



HUNTER'S HOPE

NEUROLOGY FAMILY COMPANION

*A practical approach to care of children with
leukodystrophies*

Introduction

Your child has just been diagnosed with leukodystrophy. We know you have lots of questions, and we are here to help! Before you keep reading, there are a few things we would like you to know.

THIS INFORMATION IS NOT A SUBSTITUTE FOR TAKING YOUR CHILD TO THE DOCTOR. Also, the material in this handout was not able to be based upon extensive medical research. Leukodystrophies are a group of **rare** diseases. There is very little research on how to treat basic symptoms in children with a leukodystrophy. We are working on that! While we would like to have lots of research to tell us what to recommend to children and their families, there just isn't very much available. We also know that your child needs care **today**. They cannot wait. **YOU** cannot wait. And so, we have based this handout on the next best thing: Expert advice.

In 2017, a group of parents and medical providers came together to write "A Practical Approach to Care for Children with Leukodystrophies". The group included families of children with a leukodystrophy. It also included doctors and nurses with many years of experience caring for these children.

Together, we wrote clinical guidelines to help medical professionals such as doctors, therapists, and nurses care for children with leukodystrophies. The paper focused on four symptoms that are important to families: Pain, sleep, seizures, and cognition.

The clinical guidelines were designed for medical professionals. **This handout is for YOU.** We hope that it can help you and your family along your journey.

How should I use this handout?

There is a lot of information in this handout! It is designed to help families and children across all stages of all leukodystrophies. In order to help you find the information that is most helpful for you today, look for the color-coded symbols listed below. The Family Companion is always here waiting when you are ready for more information.

Key to find information fitting your needs today:



General Information: Important for everyone.



Early Diagnosis Leukodystrophy: No symptoms/mild symptoms



Symptomatic Leukodystrophy: symptoms are impacting most or all parts of my child's day to day activities.

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Family Companion: Leukodystrophy

What is leukodystrophy?

Leukodystrophies are a group of genetic diseases that cause damage to the white matter of the brain and sometimes spinal cord. There are over 60 types of leukodystrophy. Each type is caused by a different gene. Scientists haven't found all the genes responsible for many of the types, so there are many children who are diagnosed with "unknown leukodystrophy." These children have an abnormal MRI and symptoms of leukodystrophy, but no identified genetic cause.



What is white matter?

The brain is made up of **grey matter** and **white matter**. People often describe the white matter as insulation around an electrical wire. When the white matter is healthy, messages move very quickly from the brain to the rest of the body. When it is damaged, messages are slowed down or may not get there at all.

Grey Matter

- Made up of neurons, the actual cells of the brain
- Creates electrical energy to send messages to the rest of the body
- Forms the outside layer of the brain

White Matter

- Made up of "myelin," a fatty substance
- Coats and protects the neurons (brain cells)
- Helps messages to move quickly along the nerves
- Located in the center of the brain

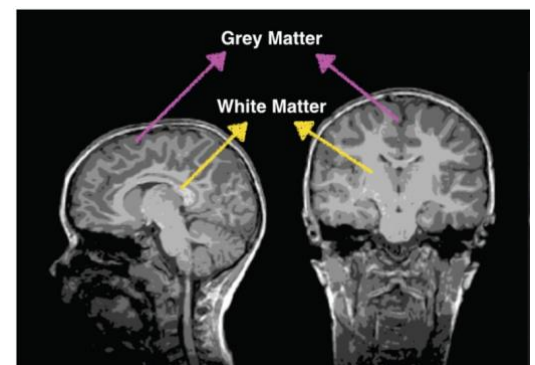


Photo from "frontiers for Young Minds", kids.frontiersin.org/article/10.3389/frym.2014.00019

How do you get leukodystrophy?

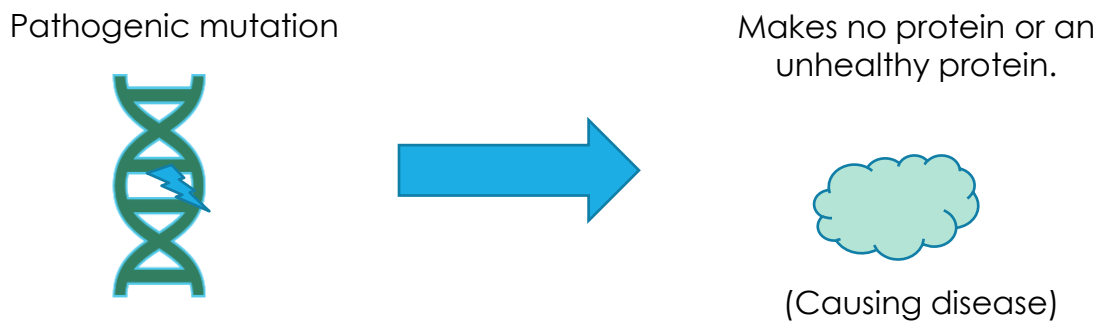
Leukodystrophies are genetic disorders. They are not contagious. You cannot “catch” leukodystrophy. It is also important to remember that people do not get to pick their genes. Just as you did not decide if your child was a boy or a girl, you did not do anything that gave your child leukodystrophy.

What is a genetic disorder?

We are all born with thousands of genes. Together, these genes make up the DNA code for who we are. You can think of your DNA as a library full of books. Each book is a chromosome. You have a total of 46 chromosomes, 23 from your mother and 23 from your father.



