p.Met8fs), one missense variant c.650A>G (p.Lys217Glu) and a splice site variant (c.225+5G>A). All male patients had predominant neurological symptoms as severe spectrum of mental, speech and motor development delay, spastic tetraparesis, strabismus, one of them had hemicrania with cyclic vomiting syndrome, another had pharmacoresistant epilepsy after an intraventricular haemorrhage at birth. Brain MRI showed a number of changes in the form of fronto-temporal atrophy, hydrocephalus, hypoplasia of the corpus callosum, MR signal enhancement from the conduction pathways. All female patients, compared with male, had severe multiple congenital joint contractures that needed orthopaedic treatment and less prominent neurological symptoms characterized by delayed psycho-speech development. A 35 years old patient carrying the c.225+5G>A variant had a mild phenotype with oculomotor apraxia, left facial nerve palsy and mild cognitive deficits.

Hence, we've presented five new pathogenic variants in the ZC4H2 gene. According to our findings all male patients in spite of the nucleotide sequence variant in the ZC4H2 gene had severe neurological symptoms and almost no congenital contractures, while in female patients the leading symptom was severe arthrogryposis multiplex congenita.

The treatment of foot deformity in patients with arthrogryposis, the first data of an italian pediatric hospital.

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Introduction: We present our data about arthrogryposis patients, collected since the start of the arthrogryposis consultancy started 2021. Methods: We report the results of 17 patients. Age: the youngest patient in treatment is actually 4 months old, the oldest 14 years old. Diagnosis: 10 patients with amyoplasia, 1 patient with Sheldon Hall, 6 Patient with distal arthrogryposis Genetic tests: all patient have done the genetic analysis, 10 patients have positiv results for gene mutations. Age of first treatment: from 2 weeks to 6 months. Number of operations (average value): 2,4. Follow up: all patients are in clinical follow up. Radiological follow up for the feet in 2 patients. Hip Problems:7. Knee Problems: 6. Other pathologies with need of surgical treatment: 2 patients with inguinal hernia. Treatment in newborn age: 14 patients undergone cast treatment and operation, 2 patient only cast treatment, 1 patient became only "tape" treatment. Early relapse: relapse in the first 6 months after treatment. Results: We divided the patients in 3 groups. 5 patients with early relapse become a second achilles tendon tenotomy. 8 patients with delayed relapse: 1 Patient become a second Dobbs Op with lenghtening of tibialis anterior, 1 patient with osteotomie and external fixateur treatment, 1 patient become correction with osteotomy, 1 patient had soft tissue release, 1 patient become achilles tendon Z- lenghtening, 1 patient is waiting for soft tissue release, 2 patients are only in treatment with custom made orthosis without new operation planned for the moment. 4 patients has no relapse. All the patients began the treatment after the cast with the Dennis Brown orthosis. 2 patients with early relapse changed to custom made orthosis. 3 patients with early relapse that continued the treatment with the DB orthosis had an other relapse in first 2 years of life. 15 patients are at the moment in treatment with custom made orthosis. Conclusion: Arthrogryposis is a rare condition, patients' s centralisation in a specialized children's hospital is important for a good and timely treatment. The orthosis are an important issue and can influence the results of postoperative treatment and relapse deformity.

Airway Team Concerns and Management of Patients with Arthrogryposis Multiplex Congenita. A Scoping Review.

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Introduction: Arthrogryposis multiplex congenita (AMC) is a heterogeneous disease that results in numerous non-progressive joint contractures, which can involve the head and neck area. This can lead to restricted neck movement, limited mouth opening, jaw, and palate anomalies. The unfavorable anatomy and often need for multiple corrective surgeries raise concerns about potential risks associated with a difficult airway and "cannot intubate, cannot ventilate" scenario. Objectives: Our study aims to assess the challenges encountered by airway teams and their management strategy for patients with AMC. Methods: A systematic search was conducted using eight databases (Medline, Embase, CINAHL, Cochrane, Global Health, Web of Science, Africa Wide Information, and Global Index Medicus) from inception to February 26, 2024. Of the 906 articles screened, 41 were included in our study. These articles comprised of 23 case reports, 7 case series, 6 correspondences, and 1 retrospective study. In all, 48 patients with arthrogryposis who underwent anesthesia intervention were identified. Results: Out of the 48 patients included, the most commonly reported abnormalities that could predispose to difficult airway scenarios were micrognathia (n=26, 54.2%), limited mouth opening (n=21, 43.8%), microstomia (n=17, 35.4%), restricted neck movement (n=13, 27.1%) and short neck (n=10, 20.8%). 20 patients (41.7%) were intubated with an endotracheal tube using direct or video laryngoscopy, 8 patients (16,7%) were managed using a flexible endoscope, 4 patients (8,3%) were intubated using a supraglottic airway (SGA) device, and 5 patients (20.4%) were intubated through an SGA assisted with a flexible endoscope. 7 patients (16.7%) were managed on spontaneous breathing due to the anticipated risk of encountering a difficult airway. 3 patients (6.3%) underwent surgical intervention (2 tracheostomy and 1 cricothyroidotomy) to secure the airway. Conclusion: Patients with AMC require careful planning and a case-based approach in airway management. The type of surgical intervention, anatomical challenges, team experience and available resources should be considered and weighed before the intervention to optimize patient safety.

Evaluation of Foot Osteotomies for Treating Residual Clubfoot Deformities in Patients with Arthrogryposis

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Introduction: Clubfoot is the most common deformity seen in patients born with arthrogryposis. The primary method of treatment for these deformities is serial casting, which has a greater frequency of incomplete correction and recurrence than in idiopathic clubfoot. For both primary and recurring cases, surgical treatments vary from soft tissue releases to talectomy. This research aims to evaluate the effectiveness of boney surgical procedures in correcting clubfoot in ambulatory children with arthrogryposis. Participants and Settings: A cross-sectional case-control study was conducted at a single center. All patients who underwent at least one foot osteotomy on their clubfoot, fixed using any method, and who had preoperative and postoperative ankle Passive Range of Motion (PROM), hindfoot-forefoot alignment (forefoot abduction), by physical examination, dynamic foot pressures (pedobarographic measurements), and Pediatric Outcomes Data Collection Instrument (PODCI) data were included in the current study. Absence of at least six months of postoperative follow-up and the presence of non-ambulatory status were criteria for exclusion from the study. Methods: This study is a retrospective case-control study. The normality of the data was assessed using the Shapiro-Wilk test. Pre-operative to post-operative comparisons of PROM, foot pressures, and PODCI scores were analyzed using the Wilcoxon Signed Rank Test. The frequency of subsequent bone surgeries following the initial operation was also investigated. Results: The study reviewed 49 surgical procedures performed on 24 feet of 20 children at age 15.1±4.5 years. Physical examinations revealed enhanced ankle dorsiflexion and forefoot abduction ($p<0.05$). Additionally, evaluating dynamic foot pressure heel contact, and the timing of heel rise demonstrated significant improvement ($p<0.05$). PODCI demonstrated improvement in happiness and global function ($p<0.05$). Two patients required an additional procedure at 26- and 37-months post-index surgery, because of the equinus deformity and insufficient correction of forefoot adduction, respectively. Conclusion: This research highlights the substantial role surgical procedures can play in improving, ankle range of motion, hindfoot-forefoot alignment, dynamic foot position and quality of life in children with arthrogryposis. It establishes a basis for future inquiries to delve into the longevity of these benefits and the overall outcomes.

Effectiveness of Achillotomy in Children With Arthrogryposis

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Introduction: Ponseti method is a wide-spread treatment option for clubfoot in children with arthrogryposis. Closed subcutaneous achillotomy in such patients does not guarantee complete correction of equinus deformity due to rigidity of tissues that often lead to reconsideration of the tenotomy principles. Aim: formulization of the anticipating criteria of effectiveness in order to develop the differentiated approach to achillotomy in children with arthrogryposis. Materials and methods. This research is based on retrospective analyze of closed subcutaneous achillotomy in 28 patients (56 feet) with arthrogryposis. Mean age of the patients at the moment of achillotomy was 5.4 months (range 2-8 months). The children were subdivided into 2 groups according to residual equinus deformity after completion of Ponseti serial casting. All patients were physically and radiographically examined. Results and discussion. The first group included 12 patients (24 feet), which achieved foot neutral position or dorsiflexion ≥5° after achillotomy. The second group consisted of 16 patients (32 feet) with residual equinus after achillotomy who required surgery. Comparison, based on X-ray, showed that patients of the second group had significantly wider tibiocalcaneal angle and significantly smaller talocalcaneal angle in lateral view ($p<0.01$). Correction value of the equinus deformity after achillotomy in children with arthrogryposis was greatly limited and averagely equal to 27° (20-30°) and 19° (10-30°) in the first and the second groups correspondingly. Conclusion. Closed subcutaneous achillotomy for effective equinus elimination during clubfoot treatment by Ponseti method should be performed only after complete correction at the level of tarsal joints. X-ray examination of the feet is recommended in children with arthrogryposis for more detailed evaluation of talocalcaneal divergence and heel position. Furthermore, the value of tibiocalcaneal and talocalcaneal angles in lateral view before achillotomy is essential prognostic factors of its effectiveness. Moreover, severity of equinus contracture should be considered before achillotomy. Achilles tenotomy is inappropriate if equinus deformity exceeds 30°. In such patients open surgery should be considered.

Technical Feasibility Study of Ultrasound Muscle Imaging in Antenatal Ultrasound Screening of arthrogryposis multiplex congenital.

