Children's Mercy

MY ANGELMAN SYNDROME CLINIC HANDBOOK

This book designed especially for:

CONTENTS

Children’s Mercy

WELCOME LETTER FROM LAURA & LEAH

Hello and welcome!

Welcome to the Angelman Syndrome Clinic at Children's Mercy Kansas City. Whether you have a loved one recently diagnosed with Angelman syndrome or you have been on this journey for some time, starting your care with a new medical home can feel very overwhelming. You are not alone.

Dr. Le Pichon heads the Angelman Syndrome Clinic and provides comprehensive support for our children. Monique, the Angelman syndrome nurse, and Dr. Le Pichon partner with specialty clinics, educational services, family support, Angelman syndrome Patient Family Advisory Council and national Angelman-centered organizations in providing optimal care for your loved one.

Please know that you are joining a clinic with an active parent presence that engages and partners with Dr. Le Pichon and Monique in the design and operations of the clinic. Your child, family and opinion matters. We look forward to meeting you and are so excited to welcome you to the Kansas City Angelman Syndrome Clinic!

With love and hope,

Laura & Leah,
Angelman Syndrome Clinic Patient Family Advisory Council (PFAC)

Guide to Children's Mercy

At Children's Mercy, we believe every kid has amazing potential.

This potential inspires us to offer more leading specialties, to research game-changing innovations, and to provide only the most compassionate, comprehensive care.

Because potential knows no bounds, we are here to help unleash the greatness in every child.

Kids come first at Children's Mercy. Always have, **for more than 125 years**. Every day employees work towards our vision:

To create a world of wellbeing for all children.

And every day our employees strive to fulfill our mission:

Children's Mercy is a leading independent children's health organization dedicated to holistic care, translational research, breakthrough innovation, and educating the next generation of caregivers.

Together, we transform the health, wellbeing and potential of children, with unwavering compassion for those most vulnerable.

The scientists at our **Children's Mercy Research Institute** are relentless in their search for answers. We work diligently every single day, making a difference for children today and into the future.

We're busy **training the next generation** of health care providers. Thousands of students each year choose to receive their training here alongside our more than 800 doctors, hundreds of allied health professionals and thousands of nurses.

But the most important thing to know about us is this: We're all about children. It's all we do. And we couldn't be more honored and proud.

Welcome to the Angelman Syndrome Clinic at Children's Mercy Kansas City

Welcome to the Angelman Syndrome Clinic at Children's Mercy Kansas City!

The Angelman Syndrome (AS) Clinic was born from the vision of Laura and Patrick Sargent, who dreamed of a specialized center in Kansas City where their daughter, Maddie, and other children with Angelman syndrome could receive expert care. When they approached me in January 2022, I was immediately inspired by their passion and commitment. Together, we brought this vision to life, officially opening the Children's Mercy AS Clinic in December 2022. Since then, our clinic has grown significantly, achieving key milestones along the way. But before I share our progress, let me introduce you to our mission and vision.

Our Mission: A medical home and a research hub for Angelman syndrome

The AS Clinic was designed to serve children with AS and their families in our region. We are guided by two primary goals:

1. Establish a medical home for children with AS.
2. Develop a regional research program dedicated to AS.

A medical home for children with Angelman Syndrome

The concept of a medical home dates back over 40 years and was formally defined by the American Academy of Pediatrics (AAP) in the 1990s as an approach to comprehensive primary care that fosters strong partnerships between families, clinicians, and support services (**AAP Medical Home**).

When you visit the AS Clinic for the first time, we take the time to fully understand your child's medical history—from pregnancy to the present day. While this might sound daunting, I will personally guide you through the process. Our discussions will focus on key aspects of daily life that often impact children with AS, including:

- Sleep
- Constipation
- Anxiety and behaviors
- School and learning challenges
- Epilepsy and movement disorders(tremor, dystonia, myoclonus, etc.)

We conclude each visit by addressing any additional concerns you may have.Your first visit will typically last an hour, while follow-up appointments range from 20 to 45 minutes, depending on your child's needs.

