

Long-Term Outcomes of Arthrogyriposis

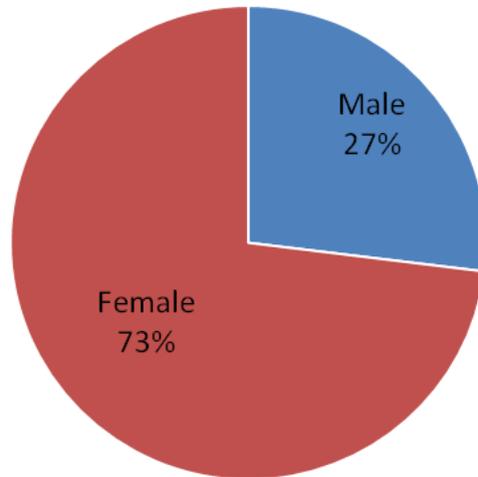
Introduction

Arthrogyriposis or arthrogyriposis multiplex congenita (AMC) is a non-progressive condition causing two or more joint contractures since birth mainly affecting the limbs and sometimes jaw, neck, and spine. The incidence of AMC is between 1:3000 and 1:5000 live births. Arthrogyriposis is an umbrella term that refers to nearly 300 specific disorders. Amyoplasia, the classic type of AMC, has an incidence rate of 1:10,000 and usually involves symmetric rigid contractures such as fixed extended elbows and club feet as well as significantly reduced limb muscle mass. At the 2014 International Arthrogyriposis Symposium in St. Petersburg, Russia, there was an agreement among attendees that very little is known about the outcomes of adults living with AMC. Understanding what happens in adults over the age of 30 is essential in providing families with better prognostic factors and identifying the best therapies in both childhood and adulthood. Thus, the primary purpose of this study was to describe the functional long-term outcomes in adults with AMC. The secondary purpose was to compare the functional outcomes as well as early and late management between subtypes of AMC.

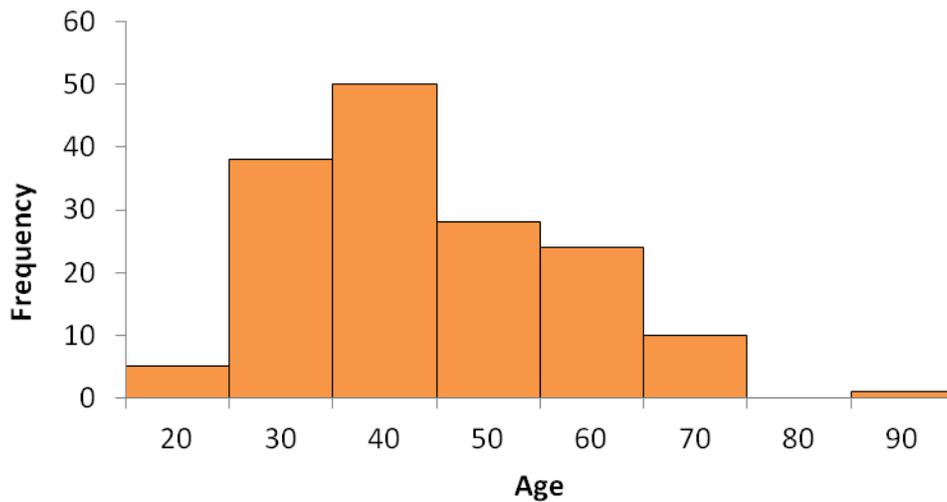
This was the largest English language survey of adults living with AMC. We advertised the study through AMC support groups and clinics in the world. Data were collected using an online questionnaire and included demographics (age, sex, country of origin, education), method of ambulation, occupation, and medical conditions. Information on description of AMC and related treatments, current Quality of Life (SF-36), and levels of physical activity (Physical Activity of Individuals with Physical Disabilities) was also collected. The Quality of Life and physical activity levels were assessed using standardized questionnaires that enabled us to compare our study data with those of other physical disabilities. Interested participants were contacted for a follow-up telephone interview and further details about the positioning of their limbs and joints at birth, their surgical and non-surgical management in both childhood and adulthood were collected.

