



We wish you all a wonderful New Year! We are excited to see what is in store for 2023. It has been a while since you last heard from us, so we're making a New Year's resolution of our own: to highlight ALL the exciting things happening in AMC Research and more.

To help with our resolution we have onboarded new team members to help translate and build knowledge about the latest information and developments in AMC Research. We would like to introduce the AMC Research communications team led by Dr. Megan MacGillivray from St. Francis Xavier University, Nova Scotia, Canada, Dr. Bonita Sawatzky from the University of British Columbia, Vancouver, Canada, Odie Huynh, and Kathleen Dolan! This means hearing from us more often, whether it be through the newsletter, website, or even webinars.

Don't see anything that catches your eye? Read on and find out how you can shape the next newsletter or even future AMC research!

Treatment from the Comfort of Home

[Using Telerehabilitation to Deliver a Home Exercise Program to Youth with Arthrogyrosis: Single Cohort Pilot Study](#)

This idea may not be unfamiliar to many of you: phone and Zoom appointments have become a necessity following the pandemic. We think it's safe to say that both patients and healthcare professionals alike became very good at virtual meetings.

But exactly how well did telerehabilitation fare in the context of AMC? That is the million-dollar question that researchers at McGill University aimed to answer in their study.

Researchers recruited people with AMC who were 8 to 21 years old to help them develop a 12-week Home Exercise Program (HEP) based on their personalized exercise goals through their patient assessment. Therapists helped patients and caregivers to achieve these goals through 15-30 minutes of physical activity, 2 to 3 times a week, providing detailed instructions, exercise videos, and Zoom assistance. By the end of their programs, most participants not only achieved their pain and exercise goals but also exceeded expectations in some of these areas like day-to-day tasks and manual labour endurance.

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This study shows the potential of telerehabilitation for overcoming geographical distance and addresses a real need for accessible exercise programs for people living with disabilities.

You can read more about their wonderful research efforts [here](#).

AMC Community Spotlight: Meet Dr. Hall!



Dr. Judith Hall

Clinical Geneticist & Pediatrician (Retired)

BC Children's Hospital

Where are you from?

University of British Columbia, Canada

What was your field of research and expertise?

I was a clinical geneticist and pediatrician, and most of my work was done by seeing and talking to patients and families (because I hated working in the lab). I was particularly involved in studying birth defects and natural history, which is studying the time from being a newborn to an adult, and what this can tell us about genes and processes. In addition, I

studied dwarfism before joining an orthopedics team specializing in AMC Research and starting my journey learning about arthrogryposis.

What interested you about arthrogryposis?

What really interested me about arthrogryposis was the question of “why was this happening?” and “what is happening?” This mystery inspired me to look into finding out what the causes were, looking into problems with genes, blood supply, and illness during pregnancy. On top of all this, seeing how incredibly smart and resilient people with arthrogryposis are sparked my curiosity even further.

Seeing the teamwork and collaboration that goes into this research, from laboratory scientists, geneticists, pathologists, and patients motivated my desire to learn more about AMC. In particular, the way parents of children with AMC engage with research and are so supportive helped inspire me to continue research in this field.

What arthrogryposis research have you worked on?

In general, as a geneticist, I have worked on research to track and understand the genes that may cause arthrogryposis, discovering that there are different types for many different genes, and targeting these genes can be a potential avenue for treatment.

Some highlights of my research include discovering that having a baby “move” in the early stages of pregnancy was effective in decreasing the severity and likelihood of AMC and contractures. This led to recommending pregnant mothers to engage in daily exercise, or even have her drink coffee, at no harm to the baby except helping decrease AMC risk factors. In addition, by researching a case of monozygotic twins where only one twin had amyoplasia (when you would have expected both!), I worked on identifying a condition during pregnancies where twins share unequal amounts of the placenta’s bloody supply led to increased chances of amyoplasia in one twin, but not the other. I have even worked on connecting arthrogryposis and how it has shown up historically throughout time in different forms of art and Indigenous cultures.

If there is something I would still love to work on, it would be writing a history of AMC to highlight our changing understanding of the condition, and the progress made in scientific research and treatments.

What advice would you give those aspiring to pursue a similar career as you?