Because our clinic was built for our local community, we prioritize efficient visits rather than overwhelming families with multiple specialists in a single day. If specialized care is needed, we arrange referrals to the appropriate experts. Genetic confirmation of AS is required before the first visit, and families should provide a genetic report from the laboratory that confirmed the diagnosis.

When scheduling your first appointment, you will be contacted by Monique Washington, RN, BSN, our AS Clinic nurse coordinator, who will help prepare you for your visit. Monique will also serve as your primary point of contact for future communication.

To date, our clinic has enrolled over 60 families, with 90% living within a 200-mile radius of Kansas City. We also have a Patient and Family Advisory Council (PFAC) composed of AS parents, who play a crucial role in shaping clinic services and research initiatives.

Our commitment to Angelman syndrome research

From the beginning, we envisioned the AS Clinic as more than just a medical home — we set out to build a regional center of excellence for AS research. Our research program focuses on two key areas:

- Advancing our understanding of the underlying causes of AS.
- Developing clinical research projects that improve the lives of children with AS.

We currently have several ongoing research projects, which I will share in more detail later. During your first visit, you will meet Eunice Ermovick, our AS Clinic research assistant, who will introduce you to our active studies and invite you to participate in those that are relevant to your family.

Looking ahead

Our clinic has grown tremendously in a short time, and we remain committed to expanding care, advancing research and building a strong community for families affected by Angelman syndrome. We are honored to walk this journey with you and look forward to continuing our mission of improving the lives of children with AS.

Angelman Syndrome Clinic Staff

Dr. J.B. Le Pichon

Physician

Dr. Jean-Baptiste Le Pichon is a pediatric neurologist, researcher, and educator with expertise in child neurology, epilepsy, and neurogenetics. He serves as a Professor of Pediatrics at the University of Missouri-Kansas City and holds the Madison Lauren Sargent Endowed Professorship in Neurology/Angelman Syndrome at Children's Mercy Kansas City.

In January 2022, Dr. Le Pichon was approached by Patrick and Laura Sargent, whose daughter Maddie has Angelman Syndrome (AS). Recognizing the critical need for specialized services in AS, together, they launched the Angelman Syndrome Clinic at Children's Mercy in December 2022, ensuring that families no longer had to travel long distances for treatment. Since its opening, the clinic has grown rapidly and now serves over 60 families.

Under Dr. Le Pichon's leadership, the clinic has gained national recognition, with mentions at the Child Neurology Society annual meeting (November 2024) and an invitation to present at the ASF meeting in 2025. His commitment to groundbreaking research is evident in the several collaborations with the Children's Mercy Research Institute (CMRI) and other centers with AS expertise in the USA. Examples of such initiatives include Emily Farrow's (CMRI) development of a low-cost genetic test for AS, supported by a FAST grant, and Scott Younger's (CMRI) work on organoid cell lines for personalized therapeutics, funded by Maddie's Mission. The clinic is also a key site for clinical trials, including two Phase 3 Antisense Oligonucleotide (ASO) Studies, which will offer cutting-edge treatment to AS patients.

A dedicated educator, mentor, and researcher, Dr. Le Pichon has received numerous teaching and research awards and has been instrumental in advancing pediatric neurology. His vision and leadership have positioned Children's Mercy as a national leader in AS research, treatment, and advocacy, ensuring that families affected by Angelman Syndrome receive the care and support they deserve.

Angelman Syndrome Clinic Staff

Monique Washington

Clinic nurse

Monique Washington is a dedicated pediatric nurse who has worked within Neurology for 11 years. She is very passionate about providing compassionate care to children and families that will have a lasting impact on their daily lives. Outside of work, she is a true Chiefs fan! She enjoys taking naps, cooking delicious meals and cherishing time with her family.

Eunice Ermovick

Research assistant

Eunice is your dedicated Research Assistant for the Angelman Syndrome Clinic, where she supports ongoing studies in neurology by contributing to data collection, analysis, and patient interactions. With a keen interest in advancing Angelman Syndrome research, she plays a vital role in helping the team further the clinic's mission in being research-forward and improving patient outcomes. Outside of work, Eunice enjoys spending time with her daughter Grace (almost 3 years), her 2 dogs (5 years and 16 years), and her husband Logan.