Preliminary Analysis of Results

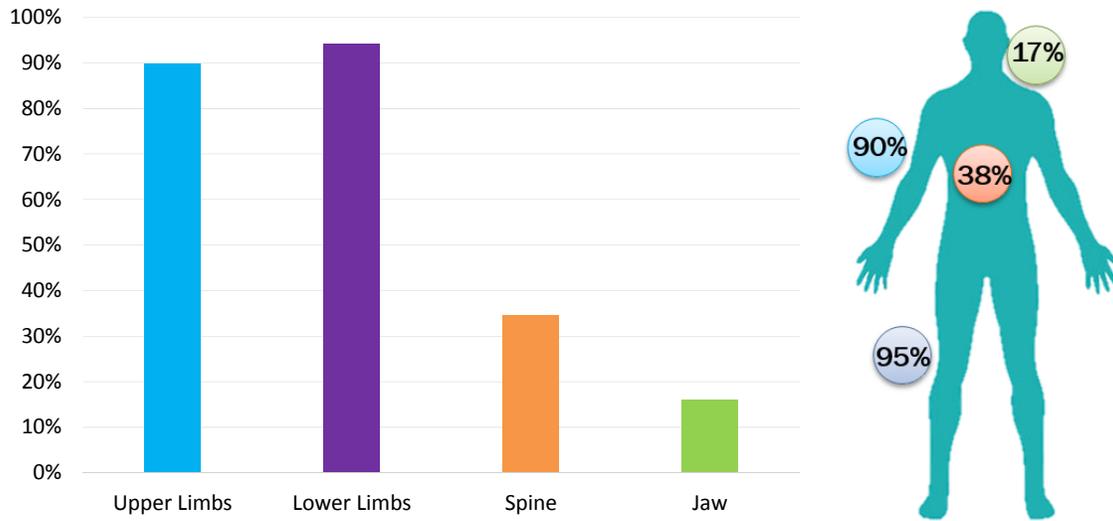
156 individuals completed the online questionnaire. There were participants. Although there is relatively little gender difference for those with AMC there was a lower number of male participants (42) vs female (114) females, which possibly suggested that women living with AMC may be more inclined in searching for information related to their condition.



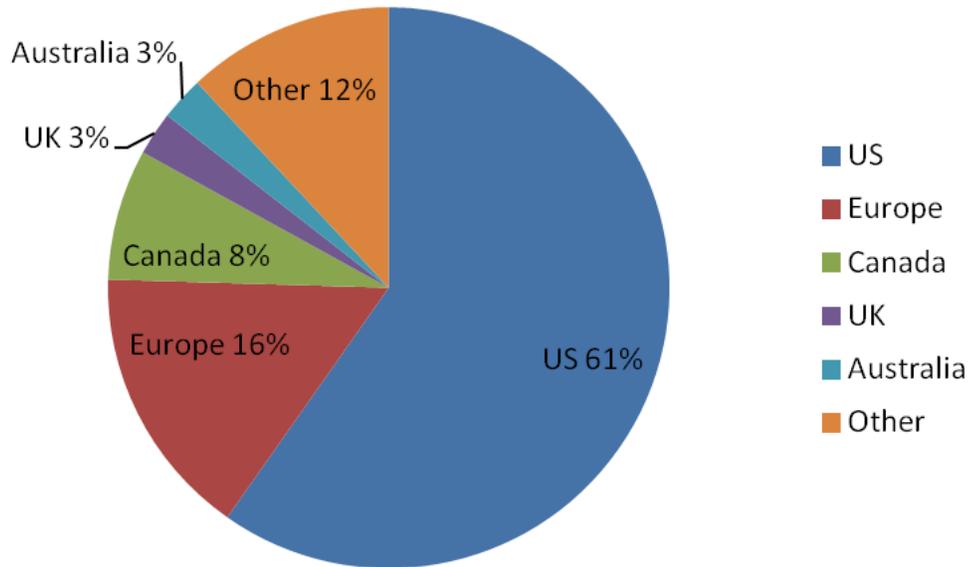
The age range of the participants was from 20 to 84 with an average age of 40 years old. The wide age range suggested the interest of individuals with AMC to participate in the study as well as the demand for information on long-term outcomes, even for those in their 80's. It also indicates a good life expectancy.



