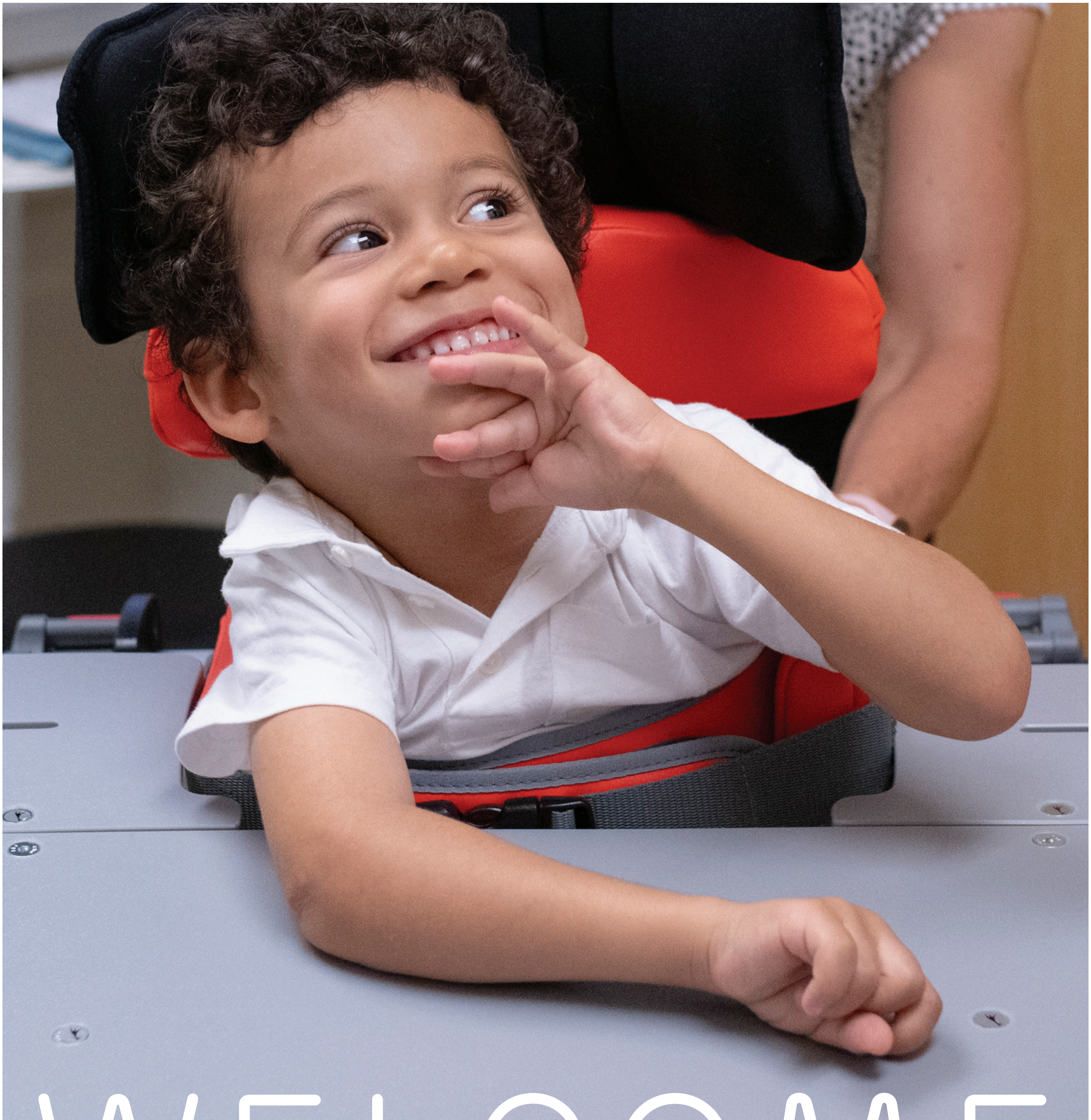

WELCOME

Children's Hospital of Philadelphia®
Leukodystrophy Center

OUTPATIENT CLINIC

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Dear Patients and Families,

Thank you for choosing the Leukodystrophy Center at Children's Hospital of Philadelphia (CHOP). Our team considers it a privilege to serve you and your family.