Each chromosome holds many genes. A gene is like a chapter within a larger book. A genetic mutation is a specific spelling error in that gene. We all have many little mutations, or spelling errors. Most of those errors do not cause any problem. The body can still read the chapter. Sometimes, though, there is a major error that changes the meaning of the entire chapter. These are called “pathogenic mutations.” Pathogenic mutations are the cause of genetic diseases such as leukodystrophies.



Is there a cure for leukodystrophy?

There is no cure for leukodystrophy. But there are lots of things that can be done to treat its symptoms. In order to do that you will need a team!

Who should be included on my child's treatment team?

Many families do not live near a Leukodystrophy Care Network (LCN) center.

That's okay! If you do not live nearby, you will need to find a local medical team to work with your LCN center. Your local care team doesn't need to know a lot about leukodystrophy, but they should be willing to learn. Your child's teachers, friends, family, and school/home-based therapy providers are also important members of the care team.

Finally, we recommend adding a palliative care expert to your team. Palliative care focuses on caring for children with a chronic illness. Many families have reported that having a palliative care team provides reassurance, support and information about the future.



How can I help my local care team?

- ✓ Give your school and local care team the Hunter's Hope website
 - <https://www.huntershope.org/>
- ✓ Give your providers a copy of the LCN's Medical Approach Paper
- ✓ Ask your doctor(s) if they are open to talking with your LCN team



Family Companion: Sleep

Why is sleep so important for my child?

Sleep is important for everyone! It is especially important for children as they are growing. Research shows that problems with sleep are common among many children. If your child is having trouble with sleep, it may have nothing to do with leukodystrophy.

What are the side effects of poor sleep?



- Feeling tired during the day
- Difficulty paying attention and staying alert
- Difficulty learning new skills
- Grouchiness and behavior problems

How much sleep does my child need?

Because each child is different, there is no set amount of sleep that a child will need. Here are some general recommendations for healthy amounts of sleep.

Age	Total Number of Hours Required	Should Not Sleep Less Than	Should Not Sleep More Than	Nighttime Sleep	Daytime Sleep	Number of Naps
Newborn	16	11	17	Varied	Varied	Varied
3 months	13	10	15	8 ½	4 ½	3-4
6 months	12 ½	10	15	9 ¼	3 ¼	2-3
1 year	11 ¾	9	14	9 ¼ - 10 ¼	1 ½ - 2 ½	1-2
2 years	11 ½	9	14	9 ¾	1 ½	1
3 years	11 ¼	8	13	9 ¾ - 11 ¼	0 - 1 ½	0-1
4 years	11	8	13	10-11	0-1	0-1
5 years	11	8	13	11	0	0
6 years	10 ½	7	11	10 ½	0	0
10 years	10	7	11	10	0	0
Teens	9 ¼	7	10	9 ¼	0	0
Young Adults	8	6	10	8	0	0

Chart from: National Sleep Foundation, n.d and Boston Children’s Hospital, 2015

My child sleeps enough, so why are they so tired?

The quality of your child's sleep is just as important as how long they sleep. If your child is falling asleep during the day, or difficult to wake up in the morning, s/he may not be getting a good quality of sleep.

What are some common causes of sleep difficulty?

- Poor sleep hygiene/sleep habits
- Behavioral issues - Sleep avoidance or fear around sleep
- Sleep apnea/Obstructive sleep apnea
- Restless leg syndrome

How do I find out why my child is not sleeping well?

Talk to your child's medical team about your child's sleep. You may be asked to complete a sleep survey and/or sleep journal to help the team understand your child's sleep patterns. The doctor may also order tests to evaluate your child's sleep. Common tests include a sleep study (polysomnogram) and/or blood work.

Questions to talk about with your care team:

- ✓ How many hours does your child sleep at night?
- ✓ Does your child snore?
- ✓ Does your child sleep through the night in his/her own bed?
- ✓ Is your child restless at night, kicking during sleep?
- ✓ Does your child go to bed at the same time every night?
- ✓ What is your child's bedtime routine like?
- ✓ Does your child wake up at the same time every day?
- ✓ Can your child fall asleep on his/her own?

What can I do to help my child sleep?

A good first step is practicing good “sleep hygiene”. This includes your child’s nightly bedtime routine, removing unneeded electronics/light sources from your child’s room, and/or increasing the time it takes for you to respond when your child wakes up at night. Some families use weighted blankets or a foam mattress topper to help their children be comfortable at night.

When our son was diagnosed we moved our bedroom to be near him. It was so hard to follow our “normal” sleep routine. What if something happened? Over time we have adjusted our sleep to healthy sleep habits, but it took time.



Sleep Hygiene Basics:

- ✓ Go to sleep at the same time every night
- ✓ Set up regular nightly bedtime routine
- ✓ Turn off electronics 60 minutes before bed
- ✓ Wake up within one hour of the same time every morning
- ✓ Use bed only for sleeping
- ✓ Encourage your child to fall asleep without a parent in bed

Can I give my child melatonin?

Practicing good sleep hygiene is **always** the first step to better sleep. If these measures are not enough, melatonin can be considered. Giving an appropriate dose of melatonin is safe but does not work for all children. Talk to your child’s medical team about the recommended dose, potential benefits and side effects of melatonin for your child.

Family Companion: Sleep

Why is sleep important for my child?

Sleep is important for everyone! It is especially important for children as they are growing. Research shows that problems with sleep are common among many children. Children with symptomatic leukodystrophy can have special sleep needs.

What are the side effects of poor sleep?