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Background and Objective(s): By its congenital nature, arthrogryposis multiplex congenita has a high impact on musculoskeletal development. The functional prognosis and muscle atrophy are often severe after birth. Symptoms at routine ultrasound screening are either non-specific (amniotic fluid abnormalities or growth retardation), not very sensitive (joint deformities), or even difficult to detect due to time constraints (decreased fetal movements). Antenatal ultrasound screening is therefore not very effective for this group of pathologies, which are often particularly serious. The objective of our study is to evaluate the performance of muscle ultrasound sections on antenatal ultrasound between 21-24 weeks of gestation for the screening of muscle atrophy, in a sample of low-risk and high-risk pregnancies for arthrogryposis multiplex congenital. Study Participants & Setting: We will prospectively include 200 pregnant women at low risk for AMC, and 30 pregnant women at high risk of AMC (abnormal position of at least one joint; abnormal fetal movement perceived by the mother or revealed on ultrasound). Our study was approved by the French medical ethical committee and declared on clinicaltrials.gov (NCT06130592). Methods: This is a prospective non randomised multicentric study carried out in the Rhône-Alpes county/ France. The main criterion will be the success rate of realization of all four ultrasound sections at the extremities (deltoide, biceps, quadriceps, triceps) and measurements of the distances "skin - muscle fascia" and "muscular fascia - periosteum". Results: Inclusions have started during March 2024. We will present the first preliminary results at the International Symposium for AMC in Montréal/ Canada. Conclusions/Significance: The interest of the study is twofold: 1) to improve in the medium term antenatal screening for AMC in the general population, and subsequently possibly other pathologies with fetal hypo/akinesia without arthrogryposis and 2) to provide more precise advice in terms of causes and prognosis for couples, to be able to discuss a medical termination of pregnancy for the most serious forms.

Spectrum of deformities in children and adolescents with AMC treated in a single center - a pilot study.

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Background: Studies demonstrating a detailed spectrum of deformities and walking capabilities of children with different types of arthrogryposis (AMC) are rare. We asked about diagnosis; distribution and severity of contractures; function among children with AMC treated in a single center. This summary presents a preliminary report on the ongoing program results. Methods: Patients treated in a single center due to multiple contractures (AMC) were called in for clinical reevaluation. Included into the study were patients with AMC. The diagnosis was further divided into amyoplasia or non-amyoplasia types; syndromic cases (with other then skeletal involvement, especially CNS) were noted as well as those with known genetic diagnosis. Excluded were cases with uncertain clinical presentation and skeletal dysplasias. On examination the following parameters were noted: trunk deformities; joint range of motion and contractures were measured using a hand-held goniometer and graded using the Bartscale. Relationship between functional status, arthrogryposis type, contracture severity will be investigated further using statistical methods. Results: Up to date 76 children (45 boys, 21 girls) aged 7,4 y on average during the examination (range: 2 months - 18y) were assessed. The cohort consisted of 40 amyoplasia cases and 36 non-amyoplasia, including 12 with involvement of other systems (9 had CNS involvement and cognitive impairment), 5 with known distal arthrogryposis. 19 patients had no obvious diagnosis. Contractures of 4 limbs were noted in 40 children, of 3 limbs - in 3, of lower extremities only - in 7, and upper extremities only - in 7 patients. Severity of the contractures according to Bartscale: in upper limbs 1 extreme, 6 severe, 23 moderate and 38 mild. In lower limbs: 1 extreme, 11 severe, 28 moderate, 28 mild cases. This resulted in overall contractures severity: 42 mild, 24 moderate, 10 severe. Conclusions: Initial results demonstrate amyoplasia constitutes the most common type of AMC. Most patients had 4 limb involvements, however 17 cases had contractures limited to upper or lower limbs or had three limb involvement. Contractures according to severity were mostly mild and moderate. As the study goes on, final results may differ and will be presented after study conclusion.

Quantitative gait analysis of youth with Arthrogryposis Multiplex Congenita to improve individualized medical and surgical treatment.

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Background and Objective(s): Individuals with Arthrogryposis multiplex congenita (AMC) have two or more joint contractures, muscle weakness, pain and, presents with mobility limitations. Clinical assessments such as range of motion (ROM) and strength testing offer valuable information into the individual's condition but offers no information on dynamic movement patterns. This case series demonstrate how quantitative gait analysis can be used to help physicians determine optimal treatment for this population. Study Participants & Setting: Two patients (Female aged 11 and a male aged 13), with a diagnosis of AMC. Methods: Patients underwent a clinical evaluation (ROM and muscle strength) and quantitative gait analysis. Patients walked barefoot over ground at a self-selected speed. A motion capture system was used to collect three-dimensional kinematic data in three planes during the gait cycle, and later used to generate a gait evaluation report. Results: Patient 1 had a right Syme's amputation with decreased right knee flexion ROM (35°), limiting his mobility and sport participation. His main gait deviation was decreased knee flexion (peak flexion 38.3°; Norms: 56.8°) during swing phase leading to compensatory patterns to help clear the foot. Based on the clinical exam and gait analysis, the following treatments were recommended: a right knee orthosis with dynamic joint for night, continuous passive motion machine and intensive physiotherapy. Significant improvements were measured on right knee flexion after three months. No surgery was required for this patient at this time. Patient 2 had increased bilateral hip flexion contracture (Left: -20°, Right:-30°) and increased back pain. On gait evaluation, patient had multiple gait deviations, including an anterior pelvic tilt during stance phase (Left: 41.9°, Right 43.0°; norms: 12.0°). Her hip flexion contracture was further increased during stance phase (Left: 63.6°, Right 64.9°; norms: 32.9°). Based on the gait report, the surgeon's confirmed his surgical plan of performing bilateral psoas release. Conclusions/Significance: The integration of quantitative gait analysis into the clinical assessment of arthrogryposis allows significant benefits by providing quantitative data of dynamic joint impairments during gait. Physicians are able to acquire deeper insights into the complexities of AMC patients and offer optimal individualized patient centered care.

Spine deformities in children with different types of AMC

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Background: Frequency of spine involvement in AMC patients remains controversial. We asked about frequency and type of spinal involvement in different types of AMC in pediatric population. Methods: Patients treated in a single center due to multiple contractures (AMC) were called in for clinical reevaluation. Included into the study were patients with AMC with diagnosed amyoplasia or non- amyoplasia types i.e. syndromic cases (with other than skeletal involvements, especially OUN) or distal arthrogryposis. Excluded were cases with uncertain clinical presentation and skeletal dysplasias. Patients underwent spine clinical examination and classic radiographic assessments including standing, sitting or lying spine AP and lateral views, depending on the patient status. We looked for trunk asymmetry, imbalance in coronal and sagittal views both clinically and radiographically. Results: Up to date 76 children (45 boys, 21 girls) aged 7,4 y on average during the examination (range: 2 months - 18y) were assessed. The cohort consisted of 40 amyoplasia cases and 36 non-amyoplasia, including 12 with involvement of other systems (9 had CNS involvement and cognitive impairment), 5 with known distal arthrogryposis. 19 patients had no obvious diagnosis. Spine deformities were noted in 53 of children, by means of clinical and/or radiographic assessment. 15 patients had a Cobb angle less than 10°. Mild scoliosis (11-20°) were noted in 15, moderate (21-40°) in 7 and severe (> 40°) in 4. Increased lumbar lordosis was detected by clinical means in 11 patients and coronal deformity was detected in 27. In 20 patients' clinical examination did not reveal the deformity that was later diagnosed in radiographs, including 8 cases of scoliosis (Cobb angle > 10 degrees). Conclusion: The study is ongoing, so definite findings will be presented on its conclusion. Up to date, the rate of spinal deformities ranges 70%, with increased lumbar lordosis rate 14,5%, scoliosis 40,1%. Clinical examination may not reveal all spinal deformities, including scoliosis in up to 15%.

Walking abilities, function and quality of life in children with different types and severity of arthrogryposis - a preliminary report from a single center.

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Background: Studies on function in pediatric patients with AMC are rare. We investigated walking abilities, overall function and QoL of patients treated in a single specialistic center. Methods: Patients with multiple contracures (AMC) treated in a single center were called in for clinical examination. Included into the study were patients diagnosed with amyoplasia and non-amyoplasia types i.e. syndromic cases (with other than skeletal involvements, especially CNS) or genetically diagnosed distal arthrogryposis. Excluded were cases with uncertain clinical presentation and skeletal dysplasias. We used a modified Functional Mobility Scale in children minimum 4 years of age to assess their walking ability. The patients were allowed to use necessary orthoses - the need to use crutches, a walker, difficulties climbing stairs or the need for assistance from another person were noted. For functional assessment we used PODCI questionnaire filled out by caregivers or the patients (older than 12 y). Results: Up to date 76 children (45 boys, 21 girls) aged 7,4 y on average during the examination (range: 2 months - 18y) were assessed. The cohort consisted of 40 amyoplasia cases and 36 non-amyoplasia, including 12 with involvement of other systems (9 had CNS involvement and cognitive impairment), 5 with known distal arthrogryposis. 19 patients had no obvious diagnosis. Walking abilities according to FMS were as follows: 18 had level 6, 24 - level 5, 4 children were graded from 2-4. 15 children were non-functional walkers or non-walkers, having FMS 1. Most of them had severe contractures of the lower limbs and/or involvement of CNS. PODCI results demonstrated mean values for the following domains: Upper Extremity - 62; Transfer - 59; Sport and Activity - 45; Pain - 75; Happiness -was 65; the overall Global Functioning - 61. Conclusion: The study is ongoing, thefore the conclusions will be presented after it ends. Patients with AMC frequently achieve walking abilities when using orthosis. When walking abilities are disturbed it is usually due to severe contractures of lower extremities and/or to cognitive impairment. Mean values of PODCI domains demonstrate positive results, indicating good life quality despite deformities. Sport and Activities were affected. There was a tendency of increasing pain complaints with age.