My Angelman Syndrome Clinic Appointment

You can use this page to help follow your journey through the Angelman Syndrome Clinic.

1

Arrive at the Angelman Syndrome Clinic at Children's Mercy

Plan for enough time to park and check in for your appointment and complete any necessary forms.

2

A care assistant will come get you from the waiting area.

They will measure your blood pressure, height and weight, and review your medical record.

3

Then you will go to an exam room and meet your AS team members:

- AS Clinic nurse
- Research assistant
- AS doctor

4

Use the iPad we provide to complete the Angelman Syndrome Clinic Survey and enroll in the Ladder Network global database for AS.

5

You may also have appointments with Angelman syndrome sub-specialists, including:

- Physical Therapy
- Occupational Therapy
- Speech Therapy
- Gastroenterology
- Ophthalmology
- Rehab Medicine
- Developmental & Behavioral Health
- Genetics

What to bring to your appointment

- Medications
- Equipment
- Assistive devices (AAC, orthotics, glasses)
- Snacks
- Toys/child's preferred items

QUESTIONS FOR YOUR VISIT

Examples: Sleep concerns, communication assistance, seizure concerns, clinical trial questions

1

2

3

4

5

6

NOTES FROM YOUR VISIT

Medication Updates or Changes:

Sleep Recommendations:

Seizure Changes and Management:

Symptom Management:

Therapy Recommendations or Changes (communication, fine/gross motor needs):

Research Discussion or Recommendations:

Please remember to complete the Angelman Syndrome Clinic Survey and enroll in the Ladder Database!

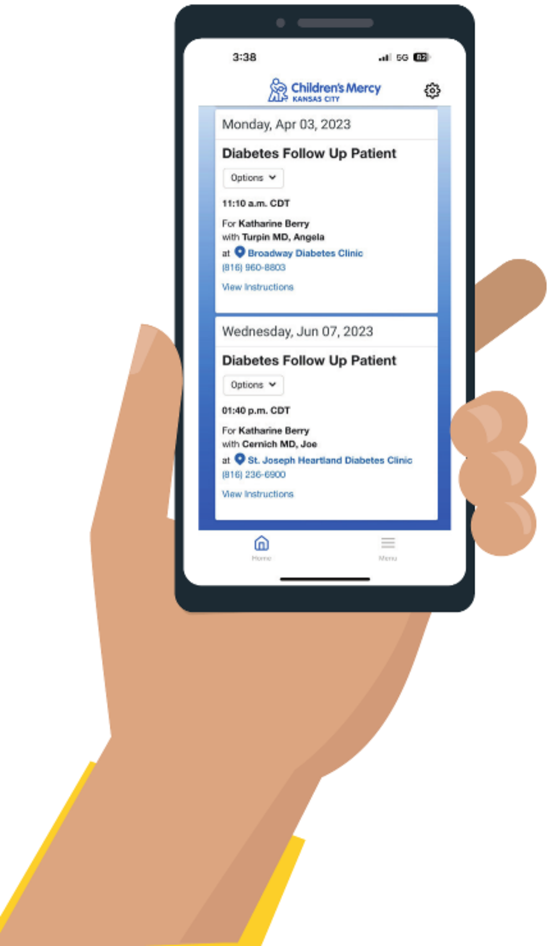
Join MyChildrensMercy Patient Portal

Not Enrolled?

You can now sign up online! Scan the QR code to get started.

The MyChildrensMercy patient portal provides you access to your child's health information 24 hours a day, 7 days a week. Additionally, you will be able to:

1. Reschedule or cancel an appointment.
2. View lab results.
3. Message your care providers.
4. View medication list.
5. Download visit summaries.
6. See immunization history.



