

Images depicted are not actual patients.

Difficulty walking or talking, frequent vomiting, or problems with memory, thinking, or learning could be signs of a neurological condition.

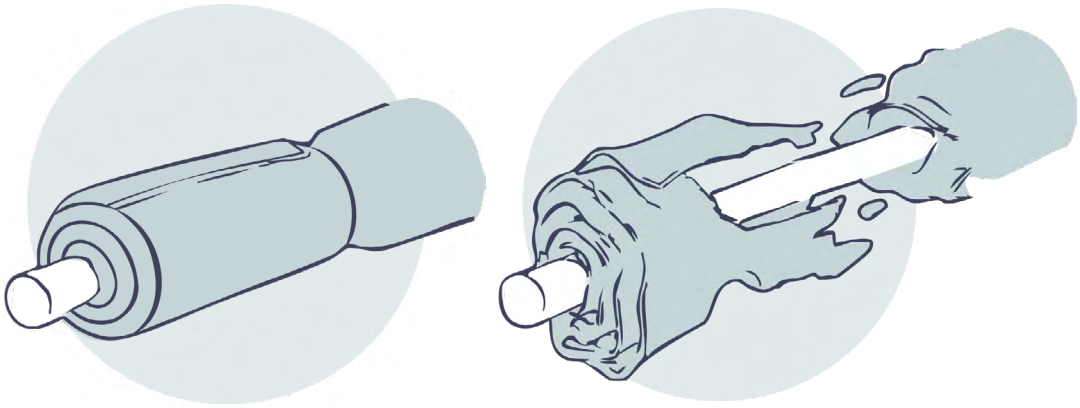
Don't wait—when something doesn't feel right, trust your instincts. Ask your doctor about seeing a neurologist who can run the right tests to help find answers.

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What is Alexander disease

Alexander disease is a **rare genetic condition that affects the brain and spinal cord**. It belongs to a group of neurological conditions called leukodystrophies that damage the **brain's white matter (myelin)**. Myelin acts like insulation around nerve fibers, helping messages travel quickly and smoothly in the brain and body.



HEALTHY MYELIN

DAMAGED MYELIN

Alexander disease is caused by changes (mutations) in a gene called ***GFAP*** (glial fibrillary acidic protein). This leads to the buildup of protein clumps—called **Rosenthal fibers**—in the brain's support cells. Over time, this buildup **damages myelin**—the protective coating around nerve cells—which can affect how the brain and nervous system work.

The abnormal *GFAP* gene can sometimes be inherited from a parent, but in most cases of Alexander disease, both parents have normal *GFAP* genes and the mutation occurs randomly.

Alexander disease:



- Can affect people of all ages, including **infants**, **children**, **and adults**
- May cause symptoms that **affect movement**, **speech**, **memory**, **thinking**, and other functions of the nervous system
- **Progresses differently** depending on the age when symptoms first appear

What are the common symptoms of Alexander disease?

Alexander disease can vary from person to person, and symptoms often depend on when they first appear. If **symptoms start in babies or young children**, they are usually **more severe and progress more rapidly**. If **symptoms begin later in childhood or adulthood**, they **may develop more slowly and be less severe**.

Infants and children up to 4 years old

- **Loss of or delays** in **reaching milestones** (such as sitting, walking, or talking)
- **Seizures** (sudden episodes of shaking or staring)
- **Trouble eating** or **swallowing** foods and liquids
- **Frequent vomiting**
- **Larger-than-usual head size**
- **Muscle stiffness, spasms, and involuntary contractions**
- **Difficulty with balance, coordination, or movement**



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Children older than 4 years of age and adults

- **Problems with motor skills** (such as balance and walking)
- Difficulty **speaking and swallowing**
- **Frequent vomiting**
- **Breathing problems**
- **Heart rate, blood pressure, or digestion problems**
- **Muscle stiffness, spasms, and involuntary contractions**
- Changes in **memory or thinking**

Remember, these are the most common symptoms by age, but they are not a complete list. Symptoms can vary from person to person and not everyone with Alexander disease will experience all of them.

Alexander disease is a progressive condition

Symptoms of Alexander disease **usually worsen over time**, though the rate of progression can vary depending on the age when symptoms first appeared. In infants, the disease can worsen quickly. When it begins later in childhood or adulthood, it often progresses more slowly—sometimes over many years or even decades.



Trust your instincts and take action. If you or a family member is experiencing any of these symptoms—or other unexplained neurological problems—it's important to see a doctor. Getting the right diagnosis can help guide care and support.

How is Alexander disease diagnosed?

Because Alexander disease can look like other neurological disorders, it is sometimes mistaken for conditions such as a brain tumor in children or multiple sclerosis in adults. This means that some people, especially **older children and adults**, **may go years without the right diagnosis**. **In infants and young children**, **the disease is usually diagnosed sooner** because their symptoms tend to be more severe and recognizable by others.

On average, it can take
6 YEARS and **12 SPECIALISTS**
for people with a rare disease **to receive an accurate diagnosis**.