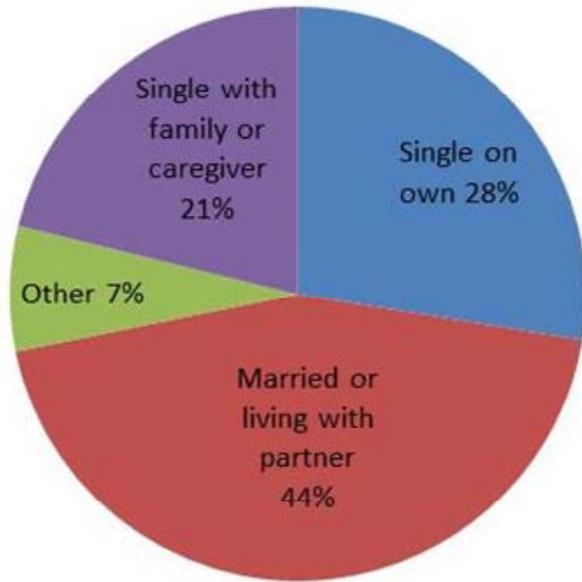
Main body areas that were involved with Arthrogyrosis included most commonly the upper limbs, lower limbs or both followed by spine involvement such as scoliosis or lordosis and jaw involvement.



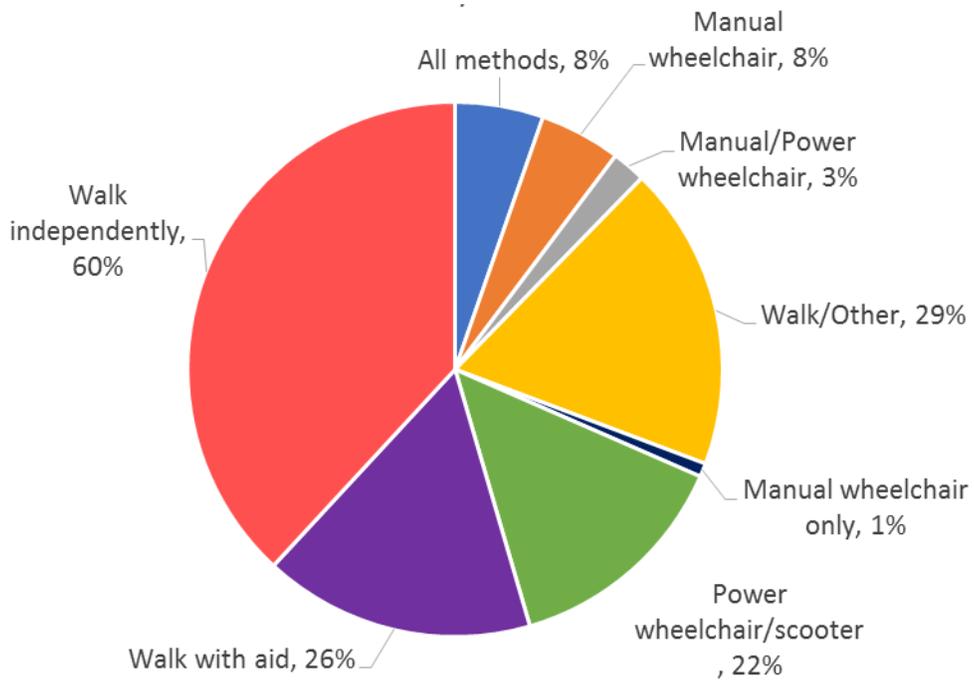
The majority of the participants were from the United States with the European Union and Canada in the second and third place respectively. Participants from a number of other countries where English is not the first language, such as Ukraine, Poland, Brazil, Peru and Germany, also completed the study.