Monday, Apr 03, 2023

Diabetes Follow Up Patient

11:10 a.m. CDT

For **Katharine Berry**
with **Turpin MD, Angela**
at **Broadway Diabetes Clinic**
(816) 960-8803

View Instructions

Wednesday, Jun 07, 2023

Diabetes Follow Up Patient

01:40 p.m. CDT

For **Katharine Berry**
with **Cernich MD, Joe**
at **St. Joseph Heartland Diabetes Clinic**
(816) 236-6900

View Instructions

Ask any of our staff or your care team for more information,or go to **cmkc.link/PortalFAQ**

Technical Questions?

Support is available 24 hours a day at (877) 621-8014.

Access Questions?

Contact Children's Mercy Health Information Management Department at **mychildrensmercy@cmh.edu.**

patient portal

GET IT ON Google Play

Download on the App Store

Access the patient portal anytime.

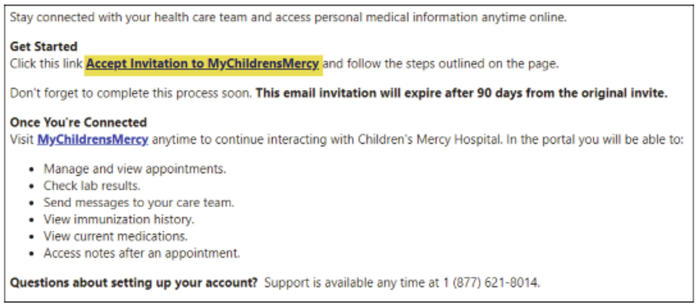
Once enrolled, you can access the MyChildrensMercy app.

Children's Mercy

MyChildrensMercy Patient Portal: Account Set-up

You will receive an invitation after the authorization form has been completed. The email usually comes to you in 48 business hours.

1. Within your email invitation, click "Accept Invitation to MyChildrensMercy."



Stay connected with your health care team and access personal medical information anytime online.

Get Started
Click this link **Accept Invitation to MyChildrensMercy** and follow the steps outlined on the page.

Don't forget to complete this process soon. **This email invitation will expire after 90 days from the original invite.**

Once You're Connected
Visit **MyChildrensMercy** anytime to continue interacting with Children's Mercy Hospital. In the portal you will be able to:

- Manage and view appointments.
- Check lab results.
- Send messages to your care team.
- View immunization history.
- View current medications.
- Access notes after an appointment.

Questions about setting up your account? Support is available any time at 1 (877) 621-8014.

2. Enter your child's date of birth and the answer to your security question. If you are a teen creating your own account, you will use your own date of birth.

Welcome to MyChildrensMercy

MyChildrensMercy is your online connection to Children's Mercy Hospital. MyChildrensMercy is an easy, secure way to log in to view health information online (your child's or – if you are an older patient – your own), all in one central location.

Answer the Security Question

Please enter your **child's date of birth** and provide the answer to your security question.

Questions about setting up your account? Support is available any time at **1 (877) 621-8014.**

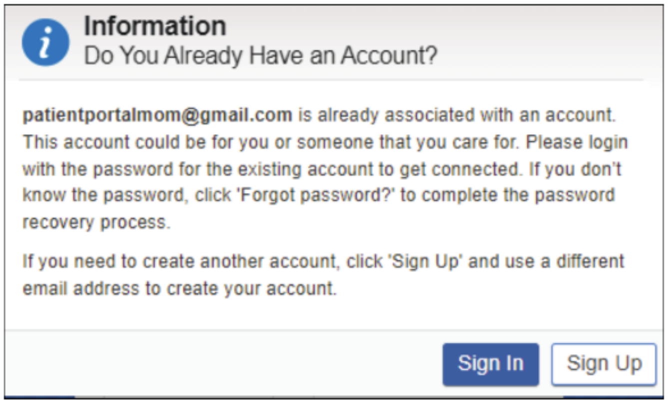
Date of birth
Month November
Day 28
Year 2017
Enter the year as 4 digits.

Your postal code
64108
The answer to this security verification question may have been provided when you were invited to join.

☑ **I agree to the Children's Mercy Hospital Terms of Use and Privacy Policy.**

3. Have you already set-up your account?

YES: If you are adding an additional child, click "Sign In."

