ARTHROGRYPOSIS NEW PARENT INFORMATION PACKET

A welcome packet for families during pregnancy and early infancy for a child with Arthrogryposis Multiplex Congenita

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AMCSI

Arthrogryposis Multiplex Congenita Support, Inc.

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AMCSI
Arthrogryposis Multiplex Congenita Support, Inc.

Dear Medical Professional,

Arthrogryposis Multiplex Congenita Support Inc. *(AMCSI)* is a 501*(c)(3)* nonprofit organization dedicated to raising awareness about arthrogryposis multiplex congenita (*AMC)*. We kindly ask that you share the enclosed information with your colleagues or any families who may benefit from these resources.

Arthrogryposis is a congenital condition characterized by multiple joint contractures and muscle atrophy. Many parents receive a distressing prognosis upon their child's initial diagnosis, prompting them to seek information and support immediately.

AMCSI provides an online support community featuring discussion boards on a range of topics, including therapy, surgeries, home modifications, and receiving an AMC diagnosis in utero. Additionally, we host an annual conference that brings together families and medical professionals for a weekend of education, connection, and support.

We hope you will review the enclosed materials, become familiar with our organization, and share these resources with those who may find them valuable. The next time you speak with families who are newly navigating an AMC diagnosis, we encourage you to provide them with the support and information available through AMCSI.

This packet includes:

- A letter for medical professionals
- An AMCSI brochure and business cards
- A Frequently Asked Questions (FAQ) document about AMC

Please feel free to contact us with any questions, and visit our website at www.amcsupport.org to learn more about the resources we offer.

Sincerely,

Ani Samargian
Founder, Arthrogryposis Multiplex Congenita Support, Inc.

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Arthrogryposis Multiplex Congenita Support, Inc.

Dear Parent,

Welcome to the AMC Family! It is probably a “family” you never knew existed until now, and being a part of it was not in your plans when you found out you were expecting. Finding out your child has arthrogryposis is a shocking experience, whether it was discovered by ultrasound during your pregnancy or not until birth. The joy, wonder, and hope for your new baby can quickly be overshadowed by fear, confusion, and isolation. The flood of medical information can be overwhelming. WE ARE HERE TO HELP! Arthrogryposis Multiplex Congenita Support, Inc (AMCSI) and Take TIME for AMC, Inc are two 501(c)3 non-profit organizations formed with the goal of raising awareness of AMC and providing pertinent information to individuals and families touched by AMC as well as the medical community who cares for them.

My name is Sara Lemin. I am the mother of two boys with arthrogryposis – one by birth and the other by adoption. I am also a board-certified OB/GYN physician near Cleveland, OH. In 2017, we found out about our son’s AMC during my pregnancy at around 22 weeks. Even though I work in obstetrics, and my husband Ian is a critical care ICU nurse, we had not learned about arthrogryposis during our training. We had to start at the beginning just like you. We were scared for our baby’s health and sad for the challenges he would face. We felt disappointed and guilty as we learned to mourn the loss of the ‘normal’ baby we were imagining and to accept with love our baby with AMC. We had a chance to meet with specialists and were offered tests to help us determine a cause, but ultimately, we just had to wait until our son was born to know exactly how he would be affected and what his abilities and needs would be.

During that waiting period, one of the most valuable sources of information, hope, and reassurance came from AMCSI at their website amcsupport.org. While other websites and Google searches offered grim outlooks and scary pictures, AMCSI showed us the wonderful ways in which children and adults with AMC are thriving. We were able to connect with other parents who gave us practical advice on how to care for our son and where to seek medical care. When our son was 7 months old, we attended our first AMCSI Annual Conference for families and felt an instant connection. It is a blessing to be around other parents who just “get it” and to see children playing together with acceptance and belonging. It was in one of the online forums where we first learned about a little boy in Ukraine with AMC who would eventually become our son! Eventually, we started our own non-profit, Take TIME for AMC, to focus on the prenatal diagnosis of AMC and care during pregnancy.

