



Newly Diagnosed Guide: **Neuromyelitis Optica Spectrum Disorder (NMOSD)**



Adapted from Neuromyelitis Optica (NMO) What You Need to Know: A guide for patients, their families, and caregivers, 3rd ed. with permission from the Guthy-Jackson Charitable Foundation.

The Guthy-Jackson Charitable Foundation (GJCF) is an American non-profit organization dedicated to funding breakthrough research, increasing public health education, and bringing physicians and researchers together to develop safe and effective treatments and ultimately find a cure for NMO.

Further acknowledgments: Special thanks to Ann Yeh MA, MD, FRCPC, Dip ABPN, The Hospital for Sick Children, University of Toronto and Lelainia Lloyd, NMO patient advocate.

Canadian adaptation: Jennifer McDonnell and Natasha Vincent

Design and Publishing:

This project has been supported by funds received from Alexion and Hoffmann-La Roche Limited. The MS Society of Canada is solely accountable for the content associated with the project and certifies that all content has been developed without the influence of our funding partners.



Neuromyelitis Optica Spectrum Disorder (NMOSD)

A diagnosis of neuromyelitis optica spectrum disorder (NMOSD) can be confusing and frightening for individuals and loved ones. The information in this guide is intended help people living with NMOSD and their families understand NMOSD.

What is NMOSD?

Neuromyelitis optica spectrum disorder (NMOSD) is a rare autoimmune syndrome of the central nervous system (CNS) where antibodies can damage the spinal cord and/or optic nerves during attacks. It is a demyelinating condition, meaning, it damages the protective myelin sheath around the nerve fibres. The syndrome of NMOSD is characterized by:

- optic neuritis (ON) that affects eye function
- transverse myelitis (TM) that affects limb function
- area postrema syndrome (APS), which are episodes of otherwise unexplained hiccups or nausea and vomiting

Symptoms may include vision impairment or loss, imbalance, incontinence, weakness, numbness or paralysis of limbs or other body parts. There is currently no cure for NMOSD, however there are medications which slow down the activity of the immune system to prevent further attacks.

What causes NMOSD?

Two different antibodies have been associated with the clinical syndrome of NMOSD. The most common cause in the adult population is an attack on the aquaporin-4 (AQP4) water channel located in the optic nerves and spinal cord. Aquaporins (AQPs) are proteins that transport water across cell membranes. Approximately 70 per cent of adults living with NMOSD test positive for an antibody biomarker in the blood called the NMO-IgG (anti-AQP4 antibody).

Up to one third of people with NMOSD who test negative for anti-AQP4 antibodies may be positive for autoantibodies directed against a component of myelin called myelin oligodendrocyte glycoprotein (MOG), including more than half of all pediatric onset NMOSD.

Who gets NMOSD?

NMOSD is most often seen in women between the ages of 20 and 40. However, NMOSD has been diagnosed in children as young as two years old and adults in their 60s.

In Canada, there are approximately 1,000 to 3,000 people living with NMOSD. Research estimates that NMOSD affects up to 10 in 100,000 persons, which means hundreds of thousands of people live with NMOSD worldwide. The prevalence of NMOSD appears to vary in different regions and among distinct populations around the world. NMOSD also appears to be more common among individuals having genetic ancestry including African, Asian, Pacific Island, Polynesian or Caribbean descent. However, anyone can be affected by NMOSD.

How common is pediatric NMOSD?

Pediatric onset NMOSD, categorized as symptoms that appear before age 18 and represents about 3 to 5 per cent of all NMOSD cases. In more than half of pediatric onset NMOSD, the first symptom is optic neuritis or longitudinally extensive transverse myelitis (inflammation of the spinal cord). A distinguishing feature of pediatric onset NMOSD is a low frequency for AQP4 seropositivity compared with adult-onset NMOSD. However, children have a high prevalence of MOG-IgG antibodies. In more than half of all cases, children with MOG-IgG

antibodies have an acute disseminated encephalomyelitis (ADEM) presentation, which is characterized by a short-lived but widespread attack of inflammation in the central nervous system (brain, including optic nerve, and spinal cord).

What are the symptoms of NMOSD?