If I were to give advice to anyone wanting to pursue pediatric research, I would break it down into three key life lessons I learned in my journey.

1. Keep asking questions, try not to get stuck on a particular idea or topic and keep exploring new interests and avenues to make meaningful developments. Never stop developing your career!
2. Collaboration is key. I would have never made it where I am today if I did not work with other pediatricians, laboratory scientists, patients, and other international healthcare professionals.

3. Finally, I think that my secret success came from parents. Parents ask the best questions and are often the most supportive and engaged in their child's research, especially in AMC!

Is there anyone who you would like to thank or has inspired you in your career?

I would have to first give credit to my undergraduate biology professor, who despite being one of the driest professors I've ever had, for helping me become a clinician by unknowingly giving me the answer to my medical school interview question.

Secondly, I would like to give a special thanks to Dr. Lynn Staheli for allowing me to work with a team of orthopedists to collaborate internationally to teach, share knowledge, and research causes and treatments for AMC.

Finally, I have to mention how women have inspired me in my journey. I especially look fondly back on my pathology professor, and how she would invite all five female medical students for dinner, creating a safe space for discussion and celebration. When I became chair of pediatrics, I would similarly take the female heads of departments to dinner as well to continue advocating for female collaboration.

Where do we find you if you are not doing research?

If I'm not doing research, one thing I would love to start is writing a memoir on my life journey and experience. I have lived through the genetics revolution, and women's revolution, and even lived through an older brother so I know I can offer some meaningful advice. If not writing a memoir, you can likely find me outside, because I love hiking, camping, and travelling. And if not doing any of those things, you can find me doing my best to age gracefully!



Sharing Your Experiences with COVID

With over 600 million confirmed cases of COVID-19 as of January 2023, everyone has been impacted in some way whether it be their physical, mental, and/or social wellbeing. The impacts of public health restrictions have had a bigger impact on the lives of people with disabilities. Reduced healthcare and mental health challenges are some problems to name a few.

The situation of the pandemic has been challenging for many; however, we have seen some positives like the greater practice of telerehabilitation. We acknowledge that each disability

and person are unique, thus we want to hear directly from AMCers like you what your experience has been throughout the pandemic.

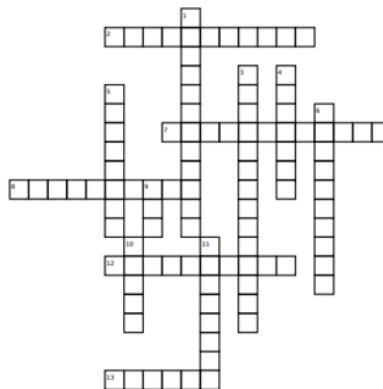
We are recruiting participants from the adult registry for this 'Covid sub-study' which involves a 15-20 minute survey about your experiences with COVID-19. There are places to provide longer answers if you want to share more.

[Click here to learn more about the COVID sub-study here.](#)

If you have not signed up for the Adult Registry and want to know more, please email Dr. Bonita Sawatzky at bonita.sawatzky@ubc.ca.

AMC Crossword: February Edition

AMC Crossword



Down:

1. Stiff joints at birth
3. When a disease does not typically get worse with time
4. Happens inside the womb
5. Treatment provided to help move joints and strengthen muscles (____ Therapy)
6. A condition from birth
9. Acronym for a splint that holds the foot and ankle
10. Acronym for an Arthrogyriposis support organization
11. Common foot problem for people with AMC

Across:

2. A type of surgical specialty to address bone and joint problems
7. Type of therapy to help with activities of daily living (____ Therapy)
8. A mobility device
12. The most common type of Arthrogyriposis
13. A device that holds a limb in place

If you would like to complete the crossword, [click here for your own copy.](#)

Contact Us & Get Involved

We want these newsletters to be meaningful to you! What would you like to learn more about related to living with AMC? What do you think AMC research priorities should be?

Please send your ideas to us at bonita.sawatzky@ubc.ca and we will see how we can explore these further in our next newsletter, or better yet, in a webinar event.

Acknowledgments | Declaration of Interest

We would like to thank Arthrogyposis Multiplex Congenita Support, Inc. (AMCSI) for supporting our knowledge translation efforts and enabling us to get the most up-to-date research findings to you!



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