Our program is dedicated to developing and providing the most advanced treatments for infants, children, adolescents and adults affected by leukodystrophies, a group of inherited diseases that affect the white matter of the brain. We created our program because we recognize that patients with these conditions need specialized and coordinated care. Our center supports families by providing efficient, comprehensive care focused on improving health and decreasing the burden of disease.

For children diagnosed with leukodystrophy prior to the onset of symptoms, possibly through newborn screening, we offer access to potentially curative therapies. Through our research program, we offer families the opportunity to participate in activities that will help us develop new therapies. Our long-term goal is to create standardized ways to treat leukodystrophy, like bone marrow transplants, stem cell transplants and gene therapy.

If a child doesn't have a confirmed diagnosis, our specialized undiagnosed leukodystrophy clinic can use cutting-edge genetic technology to try to find the cause of their symptoms. For children, teens and adults already living with the complex challenges of a leukodystrophy, we offer state-of-the-art multidisciplinary care. This includes complex care pediatricians, rehabilitation physicians, endocrinologists, physical therapists, occupational therapists, speech therapists, dietitians and genetic counselors.

We know how hard it is to care for a child with complex medical needs. We are here to support you and ease these difficulties any way we can. Again, thank you for partnering with our group to care for your child and your family.

Sincerely,

Adeline Vanderver, MD
Program Director, Leukodystrophy Center

WHO YOU'LL MEET

Care at the Leukodystrophy Center is provided by a team of specialized clinicians from many different departments at Children's Hospital of Philadelphia.

Neurologist: This attending physician is the main doctor in charge of your child's medical care. They have special training and understanding in brain, spinal cord, nerve and muscle problems. During a clinical appointment, they will examine your child and carefully review your child's medical history and test results. They will work with the rest of the medical team to develop a detailed care plan.

Complex Care Clinicians: These are pediatricians and certified registered nurse practitioners who specialize in treating children with complex medical needs. They help coordinate all the different parts of your child's medical care. These clinicians will partner with you and your child's local medical providers to make sure your child gets the medications and care they need.

Rehabilitation Physiatrist: This is a doctor trained in Physical Medicine and Rehabilitation. They are experts in improving movement and quality of life for patients with physical difficulties or disabilities caused by problems in their brain, spinal cord, nerves, bones, joints, ligaments, muscles and tendons. Many children with leukodystrophy have issues with muscle tone, ranging from floppiness to stiffness. Rehabilitation physiatrists are trained to help with muscle tone and mobility. They will try to make your child as independent as possible for everyday tasks and improve their quality of life by giving them the right medicines, selecting the best rehabilitation intervention strategies, and getting them supportive equipment like braces or splints.

Endocrinologist: This is a doctor with special training in helping patients with adrenal and bone health issues. Endocrinologists are very important for patients diagnosed with a rare disease called X-linked adrenoleukodystrophy and/or those who cannot do weight-bearing activities, like walking and running. When appropriate, our endocrinologist can recommend lifestyle changes (like changing diet, exercise and daily habits) as well as medications to treat adrenal or bone problems.

Genetic Counselor: Our genetic counselor plays an important role in diagnosing leukodystrophies by ordering the necessary genetic tests and interpreting and explaining the test results. They also provide support and education to you and your child. They explain what the genetic test results mean, which can help you make informed choices about your child's health. Our genetic counselor can help you understand and cope with the challenges of this difficult diagnosis.

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Physical Therapists (PT): Our physical therapists use exercise and fun activities to help kids and young adults with movement, coordination, balance, strength and endurance. They will do a full evaluation of your child's gross motor skills (head control, sitting, crawling, walking, running) to come up with an appropriate treatment plan. Our therapists work closely with you and your child's school and community to help your child become more independent in their daily activities. If appropriate, our PTs will also recommend supportive equipment to help your child, like wheelchairs, walkers or braces.