Open hip reduction in children with multiple contractures - early results

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Background: Teratogenic hip dislocation remains a frequent entity among children with AMC. Reduction improves hip function, but requires surgical treatment in AMC cases. Open reduction seems a liable option, however still controversy exists regarding optimal timing, approach, postoperative treatment and final results. We present early observations in children with different types of AMC treated by open reduction for teratogenic hip dislocation. Methods: Retrospective analysis of early complications including reluxation, need for additional interventions, clinical and radiographic results including late reluxation, dysplasia, femoral head growth disturbance (AVN). Results: 37 children underwent open hip reduction at our institution between 2014 - 2024. Excluded from the study were children with less than 12 months of follow up (2), those who were not diagnosed with arthrogryposis (3) or treated with an approach other than medial. The studied cohort consisted of 31 children (15 boys, 16 girls) with 50 hips. There were 19 bilateral cases (38 hips), 5 right, 6 left. All children underwent open reduction using medial approach according to Weinstein - Mau. 8 femoral shortening osteotomies were performed additionally to hip reduction. Other concomitant interventions included clubfoot correction (7 children), vertical Talus correction (4 children), knee (1) and hip (1) soft tissue releases. There were 4 cases of early reluxation which required an additional procedure. One child required soft tissue releases due to adduction contracture after immobilization. Late complications including proximal femur growth disturbance (AVN), late reluxation are under investigation. Most children present with satisfactory, painless range of motion, regardless of radiographic results Conclusions: As the study goes on we can not derive any definite conclusion. It seems that it is an efficient surgery in skilled hands, providing good clinical results, however the risk of complications remains high.

Mutation in the GLDN gene - is it always a death sentence? A case report.

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Background and objectives: Lethal congenital contracture syndrome (LCCS 11) is a very rare disease caused by a mutation in the GLDN gene which encodes the gliomedin molecule, which plays a role in the development of the nodes of Ranvier. Children with this mutation are born with arthrogryposis and severe lung hypoplasia, which in the past was the main reason for mortality in the neonatal period. The prevalence of this mutation is unknown and there is very little information available in medical literature. Therefore analysis of each patient with this diagnosis can contribute to gaining more knowledge concerning the prognosis and appropriate treatment. Method: This paper is based on a clinical observation of a 3 year old patient with LCCS 11. The patient was referred to the Arthrogryposis Treatment Center at Cracow Children's University Hospital at the age of 5 months. A treatment plan based on serial casting, rehabilitation and orthopedic surgical procedures was discussed with the parents and implemented. At the time of final follow up the patient was walking independently in ankle-foot-orthoses, with normal cognitive development and good lung function. Conclusion: Despite LCCS 11 being believed to be a lethal condition, it is important to realize that with a prenatal diagnosis (as it was with our patient) the mother can give birth in a reference center where proper care is provided for lung hypoplasia. This allows the patient to survive the neonatal period. It is important not to stigmatize patients with this type of arthrogryposis, just because the name of the disease has the word 'lethal' in it and to deny them the appropriate treatment for deformities related to the arthrogryposis.

French consensus on an Emergency card for AMC.

Klaus Dieterich[1, 2], Kelly Dilworth, Emeline Bourgeois, Véronique Bourg, Véronique Thellier, Claire Huzar, Gipsy Billy, Marjolaine Gauthier, Manon Bachy, Hassan Al Khoury, Judith Melki, Jacques Griffet, Marieke Chamberon, Marc Roquebert, Michèle Dutac, Adeline Loguiot, Pascale Boguais, Marie-Claude Lesne, Jérémy Guenezan, Guillaume Debarty,

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Background and Objective(s): People with AMC have specific needs in case of emergency. Some are only applicable to specific etiologies; others are systematic consequences of the lack of fetal hypo-/ akinesia. Study Participants & Setting: As a response to an incentive from the French rare disease consortium, we gathered a multidisciplinary panel of experts in the fields of emergency medicine and in AMC coming from nine distinct hospitals all over France. Panelists were respectively emergency doctors (n = 2, including a representative of the French National Society for Emergency Medicine), anesthesiologists (n = 1), orthopedics (n = 4), physiatrists (n = 3), occupational therapists (n = 1), physiotherapists (n = 1), research assistants (n = 1), genetic counselors (n = 1), and geneticists (n = 2). Five people with lived experience of AMC were representatives of the French Patient Association Organisation (Association Alliance Arthrogrypose). Methods: Panelists and people with lived experience met during the Scientific Counsel of the French PAO in a hybrid meeting on June 16, 2023. Results: The cover page mentions the identity, date of birth and anticipated directives in case of an unfavorable prognosis. Difficult

venous access and intubation, and avoiding over-exposure to and increasing the risk of latex allergies are also highlighted. Extra-skeletal anomalies associated with AMC can be filled out on the inside page: cardiac, renal, respiratory, neuropathic, myopathic, cerebral anomalies, communication difficulties, and miscellaneous, as well as the precise clinical and/ or molecular diagnosis if known. Particular features anticipating difficulties for anesthetic or resuscitation procedures are also mentioned: previous difficult intubation, obstructive apnea, identified risk for malignant hyperthermia, scoliosis, gastric reflux, chronic pain, current medical treatment. A figure scheme helps describe persistent deformations that should be respected in case of surgical interventions. Major previous surgical interventions should be listed. Care center(s) and health care professionals are mentioned at the back side. Conclusions/Significance: The goal of this emergency card is to improve the interaction between health care professionals and the individual with AMC and the management in case of emergency.

Long-term follow-up and outcome in autosomal dominant Larsen syndrome.

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Background and Objective(s): Larsen syndrome is an autosomal dominant syndrome characterized by joint dislocations, typical dysmorphic facies, abnormality of digits, spine and foot malformations and other skeletal defects linked to FLNB missense mutations. Clinical reports are still limited and long-term follow-up and functional outcomes scarce. Here we report on two adult patients of a single family, a mother and her daughter, addressed for multidisciplinary evaluation at our institution. The clinical diagnosis of Larsen syndrome was established early during follow-up. Study Participants & Setting: We report clinical and management data from birth to adulthood for both patients, obtaining a long-term hindsight that provides an overview of the disease course, covering several decades. Methods: This was a retrospective observational case study. Data were obtained from the PARART registry (NCT05673265). Results: Both adults presented with typical features of the syndrome, but also symptoms that had been more rarely reported such as conductive deafness. Their clinical presentation illustrates intra-familial variability, the mother having a more severe form in terms of musculoskeletal involvement. Complete functional assessment evidenced favorable outcomes even for the patient with a more severe presentation. In addition, we expose the added value of pluridisciplinary specialized evaluations in terms of medical management for patients suffering from rare diseases. Finally, we performed whole exome sequencing. We identified a novel mutation in FLNB occurring in the classical region involved in Larsen syndrome. Conclusions/Significance: Despite potential severe musculoskeletal involvement at birth, functional outcome in individuals with autosomal dominant Larsen syndrome remains good.

Developing a community and evidence based patient education program for arthrogryposis multiplex congenita: “La vie en mouvement”.

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Background and Objective(s): Patients with arthrogryposis benefit from intensive rehabilitation and orthopedic care from birth. Families are therefore confronted very early and very regularly with the management of their illness. Identifying specific needs in therapeutic education was therefore not obvious at first glance in this group of pathologies. Study Participants & Setting: At first, we asked one single open question: children and adults with lived experience of AMC, and their parents or partners, at the annual meeting of the French patient association organization Alliance Arthrogrypose in 2018 what their main difficulties were in daily living. We identified the most frequently occurring topics. We then developed workshops responding to the aforementioned topics in order to build a Patient Education Program. We established workshops for different age groups - toddlers, infants, adolescences, and adults. Methods: Community-based approach following a patient and parent questionnaire to identify their needs and knowledge gaps. Results: Thirty-one individuals answered the questionnaire. Among them, 77% (n = 24) mentioned as one of the main difficulties the lack of medical knowledge about AMC or its medical care. For another 35% (n = 11), pain and fatigue were identified in equal proportions. The psychological burden through difficulties to accept the illness or the social consequences of their physical appearance was mentioned by 26% (n = 8). How to get old with AMC was a concern for 13% (n = 4). We therefore built four workshops for adults: 1) How to handle to speak about my AMC, 2) Pain, 3) Fatigue, and 4) Aging. For children, we built workshops on 1) How to speak about my AMC, 2) How to move my body, 3) I sleep with my orthoses, 4) Well off, I do things on my own. Forty-eight individuals have benefitted from our Patient Education Program since 2020. Among those, 40 were adults and 8 were children (5 toddlers, 3 infants). Conclusions/Significance: Our Patient Education Program has helped us dedicate significantly more time to every individual. The very different setting compared to traditional consultations enhances discussions on topics less frequently treated and helps exchange on individual expertise or acquired strategies.

Apical Ectodermal Ridge Failure in Amyoplasia

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The cause of Amyoplasia remains controversial, with the predominant theory being loss of anterior horn cells as the primary insult. Failure of the apical ectodermal ridge (AER) is an extremely rare condition occurring in 1 out of 2 million live births. Amyoplasia has an incidence of 1 in 10,000 births, making the probability of both conditions occurring simultaneously by chance 1 out of 20 billion. The AER develops after the limb bud begins to form, and has a vital role in limb growth and development. Muscle precursor cells do not differentiate until they reach the limb and contact signaling factors that are AER dependent. Without sufficient AER function, muscle cell line development may be limited. We report on an association between failure of the AER and amyoplasia, which may offer a clue regarding pathogenesis. We identified 20 patients in our practice with AER failure. Loss of fingers or fingertips was seen in 25% of cases, with loss of toes in 37%, fingers and toes in 31%, and loss of a foot in 6%. No patient had ectodermal elements at the transverse amputation or amnionic bands, confirming that in no case was the deficiency due to symbrachydactyly or Streeter’s dysplasia. We have identified no patients with distal arthrogryposis that are missing ectodermal elements.

