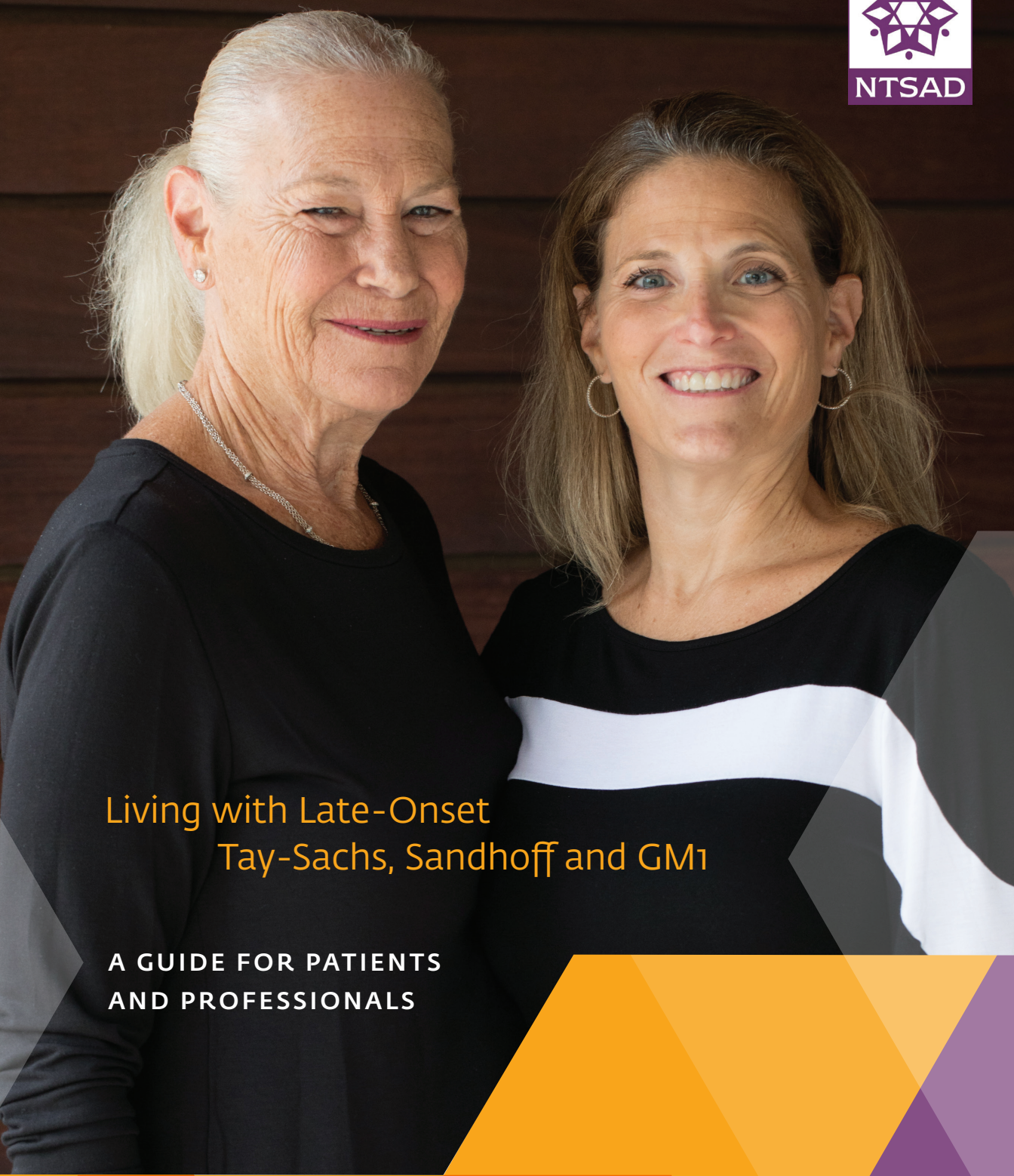
NTSAD



Living with Late-Onset Tay-Sachs, Sandhoff and GM1

A GUIDE FOR PATIENTS AND PROFESSIONALS

Understanding Late-Onset Diseases

Living with late-onset Tay-Sachs, Sandhoff or GM1 disease can be challenging. Because the diseases are rare, friends, family and co-workers may not have heard of them, and may not understand how they affect you. Even healthcare providers may be unfamiliar with these diseases. Diagnosis is often delayed because of this. Some people live with unexplained and troubling symptoms for five years or more before receiving an accurate diagnosis.