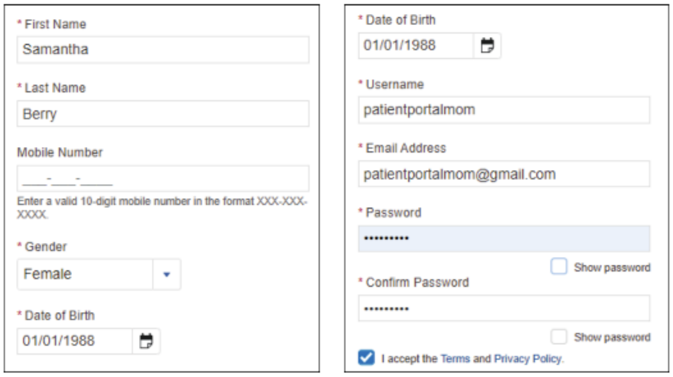


Information
Do You Already Have an Account?

patientportalmom@gmail.com is already associated with an account. This account could be for you or someone that you care for. Please login with the password for the existing account to get connected. If you don't know the password, click 'Forgot password?' to complete the password recovery process.

If you need to create another account, click 'Sign Up' and use a different email address to create your account.

NO: Enter YOUR information to create a new account.

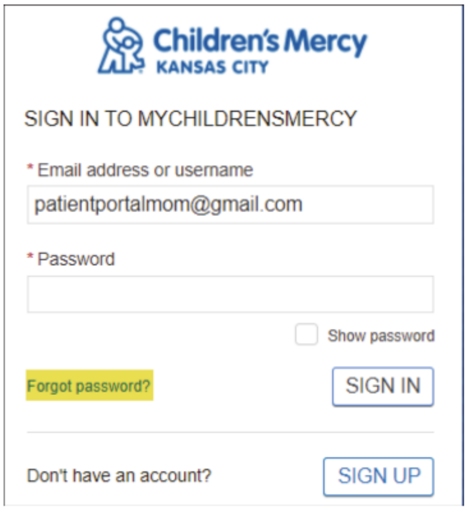


* First Name: Samantha
* Last Name: Berry
Mobile Number: ___-___-____
Enter a valid 10-digit mobile number in the format XXX-XXX-XXXX.
* Gender: Female
* Date of Birth: 01/01/1988

* Date of Birth: 01/01/1988
* Username: patientportalmom
* Email Address: patientportalmom@gmail.com
* Password: •••••••••
☐ Show password
* Confirm Password: •••••••••
☐ Show password
☑ I accept the Terms and Privacy Policy.

MyChildrensMercy Patient Portal: Password Reset

1. To reset your password, select "Forgot password?"



SIGN IN TO MYCHILDRENSMERCY

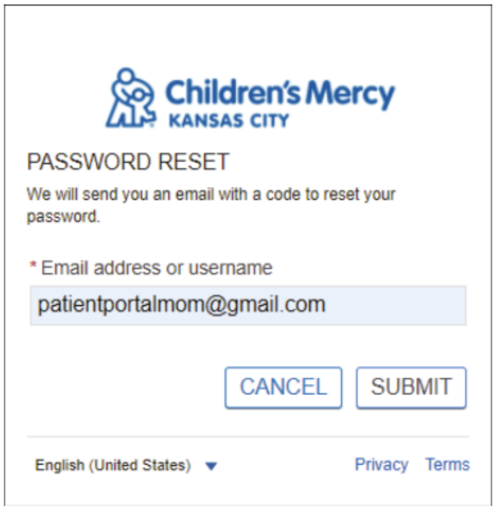
* Email address or username: patientportalmom@gmail.com

* Password

☐ Show password

Don't have an account?

2. Enter the email associated with the account.



PASSWORD RESET

We will send you an email with a code to reset your password.