Distal Carpal Wedge Osteotomy with Suture Fixation in Patients with Arthrogryposis

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Hypothesis: Distal carpal wedge excision with suture-only fixation is an effective method of treating wrist flexion deformities in children with arthrogryposis that preserves motion in the mid-carpal joint and avoids the complications associated with Kirschner wire fixation. Methods: Our technique for carpal wedge excision places the osteotomies distal to the mid-carpal joint. Fixation is achieved with polydioxanone suture. We performed a retrospective review of all patients with arthrogryposis who underwent carpal wedge osteotomy for wrist flexion deformities from 2015 to 2021. Pre and post-operative active wrist range of motion were collected along with patient demographics, Patient-Reported Outcomes Measurement Information System (PROMIS) pediatric upper extremity short form scores, and complications. Results: Forty-two distal carpal wedge osteotomies were performed in 37 patients. Patients were followed for an average of 23.8 months (range 3 to 67 months) after surgery. Pre-operatively, active wrist range of motion was 67.3° of flexion to -26.3° of extension. Post-operatively, active wrist range of motion was 31.1° of flexion to 9.4° of extension. All patients achieved union following suture fixation, and no complications were noted. The average PROMIS t-score at last follow up was 27.3 ± 11.3 (32 patients). A subset of 7 patients completed both a pre and post-operative PROMIS questionnaire and had an average improvement in t-score of 5.7. Summary Points: A distally placed carpal wedge osteotomy is an effective technique of addressing fixed wrist flexion deformities in patients with arthrogryposis. By preserving motion in the mid-carpal joint, we were able to achieve active motion in the wrist that compares favorably to previously published series, while avoiding complications associated with Kirschner wire fixation.

Complications of Single vs. Dual Plating in Humeral Rotational Osteotomies

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Humeral rotational osteotomy (HRO) has become a mainstay of treatment for children and adults with arthrogryposis to improve shoulder rotation position. Children with internal rotation contractures of the shoulder are forced to procure objects with a crossover grasp pattern, made worse with pronation contractures. Hand function is compromised due to inability to see the thumb during reach and grasp. Hand to mouth function is also compromised. Although HRO has been shown to improve function, long-term difficulties include peri-implant fractures due to an increased fall risk and recurrence of internal rotation due to skeletal remodeling. Fixation therefore needs to be rigid enough to resist a fall during the initial healing phase and compliant enough to not increase fracture risk. We retrospectively reviewed our experience with single plates and dual plates to compare the rates of peri-implant fracture in children with amyoplasia. All patients with single plates and all patients with dual plates were consecutive, with a clear time point where we converted to dual plates. Single plates were placed via both a medial and a lateral approach, whereas the dual plates were placed via a lateral approach. The lateral approach was adopted to limit the risk of re-operation in case of revision or fracture. Dual plating resulted in lower rates of fracture, with lower risk of hardware failure in the initial period. Intraoperative complications were similar between groups. Dual plating with undersized plates provides advantages over traditional single plating both in fracture risk and failure of fixation.

Best Practices of Patient Support Groups - I. Organization of patient support

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Introduction: Coordinators of the patient support groups of USA, Spain and the Netherlands are working together to determine how international collaboration with and between patient support group organisations can lead to a stronger international AMC network. This is also the subject of a workshop during the presymposium meeting. During this workshop the outcomes of a survey among coordinators of patient support groups in various

countries around the world, will be discussed, together with the experiences and questions of the participants of the workshop, concerning three themes:

1. Organisation: learning and challenges ins building and sustaining a strong support organization and network.
2. Pride and concern: what are the best practices the patient support group is proud of, and which challenges do the different patient support groups face?
3. National and international future plans: what are the most important developments and plans to strengthen patient support in the future?

Each theme will be shared during the 4th Interntional Artrhogryposis Symposiumto to a broader audience for further suggestions and more lessons to be learned. Methods: A digitized survey among patient Support groups worldwide is performed in may-july 2024. The summary of the lessons learned during the presymposium workshop will be elucidated. Results: Key topics of the organization theme will be presented:

- What are the different organizational structures for patient support groups? Do people need to be a member. Are coordinators (full time) professionals, or volunteers?
- What are the aims of the organisation?
- Accessibility: How do you reach and actívate patients (and parents)? What are successful platforms for peer support from birth into adulthood?
- How can patient support groups collaborate with medical professionals and researchers to build AMC expertise centers and strengthen multidisciplinairy care for (all) patients?

Conclusion: The possibility to share best practices of the various patient support groups among each other and with medical professionals will strengthen national and international organisations and collaboration. Plans will be discussed how to maintain this (inter)national effort to support existing and new patient support groups.

Acute knee flexion contracture correction in patients with arthrogryposis and failed distal femoral osteotomies

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Background and Objective(s): Patients with Arthrogryposis Multiplex Congenita (AMC) often present with knee flexion contractures in childhood. Distal femoral osteotomies have been reported to be successful in the cerebral palsy community in adolescents. Application of this surgery in young, school-age children, as especially seen in AMC, often recur and leave a residual deformity. Acute correction of knee contractures with a posterior knee release and femoral shortening can improve mobility in these patients but is not without risks. Study Participants & Setting: 6 patients with AMC (2 female, 4 male) had 10 knee flexion contractures treated with an acute knee flexion contracture correction in the setting of a failed distal femoral osteotomy between 2021 and 2023 at an elective orthopedic pediatric hospital. Methods: This is a retrospective study of patients with AMC who underwent acute knee flexion contracture correction in the setting of failed distal femoral osteotomy from 2021 to 2023. Demographics, pre- and post-op clinical range of motion, measurements of radiographic extension deformity and flexion/extension contracture limitations were included as available. Ambulatory status and complications were noted. Results: The average age of prior extension osteotomy was 5.8 years and 10.1 years at re-operation. Pre-operative sagittal deformity of the distal femur averaged 26.9 degrees and a radiographic extension contracture of the distal segment of 75.8 degrees sustained an average correction to 4.7 degrees radiographically. Clinically, contractures improved from an average of 40.5 to 14.1 degrees. Five complications were noted that included a physeal fracture, screw loosening, wound dehiscence, vascular injury, and heel pressure-sore. Improved gait was seen in all. Conclusions/Significance: Distal femoral extension osteotomies should not be used in children with significant growth remaining for flexion contractures. Early results suggest that in AMC, children with failed distal femoral osteotomies and recurrent severe flexion contractures may be successful treated with an acute correction using posterior knee release and femoral shortening. All patients had improvement in clinical extension and comfort in bracing that permitted improvement in ambulation as early as 4 months post-operatively.

Open Reduction of Arthrogryposis Hip Dislocations is Associated with Higher PFGD Rates Compared to DDH: A Dual-Center Study

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Background: Sequelae of open reduction of developmental/syndromic hip dislocations may include avascular necrosis (AVN)/proximal femoral growth disturbance (PFGD) and residual dysplasia. There is limited information comparing rates of these sequelae in Developmental Dysplasia of the Hip (DDH) and Arthrogryposis multiplex congenita (AMC). We performed a dual-center retrospective cohort study to compare rates of AVN/PGFD and residual dysplasia after open hip reduction between DDH and AMC for non-traumatic hip dislocations. Methods: We identified patients <18 years of age who underwent open reduction of hip dislocations between 1981-2020 at two large tertiary pediatric hospitals. Patients with AMC were matched by age against patients with DDH 1:2. Preoperative data included demographics and severity of dislocation by International Hip Dysplasia Institute (IHDI) classification and acetabular index (AI). Outcomes included AI at 2 years postop, IHDI at final follow up, presence/grade of AVN/PFGD by Salter Criteria at 2 years postop, and by Kalamachi and Macewen (KM) classification at final follow-up. Significance was set at P<0.05. Results: 82 patients (98 hips) with DDH were matched against 39 patients (49 hips) with AMC with mean follow-up of 97 months (range:24-443). There was no difference in mean age at surgery (1.46 vs 1.43 years; P=0.86), preoperative IHDI, AI, or spica cast duration (all p>0.05), but the DDH cohort had more females (83% vs 56%; P=0.003). Intraoperatively, the DDH cohort underwent more anterior approaches (75% vs 55%; P=0.02), and pelvic osteotomies (34% vs 14%, P=0.01). Postoperatively, the incidence of AVN/PFGD was higher in AMC than DDH at 2 years by Salter Criteria (57% vs 21%, p<0.001) and at final follow up by KM criteria (59% vs 16%, p<0.001). At 2 years postop, there was no difference in AI (31° vs 29°, p=0.3) or reoperation rate (24% DDH vs 20%, p=0.7), but the AMC cohort had more IHDI grade 2-4 hips than DDH (12% vs 9%, P=0.02), reflecting re-subluxation/dislocation. Conclusion: Open reduction for hip dysplasia in AMC had a significantly higher rate of AVN/PFGD and re-subluxation/dislocation compared to DDH despite similar preoperative characteristics. This information can assist in guiding perioperative counseling for families of patients with AMC.

Spectrum of orthotic treatment for AMC patients in a specialized center

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Spectrum of orthotic treatment for AMC patients in a specialized center

Background: Custom made orthotic devices constitute an important part of complementary treatment for patients with different types of arthrogryposis - from infants to teenagers and adults. The general aims of orthotic treatment includes deformity correction, increase in joint range of motion, maintaining and improving post-operative correction. Methods: We reviewed retrospectively the files of patients treated with orthotics at our Center in 2023 according to indications, type of orthotics, body area, number of provided orthotic devices generally and per patient. Results: During 2023 there were 128 AMC patients in our orthotic clinic. We provided 409 orthotic devices generally. There were 223 AFOs, 110 KAFOs, 48 WHOs, 8 body jackets, 4 EWHOs, 2 HKAFOs. Mean number of orthotics per patients was 4 per year. Mean number of exchanging orthotics was 2 per year. Conclusions: The most frequently treated parts of the body are: the foot and ankle joint, knee joint and wrist joints. Individually tailored orthotic treatment remains golden standard as part of complex treatment for limb and trunk deformities in arthrogrypotic patients .

An interdisciplinary approach integrating the use of orthotics to improve children's day-to-day function.