Symptoms of NMOSD can vary from person to person in duration and severity, including level of disability. Generally, NMOSD symptoms begin rapidly. After the initial attack, NMOSD follows an unpredictable course, and time to remission can vary. Recurring episodes of optic neuritis and/or transverse myelitis can be weeks to months in duration, and in some rare cases can last years. Usually, these symptoms are temporary and resolve fully or partially with treatment. While the syndrome of NMOSD is characterized by the following clinical features, a growing understanding of NMOSD suggests that the symptoms experienced depend on which antibody (AQP4-IgG or MOG-IgG) is found in the individual:

Symptoms and signs of optic neuritis (ON) may include:

- Rapid onset of eye pain or “eye headache” that is worsened by eye movement
- Impaired or complete loss of vision usually in one eye, but in some cases in both eyes
- Reduced light perception, color vision, visual clarity, and/or depth perception

Symptoms of transverse myelitis (TM) include:

- Pain in the neck or back
- Tightness or corset-like sensations in the abdomen, as well as arms or legs
- Sensitivity to touch, cold and heat

- Feeling of numbness, tingling, coldness, itching or burning, often spreading to large parts of the body over a period of minutes, hours or occasionally days
 - Weakness in arms or legs ranging from mild to complete paralysis in one or multiple limbs
 - Urgent need to urinate or difficulty urinating; urinary incontinence (unintentional passing of urine)
 - Constipation leading to vomiting, abdominal bloating, pain and inability to pass stool or gas; or bowel incontinence (unintentional passing of stools)
 - Muscle spasms that may last for several minutes accompanied by arm or leg pain
 - Fever in some cases
-

Symptoms of brainstem or brain involvement include:

- Area postrema syndrome (APS): episode of otherwise unexplained hiccups or nausea and vomiting
- Dizziness, confusion

How is NMOSD diagnosed?

A diagnosis of NMOSD begins with medical history, questions about signs and symptoms, a neurological examination and will include a number of other diagnostic tests.

Neurological Examination

A key step in the process of diagnosing NMOSD is a thorough examination by a qualified neurologist. The neurologist will examine an individual for two types of signs and symptoms:

- Cognitive functions such as thinking, logic, memory, and speech.
- Sensory functions such as vision, sensations (touch, taste or smell), muscle strength, balance, reflexes, and coordination.

Blood testing for antibodies

Blood tests that can detect anti-MOG IgG and anti-AQP4 IgG antibodies (or NMO-IgG) are available. Some people may test negative for NMO-IgG, but still have NMOSD. Around one-third of adults with the clinical syndrome of NMOSD who test negative for anti-AQP4 antibodies may be positive for autoantibodies directed against myelin oligodendrocyte glycoprotein (MOG-IgG). Children with demyelinating syndromes are more likely to be positive for MOG-IgG antibodies than AQP-4 IgG antibodies.

Magnetic Resonance Imaging (MRI)

MRI test results often show lesions indicative of inflammation in the spinal cord, optic nerve(s), brainstem, hypothalamus and occasionally in the brain.

Lumbar Puncture (Spinal Tap)

The lumbar puncture tests the cerebrospinal fluid (CSF) for levels of immune cells, proteins, and antibodies. In NMOSD, the CSF may show elevated white blood cell counts during the initial neurological episodes or during relapses.

Ophthalmological Tests (Vision Tests)

People may be referred to an ophthalmologist or neurological eye specialist known as a neuro-ophthalmologist to assess if there is vision impairment or loss due to damage or inflammation (optic neuritis). Visual testing will include both special testing to evaluate the structure of the back of the eye, as well as testing to look for abnormalities in color vision, contrast sensitivity, or one's field of vision.

What can I expect in the course of disease?

NMOSD is a chronic relapsing disorder, where acute attacks occur intermittently if inflammation is not controlled. Unlike other disorders like MS, current thinking does not suggest ongoing, active injury with slow progression between attacks in most people with NMOSD. However, each attack in AQP-4 related NMOSD may cause permanent neurological disability. MOG-IgG associated NMOSD is more likely than AQP-4 related NMOSD to be associated with good recovery after

an attack. NMOSD symptoms may develop quickly — even within a few hours — increase over the course of a few days and then plateau. Symptoms may improve over weeks and months with treatment.

Lasting signs and symptoms of NMOSD may differ in each person and vary according to many factors, including:

- The severity and degree of recovery from the first attack
- The number and frequency of subsequent relapses
- The effectiveness of therapies
- Other co-existing conditions or autoimmune disorders, if they are present or develop
- Gender
- Age
- Pregnancy
- Which antibody is causing the NMOSD symptoms

Early diagnosis and treatment may reduce the relapse rate and/or lessen the severity of relapses should they occur.

Recognizing an NMOSD Relapse (Attack)

People living with NMOSD can experience a recurrence of similar symptoms due to inflammation of the optic nerves and spinal cord, mimicking the initial or onset episode. Such symptoms may also be after-effects of a prior episode, referred to as “ghost” or residual pain following an attack. It is important to determine whether such symptoms represent a new relapse, or the lingering effects of a previous attack. Maintaining regular communication with your healthcare team and seeing a neurologist immediately if there are unresolved symptoms is recommended.