During these early weeks and months, you will be very busy with medical tests, doctor appointments, therapy schedules and coordinating care for your baby. When it seems overwhelming, give yourself some grace and take it one day at a time. Allow yourself the space and time to process the grief, anger, and unfairness. Remember to pause and enjoy the beautiful moments with your child. Ask as many questions as you need to understand your child’s needs and to be an active member of the team involved in their care. Reach out to AMSCI, Take TIME for AMC, and other support organizations linked to their website. Know that you are not alone in this new journey!

In kindred spirit,
Sara Lemin MD

FREQUENTLY ASKED QUESTIONS

WHAT IS ARTHROGRYPOSIS MULTIPLEX CONGENITA (AMC)?

Arthrogryposis Multiplex Congenita (AMC) is a term used when a person has contractures (stiffness) in two or more body areas that are present at the time of birth.

- Arthrogryposis = joint contracture
- Multiplex = two or more body areas affected
- Congenita = present at birth

The word "arthrogryposis" itself comes from the Greek (arthron + gryp + osis) that describes a stiffness, curvature, or contracture of a joint that gives it an abnormal appearance.

- Arthron = Joint
- Gryp = Curved
- Osis = Having the condition

People often use the terms arthrogryposis and AMC interchangeably.

WHAT CAUSES ARTHROGRYPOSIS TO DEVELOP?

Early in pregnancy, the developing baby is correctly formed and bones are connected properly at the joints. A contracture develops when that joint does not move properly during pregnancy. In order for a joint to move, the muscles attached need to be able to pull the joint back and forth, and proper signals need to flow from the brain and nerves to tell the muscles to move the joint. If there is a problem with any step in this process, arthrogryposis can develop.

The joint must also have enough room to move within the uterus. Factors that cause crowding in the uterus, such as twins, fibroid tumors, or low amniotic fluid can also lead to the development of joint contractures, though these conditions are less common.

It is important for parents to know that it is not their fault that AMC has developed. There is nothing they did wrong or failed to do during the pregnancy.

ARE THERE DIFFERENT TYPES OF AMC?

Yes! There are over 400 different diagnoses or conditions that can lead to a baby being born with arthrogryposis. What they all have in common is that the baby did not move the affected joints normally during pregnancy. No two people with AMC are the same, so it can be difficult to group them into types or categories. Researchers have proposed various ways to group the types of AMC into categories.

- Based on which body parts are affected:
    - Only limbs involved
    - Limbs, head/face, and or other organs involved
    - Limbs and severe central nervous system involvement
- Grouped into categories of diagnosis
    - Amyoplasia – often called the "classic form". It is the most common and accounts for about 1/3 of all babies born with AMC.
    - Distal Arthrogryposis – only hands and/or feet affected. Often related to a specific gene mutation.
    - "Everything else" including the remaining 300-500 conditions/diagnoses/syndromes

FREQUENTLY ASKED QUESTIONS *(continued)*

HOW IS THE TYPE OF AMC DETERMINED?

The type of AMC is usually determined by a physical exam to determine which joints are affected and whether any other organs or body systems are involved.

In addition to a physical exam, other tests to evaluate the different steps involved in joint movement can help narrow down a possible cause. For example:

- MRI of the brain and spine to look for a problem in the central nervous system
- Blood tests to screen for metabolic problems or gene mutations in the DNA

Seeking out a geneticist with knowledge of AMC is recommended early on to help determine whether a baby has a genetic cause as this may help steer the course of treatments.

WHICH JOINTS ARE AFFECTED BY AMC?

Any joint can be involved if its movement is limited during development leading to a contracture. No two people with AMC have the exact same joint involvement. Commonly involved joints include feet/ankles, knees, hips, fingers, wrists, elbows, shoulders, spine, and jaw.

ARE THERE ANY NON-JOINT RELATED PROBLEMS IN AMC?

Some, but not all, babies born with AMC can have other problems. During pregnancy, babies need to breathe and swallow amniotic fluid in order for their lungs and digestive systems to develop properly. If a baby has weakness in swallowing or breathing during pregnancy, it can lead to feeding and breathing issues after birth, such as:

- Problems with feeding that may require a feeding tube
- Problems with breathing that may require a breathing tube or ventilator
- Difficulties with anesthesia during surgery
- Other organs (undescended testicles, gastroschisis, underdeveloped lungs)

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FREQUENTLY ASKED QUESTIONS *(continued)*

HOW IS ARTHROGRYPOSIS TREATED?