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Patients with arthrogryposis show significant and variable contractures that bring important challenges in both mobility and function in daily activities. The use of orthotic devices is essential to maintain and increase their musculosqueletal integrity and quality of life. Our presentation analyses how different types of orthosis can be an effective solution for improving upper and lower limb function in activities of daily living, We will highlight the types of orthotics that can be used and adapted to meet specific client needs. Through the examination of case studies, evidence based litterature and patient's review, our comprehensive analysis will highlight the efficacy and limitations of diverse orthotic interventions on musculoskeletal integrity and daily functional activities. These findings will shed light on the key factors influencing the selection of orthotic devices based on the clinical presentation of the patient. Opportunities for innovation will be discussed, aiming to inspire healthcare professionals and researchers to explore further the potential of orthotics in improving functionality and quality of life for patients with arthrogryposis.

POSTER PRESENTATIONS

Pain and Function in Adults with Arthrogryposis Multiplex Congenita

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Introduction: Arthrogryposis multiplex congenita (AMC), a rare congenital disorder characterized by joint contractures and muscle weakness, often results in pain, which may negatively impact health. The purpose of this cross-sectional research study was to evaluate how the pain experience relates to physical and mental health in AMC. Participants: Ambulatory individuals with AMC, aged 18 years and older, were recruited from May to August of 2022. Outcome Measures: Questionnaires were administered via RedCap. Pain experience was evaluated using 7-day average pain intensity per the Patient-Reported Outcome Measurement Information System (PROMIS), pain severity per the McGill Pain Questionnaire, and pain catastrophizing per the Pain Catastrophizing Scale. Health was assessed with the Patient-Reported Outcomes Measurement Information System (PROMIS) Global Physical and Mental Health measures. Data Analysis: Descriptive statistics were calculated using SPSS statistical software. Linear regression models evaluated associations between pain experience and health outcomes, while considering upper- and lower-extremity involvement as covariates. Results: The sample included 73 participants with a median age of 31 years; 67.1% were female and 87.7% were Caucasian. After considering the extent of extremity involvement, pain intensity explained 4.2% of the variance in physical ($p<0.001$) and 5.8% of the variance in mental health ($p<0.001$), pain severity explained 10.4% of the variance in physical ($p<0.001$) and 3.0% of the variance in mental health ($p<0.037$), and pain catastrophizing explained 2.4% of the variance in physical ($p=0.003$) and 8.7% of the variance in mental health ($p<0.001$). Discussion: Pain intensity, severity and catastrophizing are significantly related to physical and mental health among individuals with AMC. Of note, pain severity appears more impactful to physical health, and pain catastrophizing to mental health. Clinical Relevance: Pain intensity, severity, and catastrophizing may be clinical targets for health promotion in adults with AMC.

Beyond Borders: The First International Dataset to Advance arthrogryposis multiplex congenita (AMC) Knowledge

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Background: Arthrogryposis multiplex congenita (AMC) represents a heterogeneous group of rare congenital contractures in two or more body joints. The development of common data elements (CDEs) has become a priority in AMC research to support etiological, epidemiological, and comprehensive phenotyping investigations. This study aimed to define a set of consensus-based standardized CDEs to enable an international registry for AMC. Methods: This is a sequential exploratory mixed-methods study among AMC experts including clinical specialists, researchers, and adults with lived experience/AMC representatives. Comprising of three consecutive phases, phase 1 aimed to identify a candidate set of data elements using focus group discussions (FGDs), phase 2 used three rounds of modified Delphi survey to achieve consensus on CDEs, and classify the CDEs, and phase 3 used FGDs to describe recommendations for implementation of the CDEs. Results: Two FGDs identified emerging themes for AMC dataset including environmental factors, cost of care, family history of AMC, and lifetime rehabilitation utilization from gestation to adulthood. In total, 45 panelists from 11 countries in North America, Europe, and Australia participated in Delphi surveys. Participation was highest for occupational therapists/physical therapists, orthopedists, medical geneticists, and adults with lived experience/AMC representatives. The CDEs included 321 data elements, starting from prenatal detection and care, to diagnoses, clinical manifestations, care and management, and socioeconomic factors from childhood to adulthood and 19 standardized measures. The data elements were 44 (14%) General core, 134 (42%) Disease core, 113 (35%) Supplemental-Highly recommended, and 30 (9%) Exploratory. To advance an international registry for AMC using the CDEs, a universal governance structure, partner-specific operating protocols, and long-term sustainability plans were the main facilitators; inconsistent databases, incompatible electronic infrastructures, and feasibility concerns were the main barriers. Conclusion: The unified, standardized data framework of CDEs facilitates multidisciplinary collaboration to augment statistical power for epidemiological and genetic studies, particularly as AMC and associated diagnoses are rare. The systematic data using the CDEs framework prompts large-scale investigations on etiological pathways, describe epidemiological profile, and establish genotype-phenotype correlations. The proposed CDEs will facilitate international multidisciplinary collaborations and opportunities for data sharing, knowledge translation, and dissemination.

Expediting knowledge using standardized annotations: Application of Human Phenotype Ontology for arthrogryposis multiplex congenita

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Background: Arthrogryposis multiplex congenita (AMC) represents a large, rare group of congenital musculoskeletal conditions, with major challenges in research posed by incompatible data frameworks, broad use of inconsistent terminologies, and text descriptions. Using the international dataset for AMC, our study aimed to systematically describe AMC phenotypic traits using Human Phenotype Ontology (HPO) terms, encode the phenotypic traits as HPO terms, and pilot test the encoding process. Methods: An international consensus-based dataset for AMC was used to extract the main AMC phenotypic traits. Systematic mapping of phenotypic traits to HPO ontology (v2022-06-12) was performed using a pre-tested algorithm. The encoding process addressed standardized annotations for joint contractures using a 3-step process. To compare the encoding process performance for systematic mapping of joint contractures to HPO terms, we extracted clinical summaries of 80 individuals with AMC from the North American AMC registry database. Results: Using a pre-tested mapping algorithm, the HPO mapping process resulted in 62% complete match, 12% incomplete match, and 26% no match. The 3-step process resulted in the extraction and review of 19 articles and book chapters (range: 2- 8 reference per joint contracture) that provided 50 suggestions, comprising of 37 (74%) new terms and (re)annotations, and 13 (26%) re-structures for joint contractures' ontology in 10 joints including Shoulder, Elbow, Wrist, Hand, Thumb, Fingers, Hip, Knee, Ankle, and Toe. The implemented annotations significantly increased the number of available HPO terms for joint contractures in a cohort of children with AMC ($p =0.04$). Our encoding and annotation approach may be used as a blueprint for systematic HPO (re)annotations for musculoskeletal and non-musculoskeletal phenotypic traits of AMC. Conclusion: Our study used an international AMC dataset to systematically map the phenotypic traits and to encode HPO ontology. Use of real-world data from the AMC registry ensured representativeness of the findings for AMC community. Our encoding and annotation approach exemplified application of a standardized knowledge base with significant improvement in phenotypic-driven classification in rare diseases. Our approach may be used as a blueprint for systematic HPO (re)annotations for musculoskeletal and non-musculoskeletal phenotypic traits of AMC and other types of rare diseases.

Health-Related quality of life in AMC: Findings of the North American registry on children with AMC

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Background: Health-related quality of life (HRQL) may be reduced in arthrogryposis multiplex congenita (AMC) due to broadly heterogeneous physical impairments and lifelong care. This study described HRQL in children and youth with AMC, compared HRQL between child self- and parent-proxy reports, and identified factors associated with better/worse HRQL. Methods: Data on 205 children with AMC (age 0-21 years) from a North American AMC registry across eight hospital sites was used. HRQL was assessed using Patient Reported Outcome Measurement Information System (PROMIS) domains of Mobility, Upper Extremity (UE), Peer Relationships and Pain Interference, and European Quality of Life-5 Dimensions-Youth- 3 Levels (EQ-5D-Y-3L) by self-report, parent proxy-report or both. Results: The sample included 41% (n=84) Amyoplasia, 28% (n=54) DA, and 14% (n=32) CNS/syndromic AMC. Overall mean (SD) PROMIS T-scores were 37.2 (10.0) for Mobility, 33.0 (12.8) for UE, 51.7 (9.8) for Peer Relationships, and 46.6 (10.3) for Pain Interference. Using EQ-5D-Y-3L, some or a lot of problems were reported by 56% of children in Mobility, 57% in Looking After Myself, 50% in Doing Usual Activities, 46% in Having Pain/Discomfort, and 37% in Feeling Worried/ Sad/ Unhappy. Compared to child self-report, parents reported significantly lower (i.e., worse) PROMIS T-scores in domains of Mobility, UE, Peer Relationships, and significantly higher (i.e., worse) T-scores in Pain Interference ($p <0.001$ for all domains). Kappa coefficients showed slight agreements for child-parent reports of Mobility ($K=0.14$), Looking After Myself ($K=0.15$), Doing Usual Activities ($K=0.07$), Having Pain/ Discomfort ($K=0.16$), and Feeling Worried/Sad/Unhappy ($K=0.06$). Wheelchair use, history of small for gestational age, prolonged hospitalization after birth, having a full-time employed parent, and caregiver's stress were associated with lower HRQL scores. Conclusion: Integration of HRQL knowledge in AMC clinical care and research will improve patient- and family-centered care, promote value-based medicine, and harness the development of social care and clinical pathways focusing on the well-being of children and youth with AMC, and their families. Findings indicate the importance of considering both the child's and parents' reports of HRQL, and promote HRQL among children with AMC, through multimodal interventions focusing on the effect of childhood and parental health outcomes.