Steps to diagnose Alexander disease may include



Referral to a Neurologist

Your doctor may refer you to a neurologist (a specialist in disorders of the nervous system)



Medical History and Exam

The neurologist may ask about symptoms, development, and family history and do a physical and neurological exam



Brain MRI (magnetic resonance imaging)

An MRI scan looks at the brain for changes often seen in Alexander disease



Genetic Testing

A DNA test checks for changes in the *GFAP* gene, which can confirm the diagnosis

Speak up for answers

Your observations and questions are valuable to your doctor.
BE YOUR OWN ADVOCATE!

If you or a loved one is experiencing unexplained neurological symptoms, ask for a referral to a neurologist and request diagnostic testing, including an MRI.

Here are some questions you might ask your doctor:

- **Could my/my loved one's symptoms be related to a neurological disorder?**
- **Do you think it would be helpful to see a neurologist for a more specialized evaluation?**
- **Are there specific tests, like an MRI of the brain or genetic testing, that could help us understand what's happening?**
- **How can I best track symptoms between visits?**

Confirming a diagnosis of Alexander disease as early as possible allows your healthcare team to develop a personalized care plan to manage symptoms.

Living with Alexander disease

Now, there is a treatment option that is an important step forward for Alexander disease. At the same time, managing symptoms and enhancing quality of life remain essential to provide comfort.

Care typically involves a **team of specially trained professionals** who support patients and their families. This team may include **neurologists**, **physical therapists**, **speech therapists**, **nutritionists**, and others who help manage the different symptoms of Alexander disease.

Examples of symptomatic and supportive care

Symptom management may include therapies and devices that help with physical, speech, thinking, or learning difficulties that some people with Alexander disease may experience.

Seizure management	• **Medicines** to help control seizures
Nutrition and feeding	• **Nutrition counseling** to maintain healthy weight and prevent dehydration • **Feeding tube** (gastrostomy tube) if swallowing becomes too difficult
Movement and muscle support	• **Physical therapy** to keep muscles as flexible and strong as possible • **Occupational therapy** to learn ways to make daily tasks easier • **Braces**, **walkers**, **or wheelchairs** to improve mobility, safety, and independence • **Medicines** to reduce muscle stiffness or tightness
Breathing support	• **Interventions to support respiratory function**, including preventing lung infections and keeping airways clear • **Breathing machines**, such as CPAP or BiPAP, to make breathing easier at night or when needed
Speech, swallowing, and communication	• **Speech therapy** to improve speaking, understanding, or using alternative communication devices if talking is difficult • **Swallowing therapy** to make eating and drinking safer and reduce choking risk
Cognitive, emotional, and family support	• **Evaluations** to check for changes in memory, thinking, or learning • **Counseling** or therapy for emotional well-being of patients and caregivers • **Support groups** to connect with other families living with rare diseases

BiPAP, bilevel positive airway pressure; CPAP, continuous positive airway pressure.

An earlier diagnosis can **HELP PATIENTS ACCESS** the care they need.

An earlier diagnosis can help patients by allowing families to access specialized care, manage symptoms sooner, and plan for needed support and resources. For those with advanced disease, palliative care to focus on comfort may be an option.

Impact on caregivers

Being a caregiver for someone with Alexander disease can be **emotionally and physically demanding**. Because the disease is rare and gets worse over time, families often face uncertainty and may find it hard to locate healthcare providers who have experience with the condition. **Managing daily care, coordinating with specialists, and advocating for your loved one can feel overwhelming.**

Caregiver coping strategies

ASK FOR HELP

Reach out to family, friends, or community services when you need support

CONNECT WITH OTHERS

Join support groups to share experiences and advice

STAY INFORMED

Learn about Alexander disease and available resources

TAKE BREAKS

Short rest periods can help you recharge

LOOK AFTER YOURSELF

Eat well, stay active, and care for your own health

SEEK SUPPORT

Counseling or respite care can help reduce stress



Connect with advocacy organizations

Patient advocacy organizations offer **education**, **resources**, **and support** for people with Alexander disease and their families:

ALEX, THE LEUKODYSTROPHY CHARITY

Provides access to vital support and information for all those affected by a genetic leukodystrophy and helps advance medical research.

LEARN MORE

ELISE'S CORNER

A community group focused on spreading awareness of Alexander disease and aiding research efforts to find treatments and a cure for this rare genetic disorder.

LEARN MORE

END AXD

Focused on driving research and development toward a treatment—and eventual cure—of Alexander disease, while helping patients get the care they need.

LEARN MORE

HUNTER'S HOPE

Provides critical information and research on leukodystrophies while supporting and encouraging patients and their families through these life-limiting illnesses.

LEARN MORE

NATIONAL ORGANIZATION FOR RARE DISORDERS

Dedicated to improving the lives of Americans living with rare diseases, NORD drives progress in research, care, and policy in partnership with more than 350 disease-specific member organizations.

LEARN MORE

UNITED LEUKODYSTROPHY FOUNDATION

Dedicated to funding research and providing disease information and medical referrals to patients and families living with a leukodystrophy.

LEARN MORE

Speak up and get support—your voice makes a difference

To learn more about recognizing the symptoms and confirming a diagnosis of Alexander disease, visit **AlexanderDisease.com/patient**.

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