Occupational Therapists (OT): Our occupational therapists work with children and young adults who are dealing with the social, emotional and physical challenges of leukodystrophy. Our OTs will do a complete evaluation of your child to assess their fine motor skills (how well they can use their hands and fingers), visual motor skills (how well they can use their eyes and hands together), and how they perform daily activities like dressing or eating. They will then work with your child to increase their participation in everyday activities like play, sports and going to school. If appropriate, our OTs will also recommend the right adaptive equipment to help your child, such as hand splints, bath chairs and positioning chairs.

Dietitian: Our clinical dietitian helps our patients with leukodystrophy by improving their nutrition status. This helps your child grow properly and stay healthy. Our dietitian will work with your family to create an individualized nutrition plan that can be adjusted as your child's needs change. They can support patients that eat food by mouth as well as those who get nutrition through a feeding tube.

Speech-Language Pathologist (SLP): Our speech-language pathologist helps children with eating difficulties. Children with leukodystrophy may have trouble with breastfeeding, bottle feeding, eating solid foods, chewing and swallowing. These can cause coughing, choking, poor nutrition, weight loss and other health issues. If appropriate, our SLP will evaluate how your child swallows by watching them eat and drink. They will then determine if your child needs additional testing or if your child would benefit from changes to their diet to increase safety when eating and drinking. Our SLP can also answer any questions you have about your child's ability to communicate.

Social Worker: Our social worker is here to help you and your child cope with the difficulties of having an ongoing illness. Our social worker is available to help answer your questions and solve any issues by providing support, information and education. They can help assess the emotional and social needs of your child and your family, provide supportive counseling, and connect you to resources in our hospital or the community that may be helpful.

Child Life Specialists: Child life specialists help patients, siblings and families by supporting them and educating them throughout their medical journey. When appropriate, a child life specialist can talk to your child and/or their siblings. They can explain your child's health issues in a way kids can understand, based on their age. This can help decrease feelings of anxiety and help children find positive ways to cope with illness.

TWO CLINICS, ONE GOAL

The Leukodystrophy Center at CHOP is proud to offer two clinics to support families.

Multidisciplinary Clinic: This clinic happens every Thursday. It is designed for all leukodystrophy patients with complex medical needs, whether they have a specific diagnosis already or not. All the specialists mentioned on pages 3 and 4 are usually available to see any patient who needs them.

Undiagnosed Clinic: This clinic happens on the second Tuesday of each month. It was created for individuals who might have signs of leukodystrophy, but who do not have a specific diagnosis. In this clinic, a neurologist and genetic counselor will meet with you and your child. They will review your child's medical history with the goal of identifying a diagnosis. If needed, the clinic can also connect your family to other diagnostic resources at CHOP.

BEFORE YOUR VISIT

Registration and Scheduling: After you fill out our Online Referral Form, one of our coordinators will contact your family. They will introduce our program, get basic information from you so we can gather your child's medical records and explain the next steps in our process. After this first call, one of our scheduling coordinators will help register your child in our electronic health record system, which is called Epic. They will also work with you to schedule your child's first appointment with us.

PLEASE NOTE: We cannot confirm your child's appointment with the Leukodystrophy Center until we receive approval from your insurance company. Please wait to make any travel plans or book a hotel room until the appointment is officially confirmed. You can find more details about insurance approval in the section that follows called "Insurance Authorization and Payment."

Medical Record Collection: It is very important that our medical team has all of your child's medical records before their appointment. This is true whether your child is a new patient seeing us for the first time, or they are returning for a regular checkup. We review medical records before appointments and use these records, as well as information we get from talking to you and your family during the intake call (see below), to make a plan for your child's care. Medical record collection helps us provide the best possible care to your child and increases the amount of time we will have during the appointment to answer any questions and concerns you have.

Key medical records we need include notes from previous doctor's visits, reports from imaging tests, and results from any genetic tests or other diagnostic tests that have already been done.