- Feeling tired during the day
- Difficulty paying attention and staying alert
- Difficulty learning new skills
- Grouchiness and behavior problems
- Higher risk for seizures
- Increased pain and/or spasticity

How much sleep does my child need?

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Age	Total Number of Hours Required	Should Not Sleep Less Than	Should Not Sleep More Than	Nighttime Sleep	Daytime Sleep	Number of Naps
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3 years	11 ¼	8	13	9 ¾ - 11 ¼	0 - 1 ½	0-1
4 years	11	8	13	10-11	0-1	0-1
5 years	11	8	13	11	0	0
6 years	10 ½	7	11	10 ½	0	0
10 years	10	7	11	10	0	0
Teens	9 ¼	7	10	9 ¼	0	0
Young Adults	8	6	10	8	0	0

Chart from: National Sleep Foundation, n.d and Boston Children's Hospital, 2015

My child sleeps long enough. Why are they still tired?

The quality of your child's sleep is just as important as how long they sleep. If your child is falling asleep during the day, or hard to wake up in the morning, s/he may not be getting a good quality of sleep.



It is also important to think about the medications your child is taking. Many medications that treat seizures, pain, and spasticity can also make children feel tired.

“We were always worried about overmedicating our son and him sleeping too much. We wanted to try to find the right balance between comfort and alertness. Seems like that was particularly difficult very early in our journey and again very late.”



What are some things that can impact my child's sleep?

Children living with leukodystrophy can have the same difficulties with sleep as other children their age. Common problems include:

- Poor sleep hygiene/sleep habits
- Behavioral issues - sleep avoidance or fear surrounding sleep

Additional causes of sleep difficulty in symptomatic children can include:



Medical issues: Obstructive sleep apnea, Restless Leg Syndrome, pain, muscle spasms, GI reflux and/or dysmotility

Overnight care: Feeding, suctioning, medicating, and need for repositioning

Look for creative ways to improve positioning for your child.

“Our son loved when we used weighted neck wraps for positioning. Shaped like cute stuffed animals, scented, and able to be heated, these were a favorite sleep trick”



How do I find out why my child is not sleeping well?

Talk to your child's medical team about your child's sleep. You may be asked to complete a sleep survey and/or sleep journal to help the team understand your child's sleep patterns. The doctor may also order tests to evaluate your child's sleep. Common tests include a sleep study (polysomnogram) and/or blood work.

Questions to talk about with your care team:

- ✓ How many hours does your child sleep at night?
- ✓ Does your child snore?
- ✓ Does your child require overnight feedings?
- ✓ Is your child restless at night or kicking during sleep?
- ✓ Does your child go to bed at the same time every night?
- ✓ What is your child's bedtime routine like?
- ✓ Does your child wake up at the same time every day?
- ✓ Can your child fall asleep on his/her own?
- ✓ Does your child need any medications at night?
- ✓ Does your child have medical equipment with lights at night?
- ✓ Does your child need to be repositioned at night?

How can a sleep journal help?

Before your next appointment, keep a sleep journal for 1-2 weeks. Write down how much your child slept and if it was a good night or a bad night. Think about what was different on days s/he slept well or poorly. Make notes of things like:

- Timing of today's activities (remember to include therapies such as P.T.)
- When was his/her last bowel movement?
- Did the child nap during the day?
- Did the child have any seizures today?
- What else was special and/or different about the day?
- School day or weekend?
- Any medication changes?

What can I do to help my child sleep?



The first step to improving sleep is always to treat any medical issue(s) that may be causing your child to have trouble sleeping. For example, a child could have constipation that is causing stomach pain. Before considering other things to improve sleep, it would be important to get rid of the pain by treating the constipation first.

Along with treating any medical issues, practicing good sleep hygiene is important.

“We used a memory foam mattress topper cut to fit our son. We could get two or three from a queen size topper. We used them on the couch, in bed, at family members’ houses, hotels, hospitals, etc. Super easy and he always had a consistent, comfortable spot to sleep.”



Sleep Hygiene Basics:

- Go to sleep at the same time every night
- Set up regular nightly bedtime routine
- Do something relaxing before bed, such as massage your child
- Remove electronics and bright light sources from your child’s room
- Turn off electronics 60 hours before bedtime
- Wake up within 1 hour of the same time every morning
- Encourage your child to fall asleep without a parent in bed
- Limit overnight feeds when possible



Some children with leukodystrophy have an exaggerated startle reflex. This startle can wake children up from sleep. A weighted blanket and/or white noise can help some children sleep, especially when they have this reflex.

Can I give my child melatonin?

Addressing medical issues and practicing good sleep hygiene are **always** the first steps to better sleep. If these measures are not enough, melatonin can be considered. Giving an appropriate dose of melatonin is safe but does not work for all children. Talk to your child's medical team about the recommended dose, potential benefits and side effects of melatonin for your child. Only a small dose of melatonin is needed. For best results, it should be given at least one hour prior to bedtime. Remember, your child's medical team should approve any over-the-counter supplement prior to use.

Can other medications help?

Because sleep difficulties can be caused by so many factors, the right treatment depends on your child's individual sleep problems. Your child's medical team may prescribe medication if your child is diagnosed with a sleep disorder. You can also talk to the medical team about using a single medication to treat multiple symptoms. It is possible that a medication your child is already taking to manage other medical conditions (such as epilepsy, pain and/or spasms) can also help improve sleep.

Online Resources:

- [American Academy of Pediatrics Sleep Guideline](#)



Family Companion: Seizures

What is a seizure?