There is no cure for arthrogryposis, but there are many therapies that can help improve the shape and function of affected areas of the body.

- Stretching and range of motion exercises
- Braces and orthotics
- Physical Therapy
- Occupational Therapy
- Speech Therapy including feeding/swallowing
- Aqua therapy
- Adaptive Equipment: walkers, wheelchairs
- Serial casting – this involves gently stretching a joint and holding it in that stretch with a cast. Each week the cast is removed and the joint stretched a little further and recasted. The process is repeated until the ideal position is achieved. The Ponseti Method is an example of serial casting used to correct clubfoot deformities of the feet and ankles, which are common in AMC.
    - Casting is often one of the first treatments done for babies, but it can wait a few weeks/months after birth to allow time for the family to bond and get to know their baby's personality before adding something new to the mix.

Surgery can help correct abnormally shaped bones and joints. Surgeries are an option after non-surgical options have been tried first. Surgeries usually do not occur before at least one or two years of age.

- Operations on soft tissues (tendons, muscles, skin) to allow a joint to move better
- Operations on bones to put a joint in a better position

Even with appropriate treatments, joint contractures often regress as children grow because the soft tissues around the joint do not grow and stretch as quickly as the bones. It is common for children to require multiple rounds of casting or repeat surgeries over the course of their growth span.

CAN AMC BE CURED?

There is no cure for AMC. It is considered a non-progressive condition, meaning that the joint contractures do not get worse or involve new joints over time. In fact, joints look their worst at birth and can improve dramatically with treatment.

WHAT CAN PEOPLE WITH AMC DO?

Each person with AMC had a unique set of abilities and challenges based on how their body is affected. Some people are affected in only their upper extremities or only their lower extremities. Many people require minimal assistance during their daily lives – they can walk, eat, write, drive, and provide their own personal hygiene care, while others need personal aides or adaptive equipment. Most people with AMC have normal intelligence. There are adults living with AMC who are doctors, lawyers, architects, authors, artists, teachers, parents, advocates, social workers, singers, dancers, athletes (even in the Olympics!) and so much more.

CAN PEOPLE WITH AMC HAVE CHILDREN?

Yes, many adults with AMC have had children of their own. Most of the time, those children are not affected with AMC. In some cases where the cause of AMC is genetic, it is possible for the children to inherit the same gene and therefore be more likely to have AMC themselves.

Adults with AMC who are planning to have children should seek advice from a medical professional (such as a geneticist, genetic counselor, obstetrician, or maternal fetal medicine specialist) to review the specifics of their particular case of AMC.

TYPE OF HEALTHCARE PROFESSIONALS

You and your child may see many *(but not necessarily ALL)* of the following types of health care professionals during pregnancy and early childhood. Not every member of a physician group may have the specific experience with Arthrogryposis Multiplex Congenita (AMC) that a family requires.

When families reach out to their local geneticists, experiences can vary significantly. Some geneticists demonstrate a strong interest and deep understanding of AMC, working diligently to establish an appropriate diagnostic pathway. Others may conduct a limited number of tests before concluding that no further action can be taken.

We also recommend that families connect with other local families who have experience with AMC to seek personal recommendations for knowledgeable providers. Additionally, for those seeking guidance on clubfoot treatment, the Ponseti International Association offers excellent resources.

OBSTETRICIAN – cares for pregnant women

MATERNAL-FETAL MEDICINE – high risk pregnancy specialist

PEDIATRICIAN – provides overall care for children and can coordinate referrals to specialists

NEONATOLOGIST – cares for newborns who are premature or need intensive care

ORTHOPEDIC SURGEON – operates on bones, ligaments, tendons, and muscles of the limbs