Intake Call: Shortly before your child's appointment, one of our team members will call you to complete an intake call. This call helps our team prepare for your child's visit by giving us more information about your family's goals. It also helps us learn more about your child's medical history. If your child is scheduled for our multidisciplinary clinic, we will also discuss the different types of doctors and specialists you would like your child to see during the appointment.

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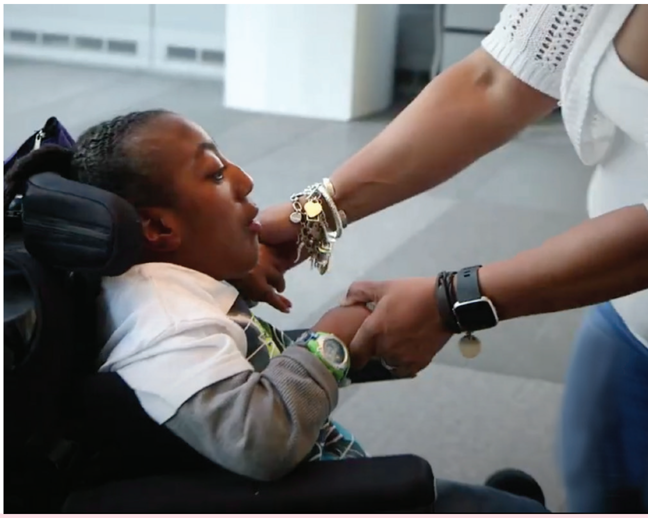
Insurance Authorization and Payment: During registration, our scheduling coordinator will ask for your insurance information. Prior to your child's appointment, our financial team will try to get insurance coverage for the appointment. It is also a good idea to contact your insurance company before making an appointment to see what services they will cover. That way, you'll know how much you might have to pay. If you need to see more than one doctor during your visit, your insurance may require you to pay more than one co-pay. Please note that you will need to pay for any costs from the visit that your insurance does not cover.

If you have Medicaid insurance from another state, it may be helpful to get a letter from your local doctor explaining why you need to see a doctor in Pennsylvania. For example, the letter could say that our Leukodystrophy Center has special expertise you can't find locally. Our financial counselor can give you a sample letter to help your local doctor, if requested.

PLEASE NOTE: If you live outside of Pennsylvania, getting your Medicaid approved to cover care at CHOP can take several months, depending on your plan. Please contact your insurance company as soon as possible to ask them about your coverage options, even if you do not have an appointment scheduled with us yet. Our financial team is here to help every family get their insurance approved for the care they need. Feel free to reach out to our financial team if you need help.



Insurance Codes:

DIAGNOSIS CODE for all procedure codes is: E75.29
PROCEDURE CODE for all physicians:
New Office Codes: 99205, 99245
Follow-up Office Codes: 99213, 99214, 99215

Physician Name (Specialty)

- Dr. Adeline Vanderver (Neurologist) // NPI Code 1578640827
- Dr. Amy Waldman (Neurologist) // NPI Code 1205991361
- Dr. Laura Adang (Neurologist) // NPI Code 1558591867
- Dr. Mariko Bennett (Neurologist) // NPI Code 1184117269
- Dr. Sarah Evans (Rehabilitation Physiatrist) // NPI Code 1184707390
- Dr. Nicole Jaffe (Complex Care Pediatrician) // NPI Code 1336451699
- Dr. Jeffrey Roizen (Endocrinologist) // NPI Code 1891951430

Other Specialties:

- Nutrition // NPI Code 1215921457 // Procedure Code 97802, 97803
- Speech Therapy // NPI Code 1215921457 // Procedure Code 92522, 92610
- Physical Therapy // NPI Code 1215921457 // Procedure Code 97161, 97162, 97163, 97164
- Occupational Therapy // NPI Code 1215921457 // Procedure Code 97165, 97166, 97167, 97168

RESEARCH PROGRAM

The Leukodystrophy Center conducts research to better understand the different types of leukodystrophy, improve how they are diagnosed, and create groundbreaking new therapies. All patients and families who visit the Leukodystrophy Center will have a chance to talk to an experienced research coordinator to learn about opportunities to participate in research studies.