This booklet explains the signs and symptoms people may experience, as well as ideas for managing them. It provides information for patients to share with family and caregivers.

Although these diseases are rare, it is important to remember that you are not alone, and that others are living with similar experiences. National Tay-Sachs & Allied Diseases Association (NTSAD) can be an important resource for you. Learn more at the end of this booklet or by visiting NTSAD.org.

About Late-Onset Tay-Sachs, Sandhoff and GM1

Tay-Sachs, Sandhoff and GM1 are rare genetic disorders that destroy nerve cells in the brain, leading to a progressive loss of muscle control.

People who have these diseases are missing specific and important enzymes (Hex A in Tay-Sachs, Hex B in Sandhoff and GLB1 in GM1). These missing enzymes break down certain substances in the cells. When the enzymes are missing, the substances build up, damaging the cells and resulting in progressive symptoms.

All three diseases occur in three different forms, depending on when symptoms first appear: infantile, juvenile and late-onset. While children with infantile forms typically have no working enzyme, those with late-onset forms have a small amount of enzyme. As a result, the onset and progression of symptoms occurs slowly. This booklet focuses on the late-onset forms of the diseases.

These diseases are autosomal recessive genetic disorders. Autosomal means they affects males and females equally, and recessive means that children are only at risk if both parents carry the gene for the disease.

If both parents are carriers, there is a 25% chance with each pregnancy that the child will be affected. Carriers of most recessive diseases do not experience any adverse health effects.

Late-Onset Signs and Symptoms

Signs and symptoms often begin in the teens or early adulthood, but may occur much later. Even within a family, one affected sibling may start to experience symptoms decades later than another. The disease is variable, and not everyone experiences all the symptoms described here.

Muscle weakness

Clumsiness and muscle weakness in the legs is often one of the earliest signs of late-onset Tay-Sachs and Sandhoff disease. Sometimes, if the muscles are unable to support the joints, joint pain can result. Once diagnosed, adults often reflect back to their childhood and realize they experienced earlier symptoms, such as not being athletic.

Tremors

Over time, people may begin to experience tremors, muscle twitches or seizures.

Incoordination/balance/falls

Some people may have trouble coordinating voluntary movements. This can lead to falls and other problems with balance.

Fatigue

Muscle weakness and problems with coordination can result in fatigue for some people.

Speech and swallowing difficulties

Over time, people may experience slurred speech and may develop swallowing issues.

Mental health symptoms

About 40% of affected adults experience mental health symptoms such as bi-polar or psychotic episodes.

Managing Symptoms

There is no treatment or cure for these diseases, but there are ways to manage symptoms. A team of healthcare professionals can help manage the wide range of symptoms that occur.

Your Treating Physician

It is more important to find a treating physician or neurologist who you are comfortable with than one who has experience with your disease. Because of the rarity of the diseases, finding a local doctor with experience is often difficult. Instead, try to find a doctor who will work with you to manage your disease and consult with expert clinicians on the NTSAD Scientific Advisory Committee (SAC) as needed. Visit NTSAD.org for more information.

The relationship with the doctor and healthcare team is a critical component of successful disease management, particularly because there are no sure-fire answers and a wide range of severity and symptoms.

Your treating physician can usually provide referrals to the other specialists listed here.

Speech-Language Pathologist

A speech-language pathologist (SLP) can be helpful at all stages of the disease. In the early stages, a SLP can assist with problem solving and developing strategies to overcome communication challenges and help you continue to live a more full and empowered life. A SLP may recommend use of augmentative or alternative communication devices and techniques, which can be as simple as a word/picture board, or more complex, such as an electronic device that speaks for you.

For example, the SLP might work with you and your family to create a word/picture board tailored to your environment (whether it be a private residence or a long-term care facility) or flexible enough to be carried around. If an electronic device might be beneficial, the SLP will evaluate your interest and motivation to use it, and will help make it easily accessible.

Physical Therapist

A physical therapist can help you manage the physical symptoms. They can work with you to keep your joints flexible and maintain your range of motion. In many cases a physical therapist will develop an exercise program to address your particular mobility challenges in order to promote independence and function in everyday life. The physical therapist also orders mobility aids and teaches you their proper use.