Pain assessment in children and youth with arthrogryposis using Adolescent Pediatric Pain Tool (APPT)

Shahrzad Nematollahi[1, 2], Emmanouil Rampakakis, Alexa Cirillo, Reggie C. Hamdy, Frank Rauch[2], Lauren C. Hyer, Michelle A. James, Haluk Altiok, Jonathan Pellett, Cary Mielke, Sarah B. Nossov, Sena Tavukcu, Philip F. Giampietro, Noémi Dahan-Oliel

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Background: Pain is very common in Arthrogryposis Multiplex Congenita (AMC), due to the phenotypic impairments and continuous treatment approaches including orthopedic surgeries. Pain is more common in adults with AMC, although most adolescents and young adults living with AMC experience at least mild pain and that pain was associated with decreased mobility. This study aimed to describe the intensity, location, quality of pain and interference of pain in AMC; and identify the associated factors. Methods: Data of 205 individuals with AMC aged 8-21 years from a North American registry (2019-2022) across eight Shriners Children in Canada and US was used. AMC was classified into Amyoplasia, Distal arthrogryposis (DA), and CNS/syndromic AMC. Pain was measured using the Adolescent Pediatric Pain Tool (APPT) and the PROMIS Pain Interference (PROMIS-PI). Results: 49% had Amyoplasia, 32% had DA, and 19% had CNS/syndromic AMC. Male-female ratio was 1.07. Twenty percent were current wheelchair users, 40% had prolonged hospitalization at birth, and 31% were small for gestational age. On the APPT pain score, 45% had no pain, 19% had 0 to 1 scores, 29% had 1 to 4 scores, 20% had score of >5. Pain was on extremities in 29%, head and neck in 4%, abdomen in 11%, back in 13%, and chest in 5%. Mean (SD) pain intensity was 1.71 (2.24) in Amyoplasia, 2.0 (2.6) in DA, and 1.0 (2.3) in CNS/syndromic AMC. Pain quality descriptor were 35% for sensory, 29% evaluative, 5% affective and 19% temporal. The final regression models are being undertaking to identify the associated factors with pain. Conclusion: Our interim analysis shows that pain is prevalent in children and youth aged 8-21 years with AMC. More than half of the children reported degrees of pain with higher proportion on body extremities. It appears that children with DA are more likely to report pain. Our findings will contribute to develop treatment plans and management strategies for children and youth with AMC. Characterizing pain that is experienced by individuals with AMC needs to be better understood, particularly factors and outcomes associated with pain in AMC.

TLR4 gene polymorphism and biochemical markers as a tool to identify risk of osteoporosis in women from Karachi

Rozeena Baig[1]

[1]University of Krachi

Background: Osteoporosis, characterized by low bone mineral density, poses a global health concern. Diagnosis increases the likelihood of developing osteoporosis, a multifactorial disorder marked by low bone mass, elevating the risk of fractures in the lumbar spine, femoral neck, hip, vertebras, and distal forearm, particularly in postmenopausal women due to bone loss influenced by various pathophysiological factors. Objectives: To investigate the association of serum cytokine, bone turnover marker, bone mineral density and TLR4 gene polymorphism in pre and post-menopausal women and to find if any of these can be the potential predictor of osteoporosis in postmenopausal women. Material and methods: Study participants were consisting of Group A (n=91) healthy pre-menopausal women and Group B (n= 102) healthy postmenopausal women having ≥ 5 years' history of menopause. ELISA was performed for cytokine (TNFß) and bone turnover marker (carboxytelopeptides), respectively. Bone Mineral Density (BMD)was measured through dual X-ray absorptiometry (DEXA) scan. Toll-like Receptors 4 (TLR4) gene polymorphisms (A896G; Asp299Gly) and (C1196T; Thr399Ile) were investigated by PCR and Sanger sequencing. Results: Statistical analysis reveals positive correlation of age and BMI with T scores in premenopausal group whereas in post-menopausal group found a significant negative correlation between age and T-score at hip (r = - 0.352**), spine (r = - .306**), and femoral neck (r = - 0.344**) and a significant negative correlation of BMI with TNF-ß (- 0.316**).No association and significant differences were observed for TLR4 genotype and allele frequencies among studied groups However, both SNPs exhibited significant association with each other. Conclusions: This study concludes that BMI, BMD and TNF-ß are the potential predictors of osteoporosis in post-menopausal women. However, CTX and TLR4 gene polymorphism did not appear as potential predictors of bone loss in this study and apparently cannot help in predicting bone loss in post-menopausal women.

Patient acceptability for pharmaceutical treatment in arthrogryposis multiplex congenita - the PERCEPTION study.

Klaus Dieterich[1, 2], Camille Berthet, Marjolaine Gauthier

[1]National reference center for arthrogryposis multiplex congenita, CHU Grenoble Alpes, France, 2Univ. Grenoble Alpes, Inserm U1209, IAB, CHU Grenoble Alpes, Medical Genetics, France

Background and Objective(s): Existing treatments in arthrogryposis multiplex congenita (AMC) are essentially symptomatic, including pain management, technical adaptations, and surgery. To date, there is no drug treatment that directly acts on functional deficits, and studies are very limited. This raises the question of patient acceptance for a drug treatment. The aim of our study was to assess the acceptability of a medical treatment in patients with AMC. Study Participants & Setting: All children and adults referred to the AMC reference center at the Grenoble Alpes University Hospital had previously undergone a multidisciplinary assessment and had had a precise diagnosis. They were all registered in the PARART database (NCT05673265). Methods: This was a prospective observational questionnaire study using the LimeSurvey® software. Results: Response rate was 33% (62 complete responses/ 189; 32 adult patients, 30 parents of affected children; 44 females, 18 males). Twenty-five responders were confronted with Amyoplasia, 21 with Distal Arthrogryposis (DA), 4 with a diagnosis from the "3rd group", and 12 did not know the exact diagnosis of AMC. A long-term medication related to AMC was rarely taken (n = 6; 10%). The acceptance rate to take medical treatment for AMC was 78%. Pain and improved autonomy seemed to be important motives for acceptance, with a 75% and 92.5% acceptance respectively. The most important drug characteristics were efficacy (important for 96% of patients), impact on daily life activities (92%), and adverse effects (90%). Among the conventional dosage forms, the oral route in liquid or solid form was the most acceptable (acceptable for 90% of patients). The most acceptable frequency of administration was once a week (89%). The best tolerated duration of treatment was only a few days (87%), and the maximum acceptable delay before noticeable effects was a maximum of 1 month. Conclusions/Significance: These results have led us to conclude that patients with AMC would be in favor of taking drugs with specific pharmacological properties and for specific motives, as to relieve the pain or improve their autonomy.

Severity of multiple congenital joint contractures in patients with ZC4H2 sequence variants depends on peripheral nervous system involvement.

Xenia Latypova[1, 2], Maryia Raykova, Hoai Thu Nguyen, Charlotte Dubucs, Delphine Heron, Sébastien LEbon, Florence Demurger, Magali Gorce, Melanie Fradin, David Genevieve, Nicole Revencu, Frédérique Nugues, Julien Faure, Bertrand Isidor, Judith Melki, Yline Capri, John Rendu, Alain Verloes, Sandra Whalen, Klaus Dieterich

[1]Département de Génétique, Hôpital Robert Debré, AP-HP, Paris, France

[2]Université de Paris, UMR7216, Epigénétique et destin cellulaire, Paris, France

Background and Objective(s): Nucleotide variants, balanced chromosomal rearrangements and copy number variations involving ZC4H2 located at the Xq11.2 locus have been identified in patients with AMC, severe psychomotor delay and distal muscle weakness. The clinical spectrum has recently extended to forms of isolated spastic paralysis or with mild cognitive deficit. The entire spectrum is currently designated ZC4H2-associated rare diseases (ZARD). Our aim was to describe in detail the central and peripheral neurological phenotypes, including psychometric data and muscle MRI. Study Participants & Setting: We formed a French-speaking cohort of twenty-five patients including three fetuses diagnosed with ZARD. Methods: Clinical, neuropsychological and imaging data were collected retrospectively using a comprehensive case report form. Results: All patients presented with AMC at birth, and with a developmental disorder of varying severity, for those born alive. Joint limitations disappeared in those with the most moderate muscular involvement on muscle MRI, but persisted at least as camptodactyly in the others. Neuropsychological assessments carried out in four patients revealed processing speed indices below normal, but with cognitive efficiency within normal values, as well as attentional deficits. More than half of all patients presented with either a Hoffmann or Babinski sign, or a full blown spastic paralysis. Muscle imaging performed in eight patients showed severe muscle atrophy predominantly in the lower limbs (thigh, calf), but preservation of the adductor longus and brevis muscles, further highlighting an involvement of the peripheral nervous system. Seven patients presented a specific unilateral or bilateral fibro-adipous infiltration of the tensor fascia lata muscle, which could constitute a diagnostic imaging biomarker. Conclusions/Significance: We highlight specific clinical, radiological and psychometric characteristics of ZARD. Our results support an involvement of the central and peripheral nervous system in patients carrying variants of the ZC4H2 gene. Additional data are needed to clarify whether there is a correlation between central and peripheral lesions.

Use of motion analysis to inform therapy in a child with amyoplasia

Stacy Stibb[1], Xue-Chen Liu

[1]Children's Wisconsin/Medical College of Wisconsin

Case Description: A 4-year-old boy with arthrogryposis with upper extremity positioning and contractures most consistent with amyoplasia is seen within the pediatric rehabilitation clinic for functional follow up. The mother reports concerns from the occupational therapist (OT) regarding treatment goals and focus. On exam the child's contractures are limited to his upper extremities, mostly shoulders, elbows, wrists, and thumbs, with sparing of his other fingers and his lower extremities. Functionally he can walk, run, and jump. He can pick up and carry items with his hands but usually does this by crossing his arms in front of him. He uses spinal mobility and swinging of his arms to get his limbs up on tables to reach for items. His OT is inquiring about how much range of motion he has in his arms, how much she should be pushing him in therapy to strengthen, as well as wondering about actual muscle activity of his upper limbs. It was recommended the child undergo assessment in the motion analysis lab. Assessment/Results: Thirty-eight reflective markers were placed at the bony landmarks and the Vicon plug-in whole-body model was used to measure the spatiotemporal and kinematic data on the trunk and upper limb during a reach and grasp cycle. Ten surface EMG (electromyography) sensors on the upper limbs and trunk were attached bilaterally. It was noted the child had decreased reaching time, forward transport, velocity reaching and transport on the right compared to left upper limb. He demonstrated increased trunk range of motion (ROM) compared to peers, decreased shoulder, elbow, wrist, and forearm ROM compared to peers with increased wrist deviation compared to peers. On EMGs of the upper limbs there was presence of activation in all muscles tested other than the right pectoralis. Conclusion: In this case the use of motion analysis provided a noninvasive way to assess active upper limb motion within a patient with amyoplasia and prove the presence of muscle activation during movement. Based on this data we were able to inform therapy regarding continued focus on strengthening of the upper limbs to assist with improved function and independence.