A seizure is a sudden, uncontrolled surge in the brain's electrical activity. Seizures can be very dramatic. Other times, they may not be as obvious. Seizures are uncommon in early stage children who do not have other leukodystrophy symptoms.

There are many kinds of seizures. Seizures can be grouped by asking a few questions:

- Where in the brain does the seizure start?
- Is your child awake & alert during the seizure?
- What does the seizure look like?



Where in the brain does the seizure start?

This question is usually answered by doing a type of test called an electroencephalogram (EEG). There are two basic types of seizure:



- **Generalized** - The seizure involves the entire brain (both sides of the brain).



- **Focal** - The seizure only involves one part of the brain. During a focal seizure, children can remain awake.

Note: EEG testing is only recommended when a child has symptoms of a seizure. An EEG is not needed in all children with a leukodystrophy.

How common are seizures in leukodystrophy?

Many children with leukodystrophy develop seizures, most commonly in the advanced stages of the disease. Seizures are rare in children who do not show other symptoms of leukodystrophy. Your child's medical team can help you understand your child's risk for seizures.

Can I prevent seizures?

Unfortunately, it is not possible to prevent 100% of seizures in children with a leukodystrophy. Again, seizures are rare in children who are not experiencing other symptoms but are common in children with advanced disease.

There are many things that can be done to reduce the number of seizures your child is having. Fever, infection, and lack of sleep are some of the things that can make it more likely to have a seizure. You and your child's medical team can work together to reduce seizure risk in your child.

Are seizures dangerous?

Seizures can look scary, but the seizure itself is not usually dangerous. Seizures become dangerous if they last for many minutes at a time. Seizures are also dangerous if they happen during activities like swimming, climbing, or driving. Frequent seizures can make it difficult for a child to learn new skills, participate in school, and sleep. Children having frequent seizures are treated with medication. The medications can help prevent seizures.



How do I know if my child is having a seizure?

Be aware of any unusual changes in your child's behavior or movements. Persistent changes in behavior or repetitive movements *could* be a seizure. Many parents call these episodes their child's "spells." If you have any concerns about your child, it is ALWAYS okay to ask the medical team.

Some symptoms of seizure include:



- Rhythmic Movements
- Sudden change in how alert your child is
- Child is much more tired than usual
- Short periods of staring off and being non-responsive

What do I do if I think my child is having a seizure?

The most important thing is to stay calm and to stay with your child. Most seizures/seizure-like events will stop quickly. Remember that you can always call 9-1-1 if you feel your child needs immediate attention, especially if they stop breathing or their skin starts to change color. Children who have had seizures should have a seizure plan that is specific to their child. Make sure that your child's seizure plan is available to all care providers.

How are seizures diagnosed?

A neurologist often uses electroencephalogram (EEG) testing to diagnose seizures. An EEG measures the pattern of electrical activity on the surface of the brain. The test is done by placing many little wires on the scalp. These wires record the electrical activity. There is no physical risk to an EEG, and it does not hurt.



How are seizures treated?

Your child's neurologist may prescribe anticonvulsant medication, which is taken every day to help prevent future seizures. The doctor may also prescribe an emergency (rescue) medication that can be given during a prolonged seizure.

Online Resources:

- [Epilepsy Foundation](#)



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How are seizures treated?

Your child's neurologist may prescribe anticonvulsant medication, which is taken every day to help prevent future seizures. The doctor may also prescribe an emergency (rescue) medication that can be given during a prolonged seizure. Rescue medication is usually only given during a seizure lasting more than 5 minutes. Alternative treatments for seizures such as CBD and the ketogenic diet have not been studied in children with a leukodystrophy. You should talk with your child's medical team before trying any treatment.



Children with symptomatic leukodystrophy almost always have an “abnormal EEG.” Many children have what we call “slowing.” Slowing is NOT a seizure. Seizures are “epileptiform discharges.”

Is it a seizure or is it leukodystrophy?

Children who have leukodystrophy can have behaviors or movements that look like a seizure. For example, they often have delays in processing speed. *Processing speed is how long the child takes to understand what he/she is seeing, hearing, or feeling. Delayed processing speed can cause a blank pause before the child responds to you.* Many children have extra reflexes such as the startle reflex and other “jerky” movements that can look like a seizure. Taking a video of these events is very helpful to show your child's medical team exactly what is happening. Your phone's video recorder is a perfect way to capture these moments.

A video of a “spell” is worth a million words to your care team!



My child had a seizure. . . now what?

When your child is diagnosed with a seizure disorder or epilepsy, it can feel scary and overwhelming. Families often wonder if they will be able to manage seizures. With time and guidance from your child’s medical team, it will be possible for you to feel comfortable managing seizures. Your child’s medical team will help design an individual treatment plan for your child.

“My daughter had her first seizure at age 3. It was a scary experience that left us feeling worried. We were on edge waiting for the next one. Fortunately, we worked with her team to find the right medications and now her seizures are well managed.”



Can I prevent my child from having more seizures?

Unfortunately, it is not possible to prevent 100% of seizures. Once your child has a first seizure, s/he is at risk for having more. Working together with your medical team, there are many things that can be done to help. Staying on schedule with daily medications is extremely important. Missing doses make it much more likely that your child will have a seizure.

Here are some other things that can increase the risk of seizures:

- ✓ **Missing medication**
- ✓ **Weight gain**
- ✓ **Fever or any infection**
- ✓ **Lack of sleep**
- ✓ **Over stimulation**
- ✓ **Pain**
- ✓ **Constipation**
- ✓ **Change in medication**
- ✓ **Puberty (birth control)**

Quick Tip: A medical alert bracelet or necklace can help make sure everyone knows that your child has seizures in case of an emergency.



