National Tay-Sachs & Allied Diseases Association – Home

Diseases Support for Families Donate to NTSAD

Events NTSAD Research About NTSAD

Contact Us

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Diseases

Support for Families

Donate to NTSAD

Events

NTSAD Research

About NTSAD

Contact Us

Skip to any section using the links below:

What is Canavan Disease?

Canavan disease is a rare **leukodystrophy** where the reduced or absent activity of a vital **enzyme** called aspartoacylase (ASPA) means the body cannot produce **myelin**, a fatty membrane that protects nerves in the brain. The nerve cells are vulnerable and unable to work properly, which damages the brain and spinal cord.

There is currently no cure for Canavan disease, but there are treatments and therapies to help manage symptoms and maintain a **baseline** for as long as possible.

Cause

Canavan disease is a rare genetic condition that's passed from parents to children. It is a recessive disorder, which means that both parents must carry a change in the **gene** that causes Canavan for it to be passed on to a child.

Symptoms

Canavan disease exists on a spectrum, and every individual experiences it differently. Sometimes symptoms are noticeable at birth, and the condition usually becomes apparent between 3–6 months of age.

Between 3–6 months of age, you may notice your child:

- Has a large head (macrocephaly) and difficulty controlling their head
- Has decreased muscle tone (**hypotonia**)
- Experiences a slowdown in development
- Experiences reduced vision

Between 6–23 months, you may notice your child:

- Begins to regress, losing the ability to crawl, turn over, sit, and reach out
- Begins to lose coordination
- Progressively loses the ability to swallow
- Has trouble breathing

At 36 months and onward, you may notice your child:

- May experience recurrent **seizures**
- Progressively loses muscle tone and function

The Canavan Disease FDA Patient Listening Session took place in March 2023. Read the summary.

Newly Diagnosed

To confirm a diagnosis, biochemical and genetic testing is conducted. Children with Canavan disease have higher levels of N-acetylaspartic acid (NAA) in the urine and a deficiency of ASPA in cultured skin fibroblasts. **DNA** testing will uncover any mutations that cause Canavan disease.

First Steps

When your child gets a Canavan disease diagnosis, it's normal to feel overwhelmed. Take the time you need to absorb the news and process your emotions. Consider reaching out to family, friends, and neighbors to build a support network to help you today and moving forward.

You're always welcome to reach out to our Family Services Team for information, advice, and support at any point in your experience.

When you're ready, the following steps will help you get organized and move forward.

- **Gather Key Information:** Use a notebook, folder, or binder to collect information, adding to it as you go. Important information to gather includes:
  - Where your child's evaluation/assessment was done
  - Where your child's diagnosis was made
  - Healthcare provider name(s) and contact information
  - Handouts and resources
  - Important telephone numbers and addresses
  - Copies of assessment reports, diagnostic and imaging tests, lab reports, and medications

  Please note: There are several health management apps available to help track this information. If you decide to use one, be sure to review its data privacy policies.

- **Apply for a Medicaid Waiver:** Typically, there is a waiting period for Medicaid – the length of time depends on the state.

  To apply for a Medicaid waiver in your state, call toll-free (877) 267-2323 or visit the Medicaid website (Medicaid.gov) for more information.

  Please note: The Kids' Waivers website is an excellent resource on Medicaid waivers and programs for children.

- **Apply for Social Security Disability Insurance:** Please note: Canavan disease is on

the Compassionate Allowance list to expedite your application on behalf of your child.

Visit the Social Security website to apply for Social Security Disability Insurance for Children.

- **Start Building a Healthcare Team:** Your child will be supported by a healthcare team that includes clinicians, pediatricians, neurologists, gastroenterologists, and pulmonologists. As well as providing physical care to your child, they will also help you advocate with insurance companies and school systems, as needed.

  Because Canavan is a rare disease, it's possible that healthcare providers in your region haven't treated a child with the disease. In this case, try to find a pediatrician or specialist who makes you feel comfortable, answers your questions, listens to your concerns, and supports your family's Philosophy of Care.

- **Get to Know Your Insurance Policy:** Review the coverage offered by your insurance policy to better understand eligibility requirements, benefits, regulatory information, and grievance procedures.