Is motor assessment of additional value in prenatal detection of AMC and FADS? A 10-year cohort study.

Jill Tjon[1]

[1]Obstetrics and Gynecology, Amsterdam Movement Sciences, Amsterdam University Medical Center, Vrije Universiteit Medical Center, Amsterdam, the Netherlands

Background/Objective(s):

The rare diseases arthrogryposis multiplex congenita (AMC) and lethal forms of AMC such as fetal akinesia deformation sequence (FADS) are often missed prenatally. This is due to varied etiology, onset and severity. To distinguish the severity of AMC, motor assessment helps to differentiate whether motility is restricted to only the contracted joints or also involve other body parts, as occurs in FADS. Therefore we studied if motor assessment was of additional value to advanced ultrasound examinations (AUE) in a population at risk for AMC and FADS. Study Participants & Setting: All consecutive fetuses with ≥2 contractures on the 20 week structural anomaly scan (2007-2016) were included, all presenting at the tertiary centre of the Amsterdam UMC. Findings at AUE including motor assessment were analysed and related to outcome. Methods: All fetuses were examined using our care pathway, first examination included structural and motor assessment, two weeks later both assessments were repeated whereafter the parents were counseled. Genetic testing was always offered. Results: Sixty-six fetuses fulfilled the inclusion criteria. Based on the first AUE, FADS was suspected in 13/66, AMC in 12/66, bilateral pes equinovares (BPEV) in 40/66 and Holt-Oram syndrome in 1/66. Based on the first motor assessment the suspected diagnosis changed in 19/66, 13 worsening to FADS, 6 amelioration from FADS, and 7 confirmed FADS, resulting in 20 FADS, 7 AMC and 38 BPEV. Second AUE in 44 fetuses showed additional contractures in 2/8 FADS, and 1 IUFD. The second motor assessment changed the diagnosis in 3/43, 1 worsening BPEV into FADS, 2 ameliorations from FADS and confirmed FADS in 7 by deterioration of motility. The result was 9 FADS, 6 AMC and 29 BPEV. We performed follow-up of 42 excluded fetuses, 27 with single contractures, healthy and walking; 14 BPEV because no motor assessment was performed and 1/14 developed FADS and was stillborn; 2 AMC were excluded because of no motor assessment, one died in utero, and one had congenital myotonic dystrophia with mental retardation and motor development delay. Conclusions/Significance: The results suggest that serial motor assessment has additional value to distinguish between FADS, AMC and BPEV.

A pathway approach to congenital bone fusions of limbs and the axial skeleton: linking distal arthrogryposes with congenital bone diseases via TGF beta and BMP signaling pathways.

Klaus Dieterich[1, 2], Déborah Kaglan, John Rendu, Frédérique Nugues, Emeline Bourgeois, Charles Coutton

[1]Univ. Grenoble Alpes, Inserm U1209, IAB, CHU Grenoble Alpes, France

[2]National Reference Center for Arthrogryposis Multiplex Congenita, CHU Grenoble Alpes, 38000 Grenoble, France

Background and Objective(s): Distal arthrogryposes and multiple synostoses syndromes are disorders characterized by skeletal abnormalities and in particular congenital joint contractures and bone fusions. Among genes that underlie these two conditions, NOG, GDF5, MYH3, FLNB, FGF9 and TAB2 are recurrently involved. Our aim was to identify common pathways between genes involved in multiple synostoses and distal arthrogryposis and understand the common phenotypic spectrum of skeletal involvement associated with these disorders according to the underlying molecular mechanism. Study Participants & Setting: We retrieved patients from the pediatric and adult registry for patients with arthrogryposis multiplex congenita PARART (NCT05673265) and the French National Rare Disease database BAMARA at CHU Grenoble Alpes and selected patients from literature review. Methods: This was a descriptive and comparative study. Results: We identified TGF beta and BMP pathways as the common denominator that link all the aforementioned genes. Mutations in genes involved in TGFβ and BMP signaling are responsible of many aspects of muscle and skeletal development, which might at least in part explain the phenotypic overlap. However, we observed phenotypic variability for a given gene and even the lack of a specific phenotypic correlation between mutations with presumed opposite consequences (loss of function vs. gain of function). The majority of patients with MYH3 and FLNB mutations carried vertebral fusions whereas symphalangism is frequently associated with GDF5 and NOG pathogenic variants. Synostosis of carpal bones was especially linked to FLNB, MYH3 and GDF5, whereas synostosis of tarsal bones was linked to MYH3, and GDF5. Conclusions/Significance: TGF-β and BMP signaling pathways link skeletal and muscular disorders through abnormal joint formation. Genotype phenotype correlations remain a challenge.

Features of patients with arthrogryposis receiving blood transfusion in surgery

Sarah Nossov[1], Cleopatra Nehme, Natalie Williams, Amanda Stutma, Bruce Brenn

[1]Shriners Childrens Philadelphia

Background and Objective(s): Patients with Arthrogryposis Multiplex Congenita (AMC) often require surgery to improve joint function. There remains a lack of literature on the specific considerations for this patient population in the peri-operative and post-operative periods. This study investigated factors potentially associated with the requirement for blood products to better understand blood loss in patients with AMC undergoing surgery. Study Participants & Setting: 73 patients with AMC (age: 1-21 years, mean: 9.4 years; gender: 38 female, 35 male) who received blood products in the perioperative or post-operative period between 2017 and 2022 at an elective orthopedic pediatric hospital. Patients underwent spine (28) or lower extremity (45) surgery. Methods: This was a retrospective study of patients with AMC who underwent spine or lower extremity surgery and required blood products from 2017 to 2022. Demographics, surgical details, and blood product administration were compared between spine and lower extremity surgery. A Fisher's exact test assessed the association between gender and blood loss. Height, weight, BMI, surgical details, and blood products were compared between the two groups using the Mann-Whitney U test. Statistical significance was set at $p < 0.05$. Results: Age was significantly higher in the spine cohort ($p<0.05$). BMI percentile was significantly higher in the lower extremity cohort ($p<0.05$). Gender and length of surgery were not significantly different. Volume of crystalloid administered was significantly higher in the spine cohort ($p<0.05$). Crystalloid/kg was not significantly different. Total blood volume and estimated blood loss were significantly higher in the spine cohort ($p<0.05$, $p<0.05$, respectively). Percent volume blood loss was significantly higher in the spine cohort ($p<0.05$). Conclusions/Significance: Patients with AMC undergoing spine procedures were older whereas those undergoing lower extremity procedures had a higher BMI percentile. This is the first study investigating perioperative and postoperative blood loss in patients with AMC. To determine factors that may predict the need for blood products in this population, future research utilizing a control population is planned. This would elucidate how patients with AMC may differ from other patient populations with regard to resuscitative and blood product needs and inform how to minimize blood product need.

TESTIMONIALS

FROM SYMPOSIUM ATTENDEES

4TH INTERNATIONAL SYMPOSIUM ON ARTHROGRYPOSIS

Dear Mr. Boissonneault,



I want to thank you from the bottom of my heart for support of arthrogryposis over the many years. As you know the registry, children, and youth clinics and genome efforts have led to amazing breakthroughs and supportive care for affected individuals.

The most recent International Arthrogryposis Symposium in Montreal particularly highlighted the remarkable advances. I was very impressed by the collaboration that is needed from the many different professionals working together as a team to provide care. I was also very pleased by the amazing collaborations of patient support groups from around the world that communicate and share information and advancements to improve the care for affected individuals.

As you may know there is also an adult registry founded by an adult PhD in Rehabilitation with arthrogryposis which helps affected adults know what to expect.

I also want to thank you and the Board for the enormous honour of a lifetime award and the Keynote Address about my 50 years journey with arthrogryposis. Interestingly it began in 1973 collaborating with Hospitals in Portland and Spokane!! Thank you so much for your engagement, and caring about affected individuals and making it all possible.

Sincerely,

Judith Hall, OC, MD, DSc honoris causa, FRSC, FRCPC, FAAP, FCCMG, FABMG, FCAHS















From Amanda Gilsin, Patient Support Group Representative, France

It was so nice meeting you and your team in Montreal! I just wanted to say what a wonderful job you all did putting this symposium together. It was really amazing!

It's so inspiring to see so many lovely, passionate people from around the world coming together for AMC.

Thanks again!

From Remco Jansen, Patient Support Group Representative, Netherlands

I'm still buzzing from the symposium. I think it was a great success and that is in no small part your doing. So thank you so much, I know it has been a hell of a job.

Tomorrow I am hosting the Dutch annual patient web conference, and of course I want to report on what we heard and learned in Montreal.

From Andres Nascimiento Osorio, MD, Neurologist, Spain

Dear Noemi,

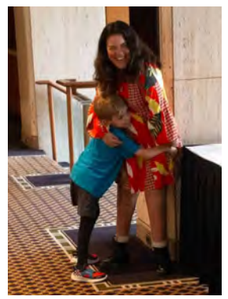
I wanted to congratulate you for the organization and for creating such a stimulating environment to continue researching and supporting families and patients with AMC. It has been a pleasure to share with this exceptional group of people, broadening the perspective and sharing different points of view.

I hope that our participation was useful and we hope to continue collaborating for a long time.

From Misha Dream Walker, international AMC ambassador, Peru

I want to take a moment to express just how deeply grateful I am for the incredible opportunity to attend the arthrogryposis symposium as a grant recipient.