Questions to talk about with your care team:

- ✓ What should I do if my child misses a dose of medication?
- ✓ What should I do if my child spits up a dose of seizure medication?
- ✓ What should I do if my child has a seizure?
- ✓ How can I reach you in an emergency?
- ✓ What is the plan if my child has a fever or other infection?

Note: Some providers recommend treating fevers more aggressively in children with a history of seizures. This is because some children are more likely to have a seizure when they have a fever. Ask about ibuprofen/acetaminophen dosing for your child.

Are there any limits on my child's activities?

Talk to your child's neurologist about what is safe for your child. It is usually recommended to use caution until your child has been seizure-free for at least 6 months. Common safety measures include:

- No unsupervised swimming or tub baths
- Showers instead of baths (if the child is old enough to shower)
- Avoid climbing to any significant height
- Avoid allowing child to lock bedroom/bathroom doors



What should I tell others about my child's seizures?

A seizure action plan is recommended for all children who have had a seizure. A seizure plan is a formal document that lists important facts about a child's seizures. Anyone who cares for your child should have a copy of the seizure plan. Many schools require a seizure action plan to be on file with the school nurse before allowing a child to return to school after his/her first seizure. Daycare providers, family members, and any other caregivers should be familiar with your child's individual plan.

EPILEPSY FOUNDATION		Seizure Action Plan	Effective Date
This student is being treated for a seizure disorder. The information below should assist you if a seizure occurs during school hours.			
Student's Name	Date of Birth		
Parent/Guardian	Phone	Cell	
Other Emergency Contact	Phone	Cell	
Treating Physician	Phone		
Significant Medical History			
Seizure Information			
Seizure Type	Length	Frequency	Description
Seizure triggers or warning signs:		Student's response after a seizure:	
Basic First Aid, Care & Comfort		Basic Seizure First Aid	
Please describe basic first aid procedures:		<ul style="list-style-type: none"> Stay calm & track time Keep airway clear Do not restrain Do not put anything in mouth Stop with child until fully conscious Record seizure in log 	
Does student need to leave the classroom after a seizure? <input type="checkbox"/> Yes <input type="checkbox"/> No		For tonic-clonic seizures:	
If YES, describe process for returning student to classroom:		<ul style="list-style-type: none"> Protect head Keep airway open/watch breathing Turn child on side 	
Emergency Response		A seizure is generally considered an emergency when:	
A "seizure emergency" for this student is defined as:		<ul style="list-style-type: none"> Convulsive (tonic-clonic) seizure lasts longer than 5 minutes Student has repeated seizures without 	
Seizure Emergency Protocol (Check all that apply unless clearly below):			
<input type="checkbox"/> Contact school nurse at _____			
<input type="checkbox"/> Call 911 for transport to _____			

What information should be in my seizure action plan?

The seizure action plan will be completed together with your child's medical team. It will include information such as:

- What your child's seizures look like
- How often the seizures occur
- What to do in the case of a seizure
- What hospital you want to go to, if it becomes necessary for hospital care
- Basic seizure first aid



You can see sample seizure action plans on the Epilepsy Foundation website. It is also helpful to provide your child's caregivers with a copy of the Seizure First Aid guidelines.

Is there anywhere I can go for more information?

Your child's medical team is the best resource for answering questions that are specific to your child. For generalized seizure information, the Epilepsy Foundation website can be a great resource. Attached are several of their handouts that many families find useful.

There are also many apps now available to help monitor and track seizures. Here are a few to consider:

- ✓ [Seizure First Aid: Epilepsy Foundation](#)
- ✓ [My Seizure Diary: Epilepsy Foundation](#)
- ✓ [Seizure Tracker](#)
- ✓ [Epilepsy Journal](#)





Family Companion: Cognition

What is cognition?

Cognition is your brain's ability to think and learn new skills.

Cognition includes the brain's ability to:

- Process things that you see, feel, or hear
- Communicate what you want or need
- Understand words
- Create opinions
- Form new memories & remember old memories
- Problem-solve



Will leukodystrophy impact my child's cognition?

Because each child is different, there is no single answer to this question. Children who are in the early stages of leukodystrophy may never have cognitive changes, or the changes may be mild. Children in the advanced stage of leukodystrophy can lose many parts of their cognition. Other children may have developmental delays.

What should I watch for?

Cognitive changes are common in children with most types of leukodystrophy. Let your child's medical care team know if you see changes in how your child is learning or if they are losing skills. The neurologist should check your child's development at every visit. They may also recommend formal testing, such as neuropsychological evaluation. Your child's school is an important partner in watching for cognitive changes.



My child is asymptomatic. Do they still need testing?

Your child's medical team may recommend baseline testing to find out how your child is thinking and learning right now. There are many ways by which people learn. Everyone has things that come easily for them, and things that are harder. Cognitive testing helps to identify an individual's strengths and weaknesses. As testing is repeated over time, it can find and track changes in cognition.

Neuropsychological testing, evaluation by a Speech Language Pathologist, and many other methods can be used. Talk to your child's medical team to find out whether cognitive testing is recommended for your child.



Quick tip: Always tell the medical team right away if you see your child losing a skill. Is there something your child used to do easily but is now getting harder? Perhaps they are starting to forget words or having trouble talking. Maybe they used to run but are falling more now.

How often should my child's language be tested?