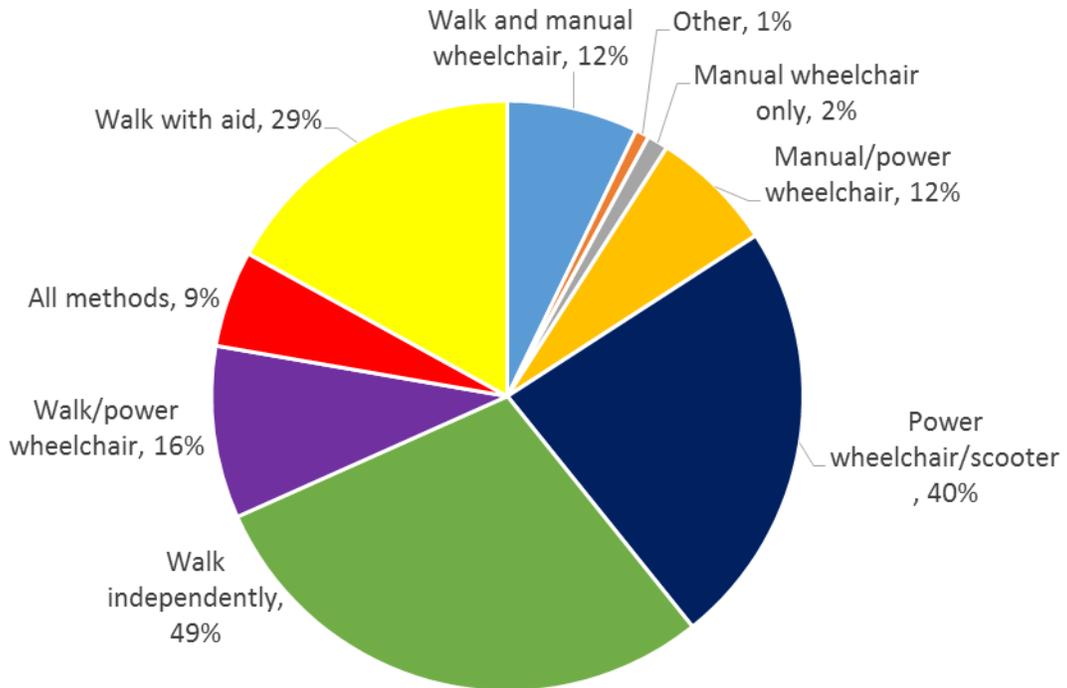
Based on the information provided on the current living situation, the majority of the participants were either living with a partner or on their own. This data indicated that most individuals with AMC have maintained a degree of independence despite their physical limitation.



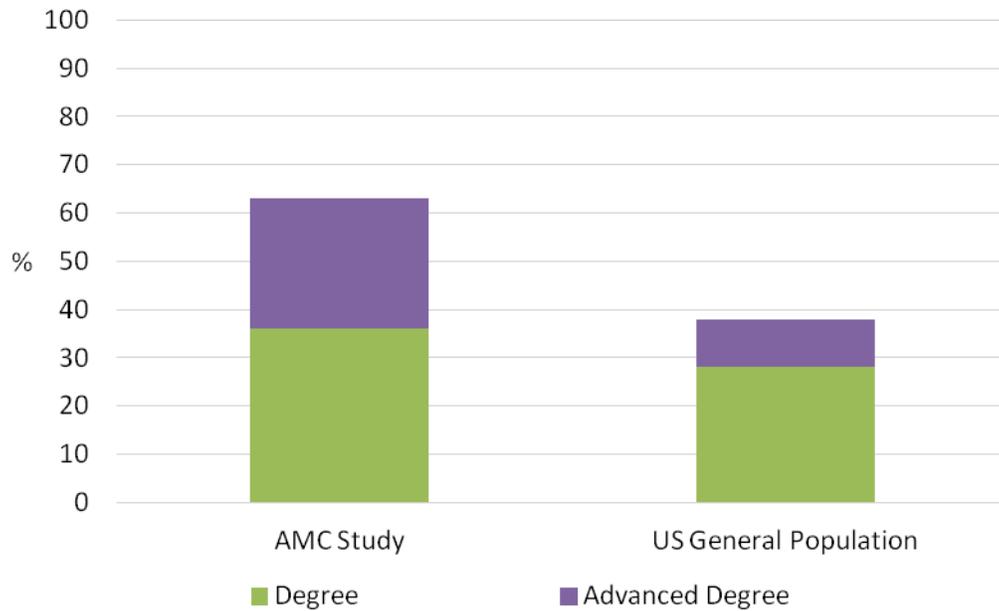
Method of home ambulation included 60% of the participants who walked independently. It appears that treatments such as correction of club feet have directly contributed to enabling these individuals to walk, based on the interviews.