The Leukodystrophy Center's research studies fall into the following categories.

Biobanking and Data Collection: The Leukodystrophy Center has one of the world's largest collections of medical records and biological samples from individuals with leukodystrophy. This collection is called the "Myelin Disorders Biorepository Project." Research on this collection has led to groundbreaking discoveries about these disorders. It has helped us make diagnostic testing faster and more accurate. It has also paved the way for new clinical trials and approved treatments. Individuals enrolled in the research biorepository can choose to participate in different activities, many of which can be done without having to come to CHOP for an extra visit. If you want to learn more about getting involved, please speak to one of our clinical research coordinators.

Natural History Studies: The Leukodystrophy Center leads several international natural history studies. A natural history study looks at how a medical condition develops and changes over time. Knowledge about how a disease progresses is very important because it helps our physicians decide the best time for medical treatments and gives families a better idea of what to expect for their child's future. It also helps to inform clinical trials and development of new treatments.

Natural history studies are currently available for the conditions listed below.

PLEASE NOTE: Your child does not need to be diagnosed with one of these conditions in order to participate in our research program.

- Adrenoleukodystrophy (ALD)
- Aicardi-Goutières syndrome (AGS)
- Alexander disease (AxD)
- Autosomal dominant leukodystrophy with autonomic disease (ADLD)
- Beta-propeller protein-associated neurodegeneration (BPAN)
- Krabbe disease
- Metachromatic leukodystrophy (MLD)
- Multiple sulfatase deficiency (MSD)
- Pelizaeus-Merzbacher disease (PMD)
- Pelizaeus-Merzbacher-like disease (PMLD)
- Pol III-related leukodystrophy (4H)
- TUBB4A-related leukodystrophy (incl. H-ABC)
- Vanishing white matter disease (VWM)

Clinical Trials: The Leukodystrophy Center has completed clinical trials for several types of leukodystrophy. Clinical trials are research studies that test whether new medications or treatments are safe and work well in individuals with a specific medical condition. If you are interested in learning about available leukodystrophy clinical trials happening at CHOP, please talk to one of our doctors or research coordinators.

FREQUENTLY ASKED QUESTIONS

Do you have resources for finding affordable places to stay or ways to travel when we come for an appointment?

Many families come from far away to get care for their child at CHOP. This can be very expensive, which we know can be hard. Our social worker can help answer any questions you have about affordable housing and travel for your child's appointment at our Leukodystrophy Center. Our social worker's contact information can be found at the end of this packet.

How long are appointments?

The length of your child's appointment will depend on your child's needs and how many specialists they are scheduled to see. On average, appointments in our Multidisciplinary Clinic last three to four hours, but they can be up to eight hours long. Each appointment with one specialist is expected to be 45 to 60 minutes. Appointments in our Undiagnosed Clinic are two to three hours long. We encourage you to bring things to keep you and your child comfortable during the appointment, including toys, activities and food.

What time will the appointment start?

Your child's appointment at the Leukodystrophy Center's Multidisciplinary Clinic will be scheduled at 8 a.m. as a placeholder time.* However, the actual appointment time will not be set until the week before the appointment date. This is because the clinic has a complex schedule with many different doctors and specialists. We will confirm your exact appointment time one week before the scheduled date by sending you a message through MyCHOP, our online patient health portal.

When your appointment is initially scheduled, please let us know if you need the appointment to start or end by a specific time.

**This does not apply to appointments in our Undiagnosed Clinic, which are always scheduled at the correct appointment time.*

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What will the schedule for my child's appointment look like?

Below is an example of the schedule for an appointment with our Multidisciplinary Clinic.