Being part of the symposium was life-changing for me. The knowledge and connections I gained are invaluable, and I'm committed to sharing everything I've learned as I visit families and communities in my Arthrogryposis awareness journey. This grant has given me the tools to extend the impact far beyond I could have imagined, and I hope that what I share will continue to make a difference in the lives of others impacted by AMC, just as it has in mine.



Thank you from the bottom of my heart.

From Deanne Suddaby, participant with lived experience

I just want to say thank you from the bottom of my heart for giving me the opportunity to attend the 4th International Symposium on Arthrogryposis!

It's difficult to express how it feels to be in a room with people who are not only familiar with AMC but are eager to continue to research and help people with this condition. It was a truly amazing experience, and I am very thankful to have met such incredible people!

From Carolina Navalon, Patient Support Group Representative, Spain

I would like to thank you for the amazing job organising and welcoming us to the 4th Arthrogryposis Symposium! These were some amazing last few days!

Tricia Bucci, Physical Therapist, USA

I have also meant to reach out and commend you and your team in Montreal for organizing and hosting a wonderful conference. There were so many moments that I just felt such gratitude to be in the room with so many amazing providers, all dedicated to the care of this special group of people. It was also so nice to see the Shriners in Montreal and to meet so many of the team members there, it's a beautiful hospital in a beautiful city.

Dr. Harold van Bosse, orthopedic surgeon, United States

The sessions that delved into the lived experiences of those with arthrogryposis were engrossing, with presentations by those with AMC, their caregivers, and sharing of results from patient related outcomes surveys on pain and function. These helped to put into perspective the talks on rehabilitation, orthotics and orthopaedic interventions, in pursuit of treatments that are the most meaningful for those being treated. Sessions dedicated to patient support groups, community, and inclusive activities highlighted the increased global recognition for the social needs of those with AMC.

Kristen Donlevie, PhD OT, USA

I attended the symposium as both a person with AMC and a recently graduated Doctor of Occupational Therapy. In a room full of experts who have worked long and hard in their fields, I felt welcomed and respected for my research and point of view despite being new to my field. The mutual respect between practitioners and people with lived experience has helped to cultivate a sense of overall AMC community that is truly astounding. I am so humbled and grateful to have been a part of this gathering and excited for what moments of community like this will mean for us and the improved worldwide understanding of AMC moving forward.

AWARDS AND HONORS

Congratulations to all our award recipients from the 4ISA!

Scientific merit

People with lived experience:

Inferring the Cellular Origins of Arthrogryposis and Other Developmental Phenotypes: A Novel Computational Framework Using scRNA-seq Data

Jennifer Sullivan[1], Andrew Adey[1], Ronen Schweitzer[1,2], Shannon McWeeney[1,3], Guanming Wu[1]

[1]Oregon Health & Science University, [2]Shriners Hospitals for Children Portland, [3]Oregon Clinical and Translational Research Institute

Trainees

1. **Genetic yield in prenatal detected Arthrogryposis Multiplex Congenita, a cohort between 2007-2021**

   Maria B. Tan-Sindhunata[1], Arda Arduc1, Ingeborg H. Linskens 1, Johanna I.P. De Vries 1

   [1]Amsterdam UMC

2. **Airway Team Concerns and Management of Patients with AMC: A Scoping Review.**

   Ostap Orishchak[1], Karina Theoret[2], Alexander Moise[3], Jiahao Deng[2], Kimberley Kaspy[4], Sam Daniel[1]

   [1]Division of Pediatric Otolaryngology, Montreal Children's Hospital, McGill University Health Centre, [2]Department of Otolaryngology-Head and Neck Surgery, McGill University, [3]Faculty of Medicine and Health Sciences, McGill University, [4]Division of Pediatric Respiratory Medicine, Montreal Children's Hospital, McGill University Health Centre

3. **Pain assessment in children and youth with arthrogryposis using the Adolescent Pediatric Pain Tool (APPT)**

   Shahrzad Nematollahi[1,2], Emmanouil Rampakakis[3], Alexa Cirillo[2], Reggie C. Hamdy[2,3], Frank Rauch[3,2], Lauren C. Hyer[4], Michelle A. James[5], Haluk Altiok[6], Jonathan Pellett[7], Cary Mielke[8], Sarah B. Nossov[9], Sena Tavukcu[2], Philip F. Giampietro[6,10], Noémi Dahan-Oliel[2,1]

   [1]School of Physical and Occupational Therapy, McGill University, Montreal, Québec, Canada., [2]Department of Clinical research, Shriners Children's-Canada, Montreal, Québec, Canada., [3]Faculty of Medicine and Health Sciences, McGill University, Montreal, Québec, Canada., [4]Orthopaedic Surgery, Shriners Children's Greenville, Greenville, South Carolina, USA., [5]Orthopaedic Surgery, Shriners Children's Northern California, Sacramento, California, USA., [6]Orthopaedic Surgery Shriners Children's Chicago, Illinois, USA, [7]Orthopaedic Surgery, Shriners Children's Honolulu, Honolulu, Hawaii, USA., [8]Orthopedic Surgery, Shriners Children's Shreveport, Louisiana, USA., [9]Orthopedic Surgery, Shriners Children's Philadelphia, Philadelphia, PA, USA., [10]Department of Medical Genetics, University of Illinois, Chicago, IL, USA.



4TH INTERNATIONAL SYMPOSIUM ON ARTHROGRYPOSIS

Clinicians

1. **Open Reduction of Arthrogryposis Hip Dislocations is Associated with Higher PFGD Rates Compared to DDH: A Dual-Center Study**

   Tristen Taylor[1], Rishi Sinha[2], Nihar Pathare[1], Caitlin Perez-Stable[1], Callie Bridges[1], Basel Touban[3,1], Laura Mayfield[2], Jaclyn Hill[4], Scott Rosenfeld[3,1], **William Morris[2,5]**

   [1]Baylor College of Medicine, [2]Scottish Rite For Children, [3]Texas Children's Medical Center, [4]University of California, San Francisco, [5]University of Texas, Southwestern Medical Center

2. **Quantitative gait analysis of youth with Arthrogryposis Multiplex Congenita to improve individualized medical and surgical treatment.**

   Katerina Jirasek[1], Marianne Gagnon[1,2], Jean-Francois Girouard[1], Reggie Hamdy[1,2], **Louis-Nicolas Veilleux[1,2]**

   [1]Motion Analysis Center (MAC), Shriners Hospital for Children (SHC), Montreal, Canada., [2]Department of Surgery, McGill University, Montreal, Canada.

3. **Spinal involvement in a pediatric and adult cohort of patients with Arthrogryposis multiplex Congenita.**

   Alicia Mom[1,2], Véronique Bourg[1,2], Shenhao Dai[3], Dominique Perennou[1,3,4], **Klaus Dieterich[1,5]**

   [1]National reference center for arthrogryposis multiplex congenita, CHU Grenoble Alpes, France, [2]Department for pediatric rehabilitation and physical medicine, CHU Grenoble Alpes, France, [3]Department for adult rehabilitation and physical medicine, CHU Grenoble Alpes, France, [4]Univ. Grenoble Alpes, UMR CNRS 5105 Neuropsychology and NeuroCognition (LPNC), Grenoble-Alpes University Hospital (South Site), Dept of NeuroRehabilitation, Cs 10217 - 38043 Grenoble cedex 9, France, [5]Univ. Grenoble Alpes, Inserm U1209, IAB, CHU Grenoble Alpes, Medical Genetics, France

4. **Functional Independence of Children with Arthrogryposis**

   Lauren Hyer[1]

   [1]Shriners Children's Greenvillle

5. **Correction of mild to moderate arthrogrypotic knee flexion contractures with guided growth**

   Harold van Bosse[1], David Teytelbaum[1], Vinieth Bijanki[1], Solomon Praveen Samuel[2], Heidi Israel[1]

   [1]Saint Louis University School of Medicine, Department of Orthopaedic Surgery, Saint Louis, Missouri, [2]Shriners Hospital for Children, Philadelphia, Pennsylvania

6. **Evaluation of Foot Osteotomies for Treating Residual Clubfoot Deformities in Patients with Arthrogryposis**

   Sadettin Ciftci[1], Anuj Gupta[1], Chris Church[1], John Henley[1], Maureen Donohoe[1], Freeman Miller[1], **Reid Nichols[1]**

   [1]Nemours Children's Health

Delegation Prizes

1. Netherlands (n=7)
2. France (n=7)
3. Poland (n=5)

4TH INTERNATIONAL SYMPOSIUM
ON ARTHROGRYPOSIS

LOOKING FORWARD

The 4th International Symposium on Arthrogryposis in Montreal was a great success, thanks to the outstanding hospitality of Noémi Nahan-Oliel and Reggie Hamdy. The event brought together professionals from across the globe who treat AMC, along with individuals living with AMC and their families, who formed a vital part of the audience.

Presentations covered a wide range of topics—from the genetic and other underlying causes of AMC to early interventions such as serial casting in infants, orthotic strategies for growing children, surgical approaches, and ways to enhance daily living. The symposium also addressed the lifelong journey of those with AMC, including the transition into adulthood and aging.

We are honored to be hosting the 5th International Symposium on Arthrogryposis in Stockholm, September 20–22, 2028. We look forward to vibrant discussions, fresh ideas, and new research on the causes, treatments, and everyday impact of AMC—working together to improve care for both children and adults living with this condition.



KAROLINSKA
UNIVERSITETSSJUKHUSET
ASTRID LINDGRENS BARNSJUKHUS

AMC-teamet i Stockholm tillsammans med ordförande i AMC-föreningen, Ulric Björkén

Maria Nikolaidou barnneurolog, Eva Pontén ortoped och handkirurg, Agnes Adestedt arbetsterapeut, Ulric Björkén AMC-föreningen, Johanna Strand fysioterapeut, Lisa Ålander fysioterapeut, Hanna Steimann ortopedingenjör
Helgi Hjartarson barnneurolog saknas

Hope to see you at the 5th International Symposium on Arthrogryposis in Stockholm, September 20–22, 2028

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