GENETICIST – evaluates the genes in DNA to identify mutations

NEUROLOGIST – cares for the nervous system including brain, spine, and nerves

PHYSICAL THERAPIST – helps patients with strength and range of motion exercises

OCCUPATIONAL THERAPIST – helps patients with performing daily tasks

SPEECH/FEEDING THERAPIST – helps with speaking, eating, and swallowing

ORTHOTIST – builds braces (orthoses) to help support joints

PHYSIATRIST – determines need for adaptive equipment (braces, walkers, wheelchairs)

PULMONOLOGIST – cares for problems of the lungs and respiratory system

CARDIOLOGIST – cares for problems of the heart and circulatory system

ENDOCRINOLOGIST – cares for problems involving hormone systems

PAIN MANAGEMENT – cares for chronic pain issues

PSYCHOLOGIST – provides emotional support and coping skills

CHIROPRACTOR – cares for body alignment issues

GASTROENTEROLOGIST – cares for problems of the stomach and bowels

EAR/NOSE/THROAT SPECIALIST – cares for problems of the head and neck

PLASTIC SURGEON – cares for skin issues and appearance of involved areas in AMC

SOCIAL WORKER – helps coordinate social services programs, government benefits and financial support programs

CARING FOR A PREGNANCY WITH ARTHROGRYPOSIS

I just found out my baby has arthrogryposis. **WHAT CAN I DO?**

It is important for parents to understand that they did nothing wrong during pregnancy that caused arthrogryposis to develop in their child.

• STAY HOPEFUL

Most expectant parents have not heard of arthrogryposis prior to being told their baby has it. It can be a lot of new and confusing information, and much of what is available on the internet projects a dismal outlook. There is a wide range of challenges that babies with AMC may face, but most babies do well at birth and beyond. Contractures look their worst at birth, but many joints relax with time, stretching, and therapy. Keep a positive outlook and seek information from reputable medical sources. In addition to all the physical uncertainty, soon-to-be AMC parents worry, “Will my baby be happy?” The answer is YES! Your baby will receive so much LOVE and ATTENTION from you, and that’s what really matters for a child to be happy. Your child will teach you more about strength, perseverance, joy, ingenuity, and love than you ever thought possible.

• STAY HEALTHY

As with any pregnancy, it is important to stay as healthy as possible. Follow a healthy diet with plenty of protein, fruits and vegetables. Take a prenatal vitamin that contains folic acid and DHA. Avoid tobacco use, vaping, alcohol consumption, and recreational drug use. Get an appropriate amount of sleep. Minimize stress where possible. If you have an underlying health condition, be sure it is under optimal control.

• PROMOTE MOVEMENT

Although your baby may have limited ability to move inside the womb, research suggests you can help promote movement in your baby by:

- o Exercising regularly
- o Drink a caffeinated beverage 2-3 times per day (maximum 200mg per day)
- o Take a brief, brisk walk 2-3 times per day

• GET ORGANIZED

You will have many questions during your pregnancy and after your baby is born. Many parents find it helpful to keep a notebook, computer file, or smartphone note. Write down questions you want to remember to ask your doctors. Print articles you find helpful. Often, your doctors may not be very familiar with arthrogryposis yet and are learning alongside you, so bring information you find to share with them.

• GET CONNECTED

Learning that your baby has arthrogryposis can be an uncertain, fearful, and lonely time in your pregnancy. **YOU ARE NOT ALONE!** There is a wonderful network of AMC families online and on social media that can be a very encouraging and informative source of support! The organization AMCSI *(Arthrogryposis Multiplex Congenita Support Inc)* is an excellent starting place that can direct you to online texts, other support groups, specialists and events.

- o Visit their website at **https://amcsupport.org**.
- o You can also search “arthrogryposis” on Facebook to find support groups.

• CELEBRATE THE NORMAL THINGS

It can be easy to get lost in the medical details, the unknowns, and the worry. Don’t forget to take time to enjoy the beautiful things about your pregnancy too! Take a picture of your belly bump. Create a baby shower gift registry. Indulge in those weird food cravings. Let the little movements fill your heart with love and wonder.

HOW CAN MY DOCTOR OR MIDWIFE HELP?

• COORDINATE REFERRALS

A pregnancy affected by arthrogryposis can benefit from a multi-specialty team approach. Your doctor should consider referrals to the following specialists as needed and when available. (Remember, every baby is unique and may not need ALL of these specialists, but this is a general list to use as a starting point.)