* Email address or username: patientportalmom@gmail.com

3. A code will be sent to your email.

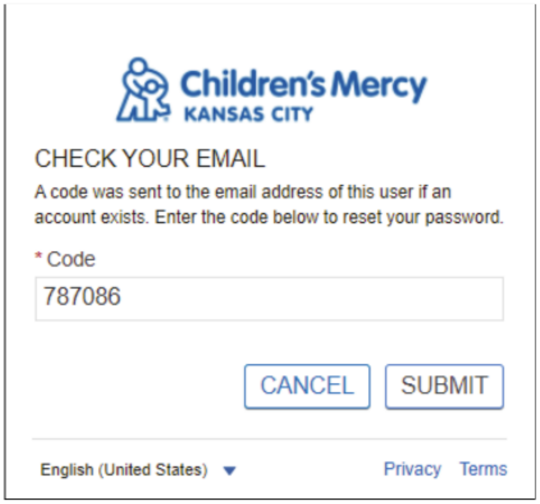
MyChildrensMercy Patient Portal
to me
0 minutes ago

Hello,

You recently requested to change your MyChildrensMercy Patient Portal password. Please enter the security code displayed below onto the verification page to continue:

This code will expire in 20 minutes.
Security Code: 787086

4. Enter the code and select "Submit."

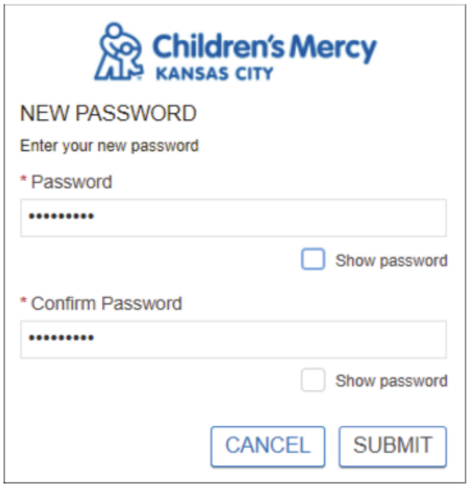


CHECK YOUR EMAIL

A code was sent to the email address of this user if an account exists. Enter the code below to reset your password.

* Code: 787086

5. Enter your new password.



NEW PASSWORD

Enter your new password

* Password: •••••••••

☐ Show password

* Confirm Password: •••••••••

☐ Show password

Children's Mercy

Types of Therapy for Angelman Syndrome

Many of the following therapies are helpful for individuals living with Angelman syndrome. There are local clinics that offer the following therapies. Many of the therapies can also be included in your child's IEP.

1. **Applied Behavioral Therapy:** Applied Behavior Analysis (ABA) therapy uses learning theory and behavioral principles to help people improve social skills.
2. **Aquatic Therapy:** Therapy performed in water for exercise, relaxation and rehabilitation.
3. **Hippotherapy (Horse Therapy):** A combination of physical, occupational and speech therapies that use the movements of a horse to provide motor and sensory input.
4. **Music Therapy:** Using music to provide stress relief, build emotional health, improve social skills, etc.
5. **Nutrition Therapy:** Helps ensure proper nutrition for growth and development.
6. **Occupational Therapy:** Aimed at improving skills needed to perform daily activities, including fine motor movements (such as self-feeding, finger isolation, etc).
7. **Physical Therapy:** Aimed at working specific parts of the body to build, improve, and/or restore function, including large motor skills (sitting, crawling, walking).
8. **Speech Therapy:** Helps individuals communicate in ways that work best for them. Also assists in feeding and swallowing.

Children's Mercy

Offered at Children's Mercy Kansas City

Child Life: Child life specialists support patient and siblings of all ages by:

- Teaching and preparing them for things that happen in the hospital or clinic.
- Teaching and helping children practice coping skills.
- Supporting children during health care procedures.
- Offering play sessions.
- Helping children express their feelings and needs.

POPS: Parents Offering Parent Support (POPS) matches parents with parent volunteers (mentors) who have been through a similar experience with their own children.

Parent and mentor interaction can happen through email, phone conversations or text messaging.

If you are interested in requesting a POPS mentor or becoming a POPS mentor, email **POPS@cmh.edu** or visit **cmkc.link/pops**.