9:00 – 10:00	Neurology & Genetic Counseling
10:00 – 10:45	Complex Care Pediatrics
10:45 – 11:30	Physical Therapy Assessment
11:30 – 12:15	Occupational Therapy Assessment
12:15 – 1:00	Physical Medicine and Rehabilitation
1:00 – 1:45	Nutrition Counseling
1:45 – 2:30	Swallow Evaluation
2:30 – 3:00	Endocrinology
3:00 – 3:15	Social Work Visit
3:15 – 4:00	Clinical/Research Lab Work

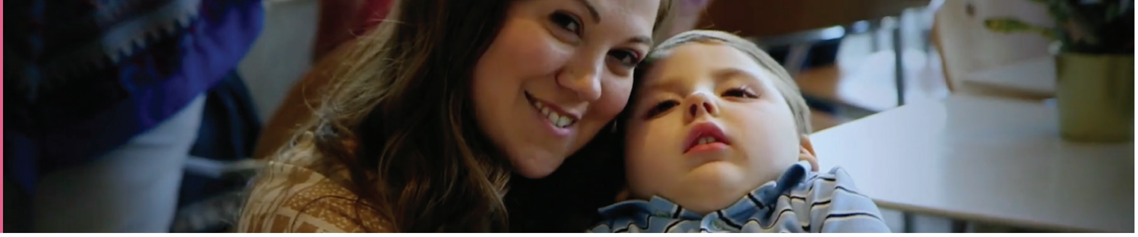
PLEASE NOTE: This is a sample schedule to show how long an appointment might take with our Multidisciplinary Clinic. In this example, the child is scheduled with every provider on our team. Your child may see fewer providers than this and appointments may vary in length, depending on your child's unique needs and symptoms. The order of providers will be random on the day of your visit. Please be patient, as there will likely be wait times between each provider. We know this can be a long and tiring day and encourage your family to request lunch breaks, if needed. We encourage open communication between your family and our team during your child's appointment, and we are happy to accommodate any of your needs that day.

What should I bring to the visit?

- ☐ Appointment information (doctor's name, clinic location, etc.)
- ☐ Insurance cards
- ☐ Co-pay(s) if required by insurance
- ☐ Medical or personal records not yet received by our team
- ☐ A list of questions you may have for our team
- ☐ Books, games, snacks, formula, diapers, medications, a change of baby clothes or other necessities (please do not bring food if your child must fast for testing)
- ☐ Any braces, splints or orthotic devices your child uses, including wheelchairs and walkers, in addition to any respiratory equipment

Any family members or friends who would like to attend the visit with you are welcome!

CONTACT US

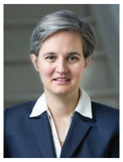


Leukodystrophy Center Outpatient Clinic

Buerger Center for Advanced Pediatric Care, 10th Floor
Children's Hospital of Philadelphia
3500 Civic Center Blvd., Philadelphia, PA 19104

- **Scheduling Coordinator**
  Melanie Fernandez
  Phone: 215-590-6053
- **Financial Counselor/Authorizations**
  Email: NeurologyAuthRequests@chop.edu
- **Social Worker**
  Sarah Stoney
  Phone: 267-426-9649
  Email: stoneys@chop.edu
- **Division of Neurology**
  Phone: 215-590-1719
- **Leukodystrophy Center Contact Information**
  Email: LCE@chop.edu
  Fax: 215-590-1234

Attending Physicians



Adeline Vanderver, MD
Program Director
Leukodystrophy Center
Attending Neurologist

Amy Waldman, MD
Medical Director
Leukodystrophy Center
Attending Neurologist

Laura Adang, MD, PhD, MSTR
Attending Neurologist

Nicole Jaffe, MD
Complex Care Pediatrician

Sarah Evans, MD
Division Chief
Physical Medicine
and Rehabilitation

Jeffrey Roizen, MD, PhD
Attending Endocrinologist

Mariko Bennett, MD, PhD
Attending Neurologist

Finding Your Way to Us • To learn how to get to the Buerger Center for Advanced Pediatric Care on CHOP's Philadelphia Campus, know where to park, and view maps to help you find your way to your child's appointment or test, scan this code or visit **chop.edu/philadelphia-campus-visit/getting-here**