- Maternal Fetal Medicine
- Genetics
- Orthopedics
- Neonatology, including a tour of NICU
- Physical Therapy/Occupational Therapy
- Physiatry
- Neurology
- Social Services and Financial Aid
- Early Intervention Programs funded by your state

• RESEARCH MEDICAL LITERATURE AND CONSULT MEDICAL PEERS

Literature searches of medical databases can be a helpful source of up-to-date information for clinicians. Many excellent, well-trained physicians and midwives may have never encountered a pregnancy affected by arthrogryposis before and are likely to be just as thirsty for accurate information as parents. A call to a Maternal Fetal Medicine specialist may provide helpful direction. Your doctor may also reach out to Take Time for AMC Inc. or AMCSI for more information. One of our medical advisory board members, Sara Lemin MD, is an OB/GYN with a special interest in the prenatal diagnosis of AMC. Dr Lemin is happy to speak with your health care team about your pregnancy. See contact info at the end of this pamphlet.

HOW CAN I PREPARE FOR DELIVERY?

DECIDE ON LOCATION

It is advisable to plan to deliver in a facility that has the capability to care for the needs of your newborn. As many as 60-70% of AMC newborns go to the Neonatal Intensive Care Unit (NICU). Your baby may not need much additional care, or he/she may need intensive support such as a ventilator or feeding tube. Since it is very difficult to predict how much care your baby may need, delivering in a hospital with a NICU available is something to strongly consider. Your doctor can help you arrange a tour of the NICU and a consult with the care team.

VAGINAL BIRTH OR CESAREAN?

Your doctor will help you decide on the most appropriate type of delivery for your baby based on your overall medical and prenatal history. Many babies with arthrogryposis are in the breech position (70-80%) due to their limited ability to move, and a cesarean delivery would be recommended. Babies with hyperextension of the neck should always be born by cesarean section. About 1 out of every 10 babies with AMC will be born with a broken arm or leg. Recent studies suggest that the likelihood of a broken bone is about the same whether born by vaginal delivery or by cesarean section. If the baby is in a head-down position, a vaginal delivery may be appropriate. The presence of arthrogryposis should not alter the timing of delivery, and your doctor will help you plan how and when to deliver based on the usual obstetric indications.

COLLECT BLOOD FROM UMBILICAL CORD

After your baby is born and the umbilical cord is cut, there is still a fair amount of baby's blood in the cord and placenta. This blood can be collected (in EDTA tubes) and saved for later use if your pediatrician or neonatologist wants to order blood tests on the baby (i.e. genetic studies). If this sample has already been collected, it could spare your baby from an extra needle stick.

RESEARCH LOCAL RESOURCES

You can ask your pediatrician about their experience with caring for a child with arthrogryposis. Routine pediatric care for most children with AMC can be done by your local providers. Many families have their local pediatrician for routine care like check-ups and treatment of common illnesses like colds, rashes, or allergies and then they travel to larger centers to see specialists for their AMC-related care. Many children's hospitals in larger cities have an arthrogryposis clinic or a multi-specialty team already in place. Check the website of children's hospitals near you. Join social media AMC support groups and ask parents in your area where they take their children.

BUILD YOUR SUPPORT NETWORK

The days after your baby is born may be very busy. Some parents are saddened that they don't have as many opportunities to snuggle, nurse, or bond with a baby who requires extra equipment or care. If you have other children at home, it can be helpful to have extra childcare arranged ahead of time. Friends who can prepare meals or fill your freezer with premade dinners can be a wonderful help. Connecting with other AMC families online can be a huge source of support from parents who have been down the path you are on.

CARING FOR A NEWBORN WITH ARTHROGRYPOSIS

Your baby may appear fragile to you, but in most cases of AMC, they are not. You will still love them and care for them just like any other baby.

WILL I NEED SPECIAL SUPPLIES AT HOME?