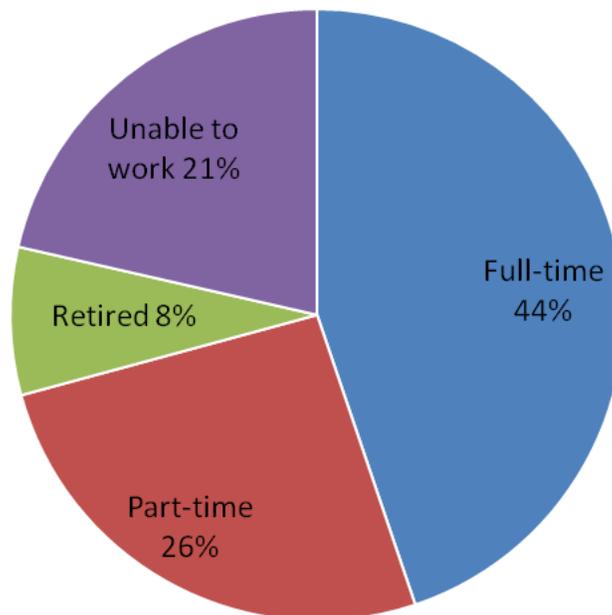
Method of ambulation outside of home indicated that individuals with AMC used alternative methods more commonly to travel longer distances, commute to work, school, and other daily destinations in order to save time and preserve their energy level.



The study participants completed a higher number of university degrees (e.g. Bachelors) and advanced degrees (e.g. Masters, PhD.) compared to the general US population. Physical limitations may have motivated these individuals further to achieve university education in order to be competitive for careers that require little physical labor. This also indicated they had the cognitive ability to do these graduate degrees.

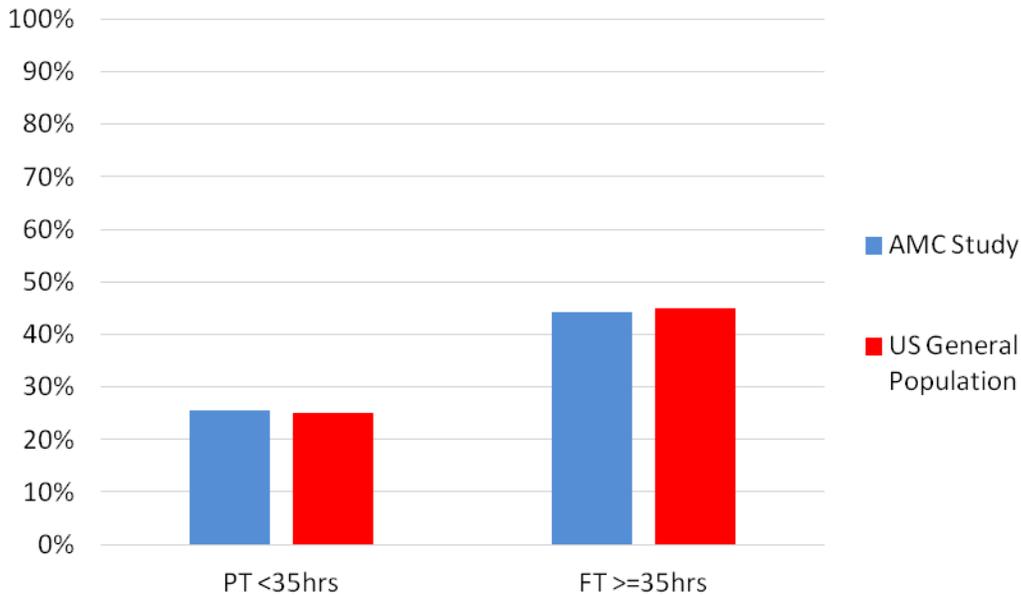


All study participants have worked in a paid or non-paid occupation. However, when asked about their current occupation status, only 21% were unable to work. Most of these individuals had to quit working due to high levels of pain (46% of our participants reported taking routine pain medication) or in order to maintain their eligibility for receiving disability funds. The type of occupation of the participants varied from office-type work to jobs requiring physical activity such as nursing.