The American Academy of Pediatrics recommends regular language screening for all children age 9 months to 30 months. Children with a leukodystrophy should have this same screening. Your pediatrician can do this screening. There are several tests your pediatrician or neurologist can use to do this screening. Testing may be performed when your child is:

- 9 months old
- 18 months old
- 24 months old
- 30 months old



Who can help in managing my child's cognition?



- Pediatrician
- Neurologist
- Early Intervention (for children under age 3)
- School (for children ages 3 and up)
- Special Education services (e.g. Speech Therapist, Occupational Therapist, educational aide)

These are all specialists who can help you understand how your child is developing. They can also help you to put services in place if needed.

How do I get my child's needs are being at school?

It can be easy to forget that your child's school is a big part of their care team. In the United States, there are two programs that can help make sure your child has the support they need at school: a 504 plan and an Individualized Educational Plan (IEP). A 504 plan provides services so that a child can access education resources regardless of any physical disability. An IEP provides services for any child with a developmental delay or learning disability.



Family Companion: Cognition Beyond the Basics

What Is Cognition?

Cognition is your brain's ability to think and learn new skills. Cognition includes the brain's ability to:

- Process things that you see, feel, or hear
- Communicate what you want or need
- Understand words
- Create opinions
- Form new memories & remember old memories
- Problem-solve



What are signs that my child is having trouble with cognition?

Every child is different. Your child's symptoms will depend greatly on the disease stage. During early onset, the child may become more irritable. S/he may have difficulty with attention and concentration. Some children with leukodystrophy have developmental delay. Other children have developmental regression.



What is developmental delay?

The pediatrician often reviews developmental milestones with you. These milestones include sitting independently, walking, talking etc. Children with developmental delay take longer than usual to master an expected skill. They are not losing skills, but rather learning at their own pace.

What is developmental regression?

Developmental regression is the loss of a skill(s) that the child already had mastered. Watching your child losing abilities can be scary and upsetting. While developmental regression can be a sign of disease progression, it can also signal other conditions that are treatable.

Always talk to your provider about other causes of developmental regression. These can include:



- ✓ Infection
- ✓ Difficulty sleeping
- ✓ Change in medications
- ✓ Increase in seizures
- ✓ Pain

How will we monitor my child's cognition?

The neurologist will check your child's cognition during every office visit, as part of their regular physical exam. The medical team will also ask about the skills your child currently has. Neuropsychological or another type of formal testing may also be recommended.



“We were fortunate enough to have access to music therapy. Jackson responded to the music by slowing his breathing, almost in a way to quiet himself to be able to better hear the music. In that moment, we realized how powerful not only the music was but also his sense of hearing.”

Is Cognitive Decline Treatable?

There are many services available for your child if s/he is experiencing cognitive difficulties. Services can be provided through your medical insurance, as part of the child's school IEP, by a palliative care organization, privately paid by families, or any combination . It can be helpful to have your child seen by a type of doctor called a physiatrist. The physiatrist specializes in rehabilitation and can help you understand what services will help your child the most.

Therapies to Help Children:

Physical Therapy	Sign Language
Aquatic Therapy	Communication Devices
Occupational Therapy	Vision Specialist
Speech Language Therapy	Hippotherapy (horse therapy)
Music Therapy	Social Work

“Hunter loved his Water Therapy/Jacuzzi - above all other therapy. There was nothing that could relax him more and give his body freedom to move, like the water. It calmed him early on when nothing else could. He spent at least an hour, sometimes two in the water every day. And, in the summer, the pool was a wonder for him. We had so much fun in the pool!”



What about communication?

Children with leukodystrophy can lose their ability to speak. Many children who lose their words find new ways to communicate. Smiles and facial movements are ways that we all communicate. There are also more augmentative communication tools. In fact, there are so many new tools that we have dedicated an entire section of this guide to communication tools.



Family Companion: Pain-the Basics

Do children with leukodystrophy have pain?

All children can have pain from common illness or injury. Pain related to leukodystrophy is uncommon in **asymptomatic** children. Children in the early to advanced stages often can have pain related to the disease.

You are your child's best advocate, and it can be helpful to keep track of pain you feel your child is experiencing as well as anything that makes your child more or less comfortable. Using a pain scale to record what your child is experiencing can help your child's medical team better understand what is happening. The medical team will help you develop a plan for pain management.



What is the Best Pain Scale for my Child?

There are 4 pain scales that are often used in children. When you pick which scale to use with your child, think about the child's age and how s/he communicates with you. Does your child use words to communicate? Does the child point to what they want? Do they use facial expressions to communicate?



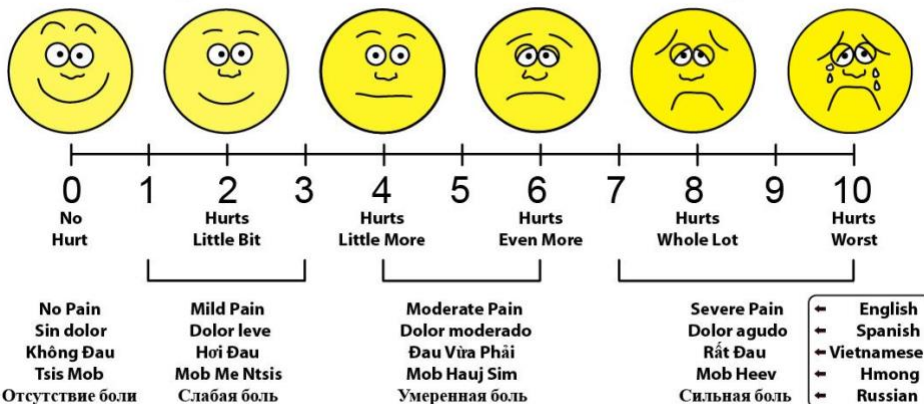
Infants and children under age 3 cannot describe their pain with words. But even very young infants can tell us about pain with their body movements and behaviors. The FLACC pain scale is best to quantify pain in these children. For children with complex medical needs, we recommend the revised FLACC (rFLACC). The faces scale is better used with older children. It pictures 5 different faces. Your child chooses the picture that looks like how they are feeling.