- **Ask for an Insurance Case Manager:** Contact your insurance company and ask for a dedicated case manager, who will get to know your loved one's specific medical needs so you won't have to explain the diagnosis each time.

A dedicated case manager will help you get the best insurance coverage possible.

Philosophy of Care

Your Philosophy of Care is a clear plan that outlines your goals for your child's care and health management. It includes the interventions you would like to use (and avoid) and reflects what you think will work best for you and your family.

Benefits of Having a Philosophy of Care

Overall, a Philosophy of Care lets you think through how you'd like to care for your child and communicate that information with your healthcare team. Specifically, your Philosophy of Care is vital during an emergency when it's difficult to think clearly and make decisions quickly.

Every family's Philosophy of Care is different—ranging from few interventions, moderate interventions, and more interventions—and we support all families in their decisions.

Make Your Own Philosophy of Care

To build a Philosophy of Care begin by jotting down thoughts and ideas as you research the disease, consult with healthcare providers, and discuss with your partner, family, close friends, and others. You may find you have a clear plan within days, or it might take weeks or months. Keep in mind that the Philosophy of Care isn't written in stone. If a goal doesn't serve your family anymore, it's okay to change it or remove it altogether.

You can make your own Philosophy of Care or download our template here.

Symptom Management

While there is currently no cure for Canavan disease, it is possible to manage symptoms like seizures and

trouble swallowing as guided by your Philosophy of Care.

Respiratory Health

We recommend that you develop a respiratory health management plan with your pediatrician and consult with a pulmonologist for advanced respiratory health needs and management.

Children with Canavan disease are prone to lung infections because of increased saliva and mucus and reduced swallowing. There are many options available to promote respiratory health, including:

- Limiting exposure to people who may be sick
- Equipment (e.g., a respiratory therapy vest or positioning equipment)
- Taking extra precaution to make sure you child does not aspirate during feeding if they are eating or drinking orally. If you notice coughing episodes after eating orally, you may want to discuss a swallow study with your child's care team.
- Speaking with your care team about how to manage secretions (e.g., suction machine, Botox treatments for the salivary glands, positioning)

Seizures

Feeding & Nutrition

Sensory Stimulation
Complementary Therapies

How to Help

If your family member or close friend has a child diagnosed with Canavan disease, you may not know how to help. The best thing you can do is be there for them. Caregiving is incredibly time consuming and is emotionally and physically draining. It can also feel very lonely, and parents may struggle to ask for help.

There are many ways to help children, families, and siblings. Here are seven ways to get you started:

1. **Offer** concrete help like picking up groceries, caring for siblings, cooking meals, doing the laundry, housecleaning, or offering to babysit.
2. **Learn** about Canavan disease to get to know what they may be experiencing.
3. **Provide** companionship by dropping in with coffee and a treat or inviting them out for a walk. If you're out of town, try to visit in a way that won't disrupt their daily routine.
4. **Listen** with empathy and understanding, knowing they will experience a wide range of emotions.
5. **Be** a resource, but don't give advice.
6. **Get** to know their special child by asking parents what they would like. You might offer to read a story, sing a song, or bring a soft stuffed toy to snuggle.
7. **Engage** with siblings and invite them on special outings or your own family gatherings.

We're Always Here to Help

Getting a Canavan diagnosis can be overwhelming. It's hard to know what to do first. Our Family Services Team is here to help. They'll answer your questions,

share information, and invite you to connect with our caring and helpful Community.

We Care for Rare

Facu y Giuli

Juvenile Tay-Sachs Disease

> “Facu (12) y Giuli (16) ellos aman viajar y divertirse en familia”

Alejandra Saipert, Madre

View All Impact Stories

National Tay-Sachs & Allied Diseases Association (NTSAD) leads the worldwide fight to treat and cure Tay-Sachs, Canavan, GM1, and Sandhoff diseases by driving research, forging collaboration, and fostering community. Supporting families is the center of everything we do.

The content of this website is intended for informational purposes only and not intended to be a substitute for professional medical advice, diagnosis, or treatment.

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Community Connections

Get the latest news from NTSAD about our Community, the research that provides hope, and what is happening in the world of rare disease.

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