Occupational Therapist

An occupational therapist helps you enhance your function and independence in everyday life by exploring new ways to engage in activities or hobbies you enjoy. They can show you adaptive techniques that can help you overcome hand tremors and improve dexterity. Depending on your specific symptoms, they may be able to suggest ideas for modifying your home and work space, provide tools and strategies to deal with symptoms that interfere with driving, or help you with adaptive aids.

Mental Health Professional

Living with a late-onset disease may trigger feelings of isolation, depression and frustration, as you learn to adjust to new developments and restrictions. An active work, social and family life promotes emotional well-being. Many people also find it helpful to talk to a mental health professional about managing their mental health and enhancing their emotional well-being. Online therapy services can provide support via phone, text, or virtual visits.

Psychiatrist

About 30-40% of people living with late-onset Tay-Sachs or Sandhoff disease experience mental health symptoms such as hallucinations, paranoia, anxiety, manic-depression, severe depression, obsessive compulsive behavior, and disorientation. It is important to seek help early if you suspect psychiatric symptoms. Generally, your treating physician provides a psychiatric or neuro-psychiatric referral.

Living with a Late-Onset Disease

Often, the biggest concerns people face are maintaining independence and staying safe by preventing falls. Your healthcare team can help with both concerns.

Physiatrists, also known as physical medicine and rehabilitation physicians, have broad expertise that allows them to treat disabling conditions throughout a person's lifetime. They often lead a team of medical professionals that may include physical and occupational therapists.

A physical therapist can work with you to help you maintain your strength, range of motion and balance. These can help you with joint protection, energy conservation and endurance. Depending on your abilities, you may want to find a therapist who can help you with water-based exercises that reduce the load on your joints. Over time, you may be able to move on to land-based exercises.

The goal is often to help you find a level of intensity that lets you maintain or improve your ability to function without overstressing your muscles and causing damage. A physical therapist can help you track your progress with simple tests like a gait test or a 10-meter walk, to see how your abilities change over time.

A physical or occupational therapist can also help you with practical concerns, like standing up from a chair, getting out of bed, or getting into a car. They can also help you find and use devices ranging from trekking poles, to walkers, wheelchairs or scooters. Depending on your ability, they may be able to help you find devices that enable you to continue driving a car.

Community Resources

Most communities have resources to help people maintain their independence. Depending on where you live, these may include:

- Ride shares and van services to help you get around
- Housing advocates to help you find accessible and affordable housing
- Independent living facilities
- Vocational groups to help with job training if your abilities change
- Recreational opportunites and resources for people at all levels of ability
- Opportunities for social activities

A call to a local Senior Center or an independent social worker is often a good place to start asking about community services.

Other Resources

- Social Security Disability Insurance: www.ssa.gov/benefits/disability
- Americans with Disabilities Act: www.ada.gov
- Wheelchair Travel: wheelchairtravel.org
- Canine Companions for Independence: www.cci.org
- Clinical trials: Contact NTSAD for up-to-date information. info@ntsad.org

Although these diseases are progressive, many people find that they can work with their healthcare team and community to make the most of their abilities and find ways to maintain a level of independence.

A Family Gathering

The NTSAD Annual Family Conference provides over 300 parents, grandparents, affected children, healthy siblings, affected adults and their families the unique opportunity to gather with people that truly understand; learn about latest research and symptom management approaches; and discuss other important topics.

The primary goals of the conference are to empower, support and connect families coping with Tay-Sachs, Canavan, GM1 or Sandhoff. These diseases are always fatal in children and extremely debilitating in adults. The Annual Family Conference directly achieves our mission to support affected families and individuals in leading fuller lives.

Other examples of the powerful impact of the conference include: people donating equipment they no longer need, the strong bonds and friendships formed over just a few days, and incredible sense of hope and empowerment that comes from attending the conference.

About NTSAD

National Tay-Sachs & Allied Diseases Association (NTSAD) is one of the oldest patient advocacy groups in the country. We focus on funding research, supporting families and individuals worldwide, and raising awareness to prevent disease.

Today, NTSAD is recognized as a leading non-profit patient group with a demonstrated commitment to service, science and support. NTSAD gives help and hope to thousands of individuals and families from many backgrounds and ethnicities who have been or are affected by Canavan, GM1, Sandhoff, Tay-Sachs and related genetic diseases all over the world.

Learn more at NTSAD.org or contact us at info@ntsad.org

This brochure was made possible by a grant from Sanofi Genzyme.
Special thanks to Andrew Schaefer, Judy Kaplan, and Dr. Cynthia Tifft and her team for their input.