Finally, there is the 0-10 number scale that is used by adults. This scale goes from 0 to 10, with 0 being no pain and 10 being the worst pain possible.

Here is a chart to help you think about what pain scale would be best to use with your child:

Pain Scale	Best if your child is...
FLACC	<ul style="list-style-type: none"> • Child is under 3 years old • Child is non-verbal at any age • Child has normal “tone” in arms and legs
Revised FLACC	<ul style="list-style-type: none"> • Child is non-verbal at any age • Child has “increased tone” • Child has special needs
Faces Scale	<ul style="list-style-type: none"> • Child is around 3 years old • Child can point to what they want • Child has good vision • Child understands the faces
Number Scale	<ul style="list-style-type: none"> • School age children • Created for children 9 years and older • Child has to be verbal • Child has to understand numbers

Wong-Baker FACES Pain Rating Scale



From Hockenberry MJ, Wilson D: *Wong's Essentials of Pediatric Nursing*, ed. 8, St. Louis, 2009, Mosby. Used with permission. Copyright Mosby.

FLACC Scale:

	DATE/TIME					
Face 0 - No particular expression or smile 1 - Occasional grimace or frown, withdrawn, disinterested 2 - Frequent to constant quivering chin, clenched jaw						
Legs 0 - Normal position or relaxed 1 - Uneasy, restless, tense 2 - Kicking, or legs drawn up						
Activity 0 - Lying quietly, normal position, moves easily 1 - Squirming, shifting back and forth, tense 2 - Arched, rigid or jerking						
Cry 0 - No cry (awake or asleep) 1 - Moans or whimpers; occasional complaint 2 - Crying steadily, screams or sobs, frequent complaints						
Consolability 0 - Content, relaxed 1 - Reassured by occasional touching, hugging or being talked to, distractible 2 - Difficult to console or comfort						
	TOTAL SCORE					

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What can I do to help my child?

Figuring out the cause of the pain is **always** the first step to treatment. It is helpful to track your child's pain with a pain scale and/or pain journal in order to share with their medical team.

Common causes of pain in all children:

- Injury (from a fall, for example)
- Headache or migraine
- Teething/dental pain
- GI reflux
- Stomach pain (constipation is very common)
- Illness (such as the flu)

What are some sources of pain specific to leukodystrophy?

Pain can be a symptom of leukodystrophy even in children with early symptomatic disease. Pain and irritability are the most common first symptoms in infantile Krabbe.

Pain can also occur early in leukodystrophies that are known to cause neuropathy. **Neuropathy** is pain due to nerve damage. It most often starts in the feet and legs. Neuropathy is most common in Krabbe, MLD and adult AMN. Neuropathy pain is treatable! Over-the-counter medications such as acetaminophen and ibuprofen do not usually help neuropathy. However, there are prescription medications that your child's team can prescribe.

Note: For children with MLD, the doctor should also check for gallbladder disease. It is common for children with MLD to have sludge in the gallbladder that can cause pain.

What are some ways to help pain, other than medication?

Light massage on the stomach can help pain related to constipation. Increasing fiber & water intake, as well as daily activity & movement, are more good ways to treat constipation. If your child's pain is caused by an injury or illness, talk to your child's pediatrician about how to manage the pain.

Distraction with games, screen time, friends or other enjoyable activities can be a good treatment for mild pain. Therapies can help manage chronic pain. Some therapies include:

- Sensory therapy: rocking, music, lighting, fans, warm water, vibration
- Heat and cold application
- Aquatic therapy
- Music therapy
- Massage
- Reiki
- Physical and occupational therapies



What medications can reduce my child's pain?

Your child's medical team will decide what medications are best for your individual child. These may include over-the-counter medications (such as ibuprofen or acetaminophen) for mild pain. Prescription medications may be used for moderate to severe pain. Other medications may be used to target specific kinds of pain common in leukodystrophy, such as muscle spasm. Sometimes pain medications have side effects. Your child may be more sleepy or drowsy than usual. If you notice this, make sure to let your team know so they can adjust your child's medications to keep them comfortable without unwanted side effects.

How will I deal with seeing my child in pain?

It can be incredibly difficult to see your child in pain and not be able to immediately comfort them. You might feel frustrated, sad, or powerless. Remember that you are your child's best advocate, and that there are ways to manage their pain and keep them as comfortable as possible. Feel empowered to educate others, including your leukodystrophy care team, about how your child expresses their pain or discomfort.

Family Companion: Glossary

Anticonvulsant: A medication used to prevent and/or reduce seizures

Asymptomatic: Having no symptoms

Awake and Alert: Aware of things that are happening

Baseline: A base for measurement; A starting point used for comparison

CBD: Cannabinoid (CBD) is a chemical part of marijuana. CBD does not cause the “high” associated with marijuana. There has been interest in CBD being used as a seizure medication, but it has not been tested in children with a leukodystrophy.