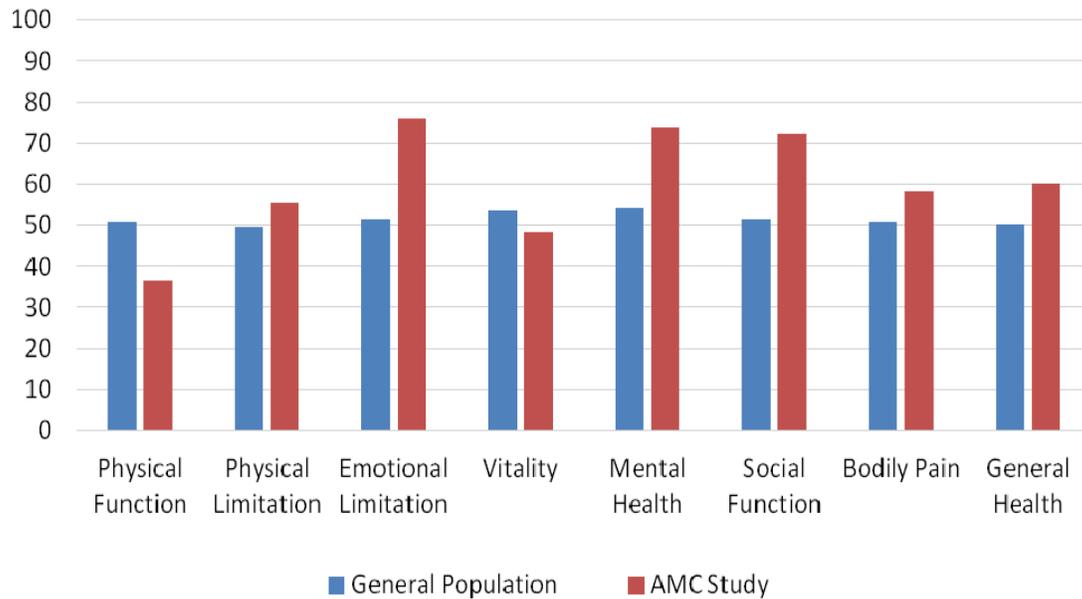


A comparison of the percentage of the study participants who worked full-time (FT) and those who worked part-time (PT) is shown with the general US population. Interestingly, individuals with AMC work

almost as much as the general US population despite their physical limitations and high level of daily chronic pain.



The Quality of Life questionnaire assessed physical function, physical limitation, social function, emotional limitation, vitality, mental health, bodily pain and general health. Physical function is assessed by questions on capacity for physical activities such as carrying groceries or climbing a flight of stairs. Physical limitation is assessed in terms restrictions in physical activities. Similarly mental health and emotional limitation assess the functional capacity and limitations that interfere with normal social activities and mental and emotional aspects of life. Vitality assesses the level of energy and fatigue. Bodily pain assesses the overall severity of experienced pain. This was a self-reported questionnaire and was a reflection of each individual's opinion about his or her own health status. A higher score in each category indicated a better Quality of Life. The Quality of Life measure was compared for the study participants and the general US population. With the exception of vitality and physical function, individuals living with Arthrogyrosis had better outcomes than the general population.



The physical activity questionnaire assessed the amount of time spent doing stationary activities as well as exercise, indoors and outdoors house-related work, and occupation-related activities. The combination of the different categories then provided a total score, which was representative of how active an individual was. This total score could be used to compare different groups such as the general population with adults living with AMC. A higher physical activity score meant more time was spent performing non-stationary activities. The average score for the level of activity of the individuals with AMC was at 13.5 while those with other types of disabilities maintained an average score of 11. Able-bodied individuals, who maintained moderate activity defined as three 30-min workouts per week, received an average score of 22.

Adults living with AMC did remarkably well compared to able-bodied population despite dealing with pain on a daily basis. Individuals with AMC exhibited persistence and determination and were enthusiastic to gain knowledge about their condition. At this stage, we are compiling and analyzing the study data with the aim of publishing the results in the near future.

Questions

Please directly contact the study’s primary investigator, Dr. Bonita Sawatzky (bonita.sawatzky@ubc.ca) if you are interested in taking part in the study or if you have any questions.