Most babies with AMC can use typical baby care supplies such as car seats and bassinets/cribs. Changing diapers and dressing them may be more difficult because of the lack of joint motion. Clothing that does not need to go over a baby's head, such as kimono-style or wrap around shirts, can be very helpful. Many parents prefer sleep sacks or pajamas without feet to accommodate casts or braces. As your baby grows, there may be items that are particularly helpful to their needs, such as toys, bath seats, utensil, clothing, or seating. Parent support groups online are excellent for answering questions about such preferences and places to shop! Physical therapists and occupational therapists are also a wealth of knowledge about adaptive items and tricks for performing various tasks.

WHAT WILL THE FIRST FEW MONTHS BE LIKE?

Caring for any newborn child, especially for first-time parents, can be overwhelming! In addition to the joy of welcoming a new family member, sleep deprivation and the uncertainty of so many new duties as a parent can lead to anxiety and self-doubt. Adding AMC to the picture can multiply these feelings! Mothers of babies with special needs are more likely to develop postpartum blues or depression, so do not hesitate to reach out to your doctor if you have any concerns about your mental health! Every child with arthrogryposis is unique, and your path will have its own course. Be patient and take it one day at a time.

It is reasonable to expect a possible stay in the Neonatal Intensive Care Unit. Some babies will need treatment for essential functions of the heart, lungs and digestive systems long before attention can be focused on treating the joints. Treatments such as massage and gentle stretching can be started while prioritizing overall vital functions first.

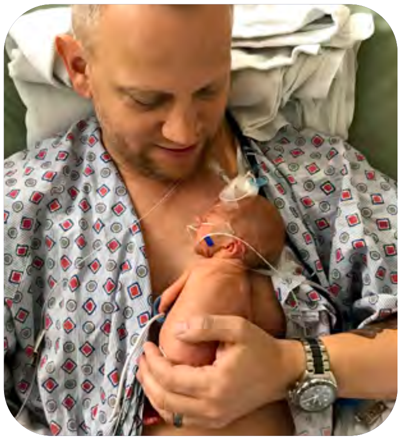
Once your baby comes home, he/she may have extra equipment such as casts, splints, monitors, oxygen, feeding tubes, etc. Don't panic! The hospital will be sure you understand how to care for these things before you come home.

Remember to take time to care for yourself and your close family relationships. Older children will benefit from keeping routines as close to normal as possible. Some family and friends may feel awkward about visiting or asking questions due to their own lack of understanding about arthrogryposis. Try not to be offended but rather use opportunities to educate others in a positive way.

You can expect to have many appointments for your baby in the first few weeks, months, and years of life. Many parents are overwhelmed by the various types of appointments and providers their child may need, but it helps to focus on short term goals and take it one step at a time. Every child is unique and will require an individualized group of providers tailored to his/her specific needs.

IS ASSISTANCE AVAILABLE FOR MY CHILD WITH AMC?

Your child will most likely qualify for early intervention services provided through your local government agencies. Many states provide these services free of charge or on a sliding scale based on family income.



Early Intervention services can include

- Access to therapy (physical, occupation, speech, etc)
- Access to specialized daycares or schools
- Providing adaptive equipment
- In-home visits with nurses

Your child may also qualify for financial medical assistance. Talk to a hospital social worker who can help direct you to state and local resources and appropriate organizations, such as:

- Medicaid or additional health insurance coverage
- State funded programs
- Local Health Department
- Local Board of Developmental Disabilities
- Charity organizations

Programs vary widely from state to state. It is beneficial to educate yourself on the programs and assistance available in your area.

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DON'T HESITATE TO REACH OUT TO US!

Part of the mission of AMCSI and Take TIME for AMC is to educate and support parents who just received the diagnosis of AMC in their newborn or pregnancy. We understand that sometimes the internet does not provide the one-on-one support that parents may need. Below are the names and contact information for parents who are raising a child with AMC who are eager to support new parents as they start this journey with AMC. We encourage new parents to contact these individuals by any means they feel comfortable with!

Ani Samargian
AMC Mom & Founder of AMCSI
Website: **https://AMCsupport.org**
Email: anisamargian@gmail.com
Phone/text: (864) 809-6099

Sara Lemin
AMC Mom, OB/GYN, & Founder of Take TIME for AMC
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