Social Work: Medical Social Workers provide supportive counseling related to:

- Adjustment to illness.
- Lifestyle changes.
- Grief and bereavement.
- Financial issues.
- Family stressors and other concerns.
- Mental health and substance abuse screening, assessment and intervention.

Angelman Syndrome Resources

Offered Nationally

Angelman Syndrome Foundation: The mission of the Angelman Syndrome Foundation is to advance the awareness and treatment of Angelman syndrome through education and information, research and support for individuals with Angelman syndrome, their families and other concerned parties. We exist to give all of them a reason to smile, with the ultimate goal of finding a cure. More information online at **www.angelman.org**.

Counseling Services: Shannon Barlow is a Licensed Clinical Professional Counselor offering her services at no charge. Contact her at **ShannonbarlowLCPC@gmail.com** or (773) 259-4200.

Foundation for Angelman Syndrome Therapeutics (FAST): FAST is the largest non-governmental funder of Angelman syndrome research. Since 2011, we've committed over $20 million to life-changing research.

Genetic Counselor: Niki Armstrong is a board-certified genetic counselor specializing in rare diseases. Contact her at **niki@cureangelman.org.**

Angelman Syndrome Resources

In the Kansas City area

Early Intervention Evaluation/Information: Contact your county to schedule an evaluation for early intervention services.

Ability KC
3011 Baltimore Ave
Kansas City, MO 64108
Phone: (816) 751-7700

Britain Development
7415 Grandview Street
Shawnee Mission, KS 66204
Phone: (913) 676-2253

Eitas
8511 Hillcrest Rd.
Kansas City, MO 64138
Phone: (816) 363-2000

Kansas City Regional Office
821 E. Admiral Blvd.
Kansas City, MO 64106
Phone: (816) 889-3400

Maddie's Mission Foundation
PO Box 8552
Prairie Village, KS 66208
Phone: (913) 530-6274

Playabilities
8340 Mission Road, Suite B
Prairie Village, KS 66206
Phone: (913) 213-3531

Special Olympics – Kansas City Metro Area
9150 E 41st Terrace, Ste. 213
Kansas City, MO 64133
Phone: (816) 535-9610

University of Kansas Department of Hearing & Speech
Augmentative & Alternative Communication
(AAC) Evaluations and Training
3901 Rainbow Boulevard
Kansas City, KS 66160
Phone: (913) 588-5937

Variety KC
4050 Pennsylvania Ave
Suite 115-77
Kansas City, MO 64111
Phone: (913) 308-4042

Additional State Resources Provided by the ASF: Kansas and Missouri

Genetic Resources

National Library of Medicine Genetics Home Reference

NCBI Genes and Disease

Epilepsy Resources

American Epilepsy Society (AES)
342 North Main Street
West Hartford CT 06117-2507
Phone: (860) 586-7505

Epilepsy Foundation
8301 Professional Place
Landover, MD 20785-7223
Phone: (800) 332-1000 (toll-free)
Email: **info@efa.org**

Clinical Trial Information

ClinicalTrials.gov
Search for "angelman syndrome" to view a list of current/pending federally or privately funded clinical trials involving patients with Angelman syndrome.

Guardianship Information

Recommended Books about Angelman Syndrome/Special Needs Parenting

Stories for Siblings and Angels:

Getting to Know You (Sibling Stories)

Getting to Know You (Making Memories)

Genetic Gems

Good Morning Gus

Stories/Books for Parents:

Angelman Syndrome Demystified

Angelman Syndrome : The Guide To Complete Knowledge About Angelman Syndrome, Treatments And Natural Remedies That Work

See the Hope

.

***The books listed can be found at the Kreamer Resource Center, but supply is not guaranteed.**

Angelman Syndrome Clinic PFAC Members

Rob and Heidi Burns

Rob and Heidi Burns are parents to Keegan (19) and twins Kenley and Kamden (14). With the help of Children's Mercy, Keegan was diagnosed at the the age of two with a non-deletion imprinting center defect consistent with Angelman Syndrome. Heidi has spent 23 years in education as a Reading Specialist, and Rob works for J&J helping patients and providers access the organization's immunology products. Keegan currently attends the Blue Valley 18-21 Year Old Program, and loves to see his teachers and classmates every day. The Burns enjoy spending time watching sports and getting together with friends and family.