How is NMOSD treated?

There are a variety of effective treatment strategies for individuals who are both AQP4-IgG positive and negative. In addition to medications, there are non-medicinal therapies such as physiotherapy and rehabilitation.

AQP4 seropositive treatment options

Current disease-modifying therapies (DMTs) are effective in suppressing the inflammation that occurs in NMOSD and can prevent relapses of people with NMOSD who are AQP4-IgG positive. For more information about disease-modifying therapies please visit mssociety.ca or connect with a Navigator at 1-844-859-6789 or msnavigators@mssociety.ca

Prevention of NMOSD attacks

Individuals who are not AQP4-IgG positive may prevent attacks with other types of medications following careful assessment by a neurologist. Individuals may be prescribed an immunosuppressant – which slows down the activity of the immune system – to prevent further attacks. NMOSD does not progress between relapses so preventing attacks is essential.

Treatment of NMOSD attacks

Standard treatment involves high dose intravenous steroids, and sometimes, additional treatments (intravenous immunoglobulin or plasmapheresis/plasma exchange). These treatments should be started as soon as possible at onset of a severe attack.

Steroids

A course of methylprednisolone, a type of corticosteroid, is usually given to manage acute attacks of NMOSD – either intravenously or orally. Steroids work to reduce inflammation and can decrease the severity and duration of NMOSD relapses.

Plasma exchange

If a course of steroids hasn't helped your attack, or if your attacks have progressed, you may be offered plasma exchange (PE). This procedure involves taking some of your blood and mechanically separating the blood cells from the fluid (plasma). The blood cells are then mixed with a replacement solution (typically albumin or a synthetic fluid with properties like plasma). The solution with the blood is then returned to your body. PE temporarily clears the blood of antibodies that attack the myelin.

Immunosuppressive medications

If a course of PE doesn't help the attack, some doctors may use a therapy to 'reboot' the immune system. It is important to speak with your doctor about the range of possible side effects from these medications.

Intravenous immunoglobulin (IVIg)

IVIg is a blood product containing the plasma of thousands of donors and is administered by intravenous infusion. IVIg works to reduce inflammatory attacks. In some cases, it is given in combination with or after high-dose steroids.

Treatment of symptoms

Symptoms such as neuropathy (nerve pain), pain, stiffness, muscle spasms, bladder and bowel control problems and depression and anxiety can be managed with various medications and therapies.

Physical activity

Exercise is recognized as an important step toward wellness when living with a chronic health condition. The benefits of exercise are numerous and include improvement in balance and coordination, prevent stiffness, increase energy, decrease pain, as well as improvement in muscle tone, strength and overall endurance. Before you begin any form of exercise, please be sure to consult your healthcare provider. You may also be referred to a physiotherapist or occupational therapist.

Physiotherapists (PTs) evaluate your movement and functioning—including your strength, mobility, balance, posture, fatigue, and pain management. PTs can help you meet the physical challenges and demands of your life. They can also suggest an appropriate exercise regime and should be consulted regarding the proper use of mobility aids such as orthotics (shoe inserts or braces), canes, crutches, walkers, wheelchairs, or scooters.

Occupational therapists (OTs) help work on the everyday skills that you need to function as independently as possible at home and at work. They target upper body strength, mobility, and coordination and can help you use assistive technologies to increase ease of access and independence.

Healthy Eating

There is no special diet for NMOSD. Many people find they can improve their quality of life and sense of well-being by focusing on aspects of health that can be controlled and changed – such as eating a nutritionally balanced diet as recommended by Canada’s Food Guide. This, combined with appropriate exercise, can help control weight, decrease fatigue, maintain regular bowel and bladder function, minimize the risk of skin problems, keep bones healthy and strong, maintain healthy teeth and gums and strengthen the heart.

Additional Resources

The Sumaira Foundation



The Sumaira Foundation for NMO is a 501(c)(3) organization dedicated to generating global awareness of neuromyelitis optica spectrum disorder (NMOSD), fundraising to help find a cure, and creating a community of support for patients + their caregivers.

www.sumairafoundation.org

info@sumairafoundation.org

Contact Us

Contact a Navigator with the MS Knowledge Network to learn about what programs and services are offered in your community. Navigators provide trusted, consistent, quality information and support and are available to assist anyone in Canada, from 8am to 8pm ET, Monday to Friday.

Phone:1-844-859-6789

Email: msnavigators@mssociety.ca

Live Web Chat: visit the **MS Information** or **Support & Services** sections of our website.