Electroencephalogram (EEG): An EEG is a test that is used to find out what part of the brain is causing seizures. A special technician uses stickers and glue to attach small wires to the child’s head. The wires connect to a machine that can read the electric waves produced by the brain. The EEG reading allows the neurologist to see what is happening inside the brain. An EEG can show normal brain activity, seizures, or slowing.

Brainwaves seen on an EEG Child having an EEG



Epilepsy: A disorder relating to a person who has had multiple seizures. Also known as **seizure disorder**

Epileptiform Discharge: The type of brainwave seen on an EEG that indicates a seizure

Fatigue: Feeling tired during the day. Low energy, sleepy, or drowsy

GI/Gastrointestinal: Refers to the stomach and intestines

GI Dysmotility (Gastrointestinal Dysmotility): This is when the stomach and intestine have difficulty moving food through the system. Children often have pain with this. Children can have bloating (gas), constipation, and/or nausea.

GI Reflux (Gastrointestinal Reflux): A common problem in young children. The stomach uses special acid to break down food. When a person has GI reflux, this acid flows up the throat and causes a burning feeling. This is sometimes called “heartburn”.

IEP (Individualized Education Program): This is a legal document designed specifically for your child to help them reach their academic goals. An IEP documents your child’s current performance, annual goals, special education and related services, participation in testing, participation with nondisabled peers, transition services, and methods of measuring progress.

Ketogenic Diet: This is a special diet that may help control seizures in some children. This diet dramatically limits carbohydrates and is very high in fat. This type of diet should **only** be considered with the help of a doctor and dietician. It is not recommended for boys with ALD because these boys are not able to digest certain types of fats.

Muscle Spasms: A sudden, uncontrolled tightening of a muscle. Can be seen as quick jerking movement of a muscle, or a prolonged contraction. This tightening is often painful.

Processing Speed: Processing speed is how long the child takes to understand what s/he is seeing, hearing, or feeling. Delayed processing speed can cause a blank pause before the child responds. This is because it takes the brain longer to understand what was just said. A blank stare because of delayed processing speed is not the same as an absence seizure.

Reflexes: Reflexes are automatic responses that happen without thinking, such as kicking your leg when the doctor hits the knee with a reflex hammer. Infants have many more reflexes than adults. In normally developing children, many reflexes go away as they grow. Others should stay. Doctors use examination of these reflexes to help test how well the brain and nervous system are working.

Responsive: Awake and alert; able to respond to stimulation such as being touched, or things that are seen or heard

Seizure: A seizure is a sudden, uncontrolled surge in the brain's electrical activity

Seizure Disorder: A person who has had multiple seizures. Also known as **epilepsy**

Seizure Plan: A seizure plan is a formal document that allows all caregivers to know important facts about the child's seizures. A seizure plan should be completed together with your medical team. It will include information such as:

- What the child's seizures look like
- How often the child has seizures
- What should be done when the child has a seizure
- The hospital of choice, if it becomes necessary for hospital care
- Basic seizure first aid

Seizure Action Plan		Effective Date	
This student is being treated for a seizure disorder. The information below should assist you if a seizure occurs during school hours.			
Student's Name	Date of Birth		
Parent/Guardian	Phone	Cell	
Other Emergency Contact	Phone	Cell	
Treating Physician	Phone		
Significant Medical History			
Seizure Information			
Seizure Type	Length	Frequency	Description
Seizure triggers or warning signs:			Student's response after a seizure:
Basic First Aid: Care & Comfort			Basic Seizure First Aid
Please describe basic first aid procedures:			• Stay calm & track time
Does student need to leave the classroom after a seizure? <input type="checkbox"/> Yes <input type="checkbox"/> No			• Do not restrain
If YES, describe process for returning student to classroom:			• Do not put anything in mouth
Emergency Response			• Stay with child until fully conscious
A seizure emergency for this student is defined as:			• Record seizure in log
Seizure Emergency Protocol (Check all that apply, preferably below):			For individuals wearing:
<input type="checkbox"/> Contact school nurse or _____			• Present head
<input type="checkbox"/> Call 911 for transport to _____			• Keep airway open/clear
<input type="checkbox"/> Notify parent or emergency contact			• Keep child on side
<input type="checkbox"/> Administer emergency medications as indicated below			A seizure is generally considered an emergency when:
<input type="checkbox"/> Notify doctor			• Convulsion (stereotyped) seizure lasts longer than 5 minutes
<input type="checkbox"/> Other _____			• Student has repeated seizures without regaining consciousness
			• Student is injured or has injuries
			• Student has a broken bone
			• Student has breathing difficulties
			• Student has a seizure in water

Seizure plan forms can be found at the Epilepsy Foundation website. Your child's medical care team may have their own version.

Sleep Hygiene: Sleep hygiene is a set of behaviors you can do at night to help you fall asleep. This often includes things like going to bed at the same time every night and turning off electronics one hour before bed

Startle Reflex: The startle reflex is a normal reflex in an infant for the first months of life. When a baby is frightened, feels they are falling, or hears a sudden sound, the arms and legs kick out suddenly. This reflex is normal in infancy but should leave during development. A child with impaired brain function may keep reflexes even as s/he grows. A sudden jerk due to the startle reflex should not be confused with a seizure.



White matter: The brain is made up of **grey matter** and **white matter**. People often describe the white matter as insulation around an electrical wire. When the white matter is healthy, messages move very fast from the brain to the rest of the body. When it is damaged, messages are slow.