Terrence Gallagher

Terrence is an architect and full-time community diverse volunteer at Children's Mercy for the past 15 years. He is the proud father of three girls with his best friend and wife, Diane, who is also highly active in the Children's Mercy family and parent voice as a member of the Children's Mercy Board of Directors. Their love for Children's Mercy grew from the care, support and love of their middle daughter, Elizabeth, who is diagnosed with Angelman Syndrome, an unknown genetic disorder and calcification of the basal ganglia with multiple medical complexities deriving from these.

Ashlyn Johnson

Ashlyn and her husband, Jay, have three wonderful boys. Their oldest, Parker, was born in 2011 and diagnosed at a year and a half with Angelman Syndrome. Parker Dean has taught them everything they know about unconditional love, patience and resilience. In 2021, he was joined by his middle brother, Jameson and then again in 2023 by his little brother, Hudson. They have been a part of the Children's Mercy AS Clinic since April of 2023 and consider themselves incredibly blessed with the staff and care plan. This has been a fantastic opportunity to be a part of the PFAC and share insights from Ashlyn's 10+ years as an educator and the situations their family has experienced through Parker's school journey. Ashlyn is honored to share her story and hopes to help lend support to families, new and experienced.

Angelman Syndrome Clinic PFAC Members

Tasha Martin

Tasha and her husband, Alex, have two wonderful boys. Reis was born with bi-lateral cleft lip and palate in 2009. He has been seen at Children's Mercy since he was born. He is seen in several clinics at CMKC. Our youngest, Lynkin, was born in 2010. In December of 2011, he was diagnosed with Angelman Syndrome. He has been seen at Children's Mercy since he was 14 months old. He is seen in several clinics to help with all the medical needs that come along with AS. Children's Mercy has and will always be a big part of their lives. Tasha is so thankful to be able to join PFAC and help support the hospital and other families!

Laura Sargent

Laura and Patrick are parents to Colin (8yo) and twin girls Kellie & Maddie (6yo). Maddie lives with deletion positive Angelman syndrome. Laura is a former pediatric nurse practitioner at Children's Mercy and now works in her family business and runs Maddie's Mission Foundation. Maddie's Mission Foundation partners with area organizations working to improve the lives of individuals living with rare diseases and special needs. Laura is proud and honored to be part of the Angelman Syndrome PFAC in support of other families and patients at Children's Mercy.

Patrick Sargent

Patrick's journey with Children's Mercy began in 2020 at the Genetics Clinic when his daughter, Maddie, was diagnosed with Angelman syndrome. He believes that Maddie's biggest advocates -- her mom, Laura, her brother, Colin, and sister, Kellie, and the community surrounding them, including CMKC, are why she thrives today. Patrick joined the PFAC as a way to give back to the organization that has been a significant part of his family's journey. He continues to be grateful for the opportunity to advocate for Angelman children and all special needs families and patients.

Angelman Syndrome Clinic PFAC Members

Leah Scott

Leah and her husband Bobby, live in Blue Springs with their sons, Caden and Sawyer. Caden was diagnosed with Angelman Syndrome in January 2020 after missing several early developmental milestones, but Children's Mercy has been with them every step of the way. They are "frequent fliers" of several different clinics at CMKC, and are thrilled to now have an Angelman Syndrome Clinic here in Kansas City! Leah is excited to be a part of the Angelman PFAC and looks forward to advocating for other Angelman families who make this fantastic clinic their medical home.

Helal Shah

Helal Shah and Manish Mistry are parents to Jian (5yo) and Vira (3yo). Vira has Uniparental Disomy (UPD) Angelman Syndrome and was diagnosed at the age of 2. She looks up to her big brother Jian, often imitating his every move. Despite the challenges of her condition, Vira refuses to be defined by it, embodying strength and perseverance every day.

Children's Mercy