HSS

# **HealthConnection**

HOSPITAL FOR SPECIAL SURGERY'S GOOD HEALTH NEWSLETTER

#### NEUROMUSCULAR CONDITIONS

WINTER 2018

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Check with your doctor before embarking on any new diet or exercise program.

No use of medication should be initiated without first consulting with your healthcare provider regarding the potential benefits and potential risks of use.

# When Muscles and Nerves Don't Talk

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Every day, nerve cells ("neurons") in your brain and spinal cord send a continuous stream of messages to the muscles of your body, telling them what to do. You control these messages each time you move—from the moment you wake up and decide to get out of bed, through showering and dressing, walking or driving to work, eating your meals, interacting with family, friends, and coworkers—and all the way until the end of your day, when you wind down and climb back into bed.

Now imagine that the highway of communication between your nerves and muscles is impaired. Perhaps the road is washed out at some point, or there are too many potholes for the messages to get through smoothly—or at all. When neurons become unhealthy or die, communication between your nervous system and your muscles breaks down, causing the muscles to gradually weaken and waste away.

This is what happens in people with neuromuscular disorders, which can cause twitching, cramps, aches and pains, joint problems, trouble walking or balancing, weakness, and numbness. As the nerves controlling voluntary muscle movement are damaged, everyday tasks we take for granted can become a major chore. Treatments for these conditions vary widely, and a neurologist can review your options with you. Research is actively under way across the country and around the world to better understand the causes of neuromuscular disorders that have no cure. Targeted therapies have already been developed for some serious illnesses that slow disease progression and reduce symptoms while scientists continue to search for a cure. In the meantime, healthcare providers collaborate to provide therapy, education, and support to patients and their caregivers to optimize independence, comfort, and quality of life.

#### Is It in the Genes?

While many neuromuscular disorders arise without any known cause, others are related to genetic mutations that can be passed on in families. Genetic counseling is beneficial for these families to identify family members who may be at risk. Examples include:

- Spinal muscular atrophy (SMA) is caused by an abnormal or missing gene known as SMN1, which the body needs to make a protein required by motor neurons (the nerve cells controlling movement). SMA affects motor neurons in the spinal cord, impairing walking, crawling, breathing, swallowing, and control of the head and neck.
- Muscular dystrophies (MD) is a group of more than 30 genetic diseases characterized by progressive weakness and degeneration of the skeletal muscles and sometimes the heart muscle. Some forms of MD arise early in life, such as Duchenne (the most common form) and Becker, while others (such as facioscapulohumeral MD and myotonic MD) don't cause symptoms until the teenage or adult years.



- Some forms of peripheral neuropathy (numbness and weakness in the arms and legs) are attributable to genetic errors. Mutations in the SCN genes can cause a disorder called small fiber neuropathy, which causes severe attacks of stabbing or burning pain starting in the feet or hands and eventually other parts of the body. Patients with small fiber neuropathy cannot feel pain concentrated in a small area, such as a pin prick, but they have an increased sensitivity to pain in general, and sometimes difficulty differentiating hot from cold.
- Pompe disease is a rare and often fatal illness that disables the heart and skeletal muscles and is caused by a mutation in a gene that makes an important enzyme called GAA.
- In some people, such as those with amyotrophic lateral sclerosis (ALS or "Lou Gehrig's disease"), the weakness first strikes the voluntary muscles—causing trouble walking, writing, or speaking—and gradually extends to those controlling respiration, eventually depriving patients of the ability to breathe on their own. About 10 percent of these cases are inherited.



Some non-inherited ("acquired") neuromuscular diseases are caused by autoimmune disorders, where the immune system goes into overdrive and attacks the body's own tissues. Examples include myasthenia gravis, myositis, and multiple sclerosis. Other acquired neuromuscular disorders may be caused by age—such as spinal degeneration, arthritis, or spinal stenosis that causes nerve problems sports injuries, overuse injuries (such as carpal tunnel syndrome), or "trapped nerves" in the elbow, knee, ankle, or shoulder. (**See page 5** to learn more about common neuromuscular conditions.)

Many neuromuscular disorders can be relieved with medication, injections, physical therapy, and in some cases, surgery. But others are more difficult to treat. If you have any neuromuscular symptoms that won't go away or continue to get worse, see your primary care physician for a referral to a neurologist right away for a full evaluation. (See the article on page 3 to learn more.) The sooner you receive a diagnosis, the quicker you can learn what you can do to feel better.

# Diagnosing Neuromuscular Disorders

When It's Time to See a Doctor

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Neuromuscular disorders may present in many ways, but most commonly with numbness, tingling, weakness, or pain. You should generally consider seeing your primary care physician if your symptoms persist or worsen. If you are developing severe or rapidly progressive weakness or if you have any breathing issues, an emergency room may be the best place to start. You may be referred to a neurologist for more detailed evaluation and diagnostic testing. The goal is to determine the cause of your symptoms and offer the best course of management for you.

#### What Are the Symptoms?

Neuromuscular disorders can cause a variety of symptoms, such as:

- Numbness, tingling, and/or burning in the arms or legs
- Pain that radiates from the back or neck into the legs or arms
- Muscle weakness, cramping, or fatigue that may impair your ability to perform daily activities; however, general fatigue and lack of energy are less often due to a neurologic condition
- Walking and balance problems

Pain in the joints, localized back pain, and localized muscle spasm are usually not neurologic. If you have these symptoms, start with a visit to your primary care physician.



#### Making the Diagnosis

Because the symptoms of neuromuscular disorders are so nonspecific, your doctor will run a series of tests to try to figure out the cause. To prepare for your visit, try to write down as accurately as possible the chronology of your symptoms and the dates, nature, and outcomes of any consultations, tests, and procedures you've already had. Make sure to bring copies of these reports with you. Be prepared to answer questions such as:

- What are you feeling? What is the nature, location, and pattern of your symptoms?
- When did your symptoms begin, and how did they progress?
- Do they get worse or better with certain activities or at certain times of day?
- Are the symptoms impairing your ability to perform your routine daily activities, such as grooming, eating, cooking, or writing?
- Does your family have a history of similar symptoms? (This question can determine if there may be a genetic cause.)

You will have a neurological examination, which will involve assessing your strength, sensation, and reflexes, among other things.

The neurologist may order tests, including:

- Imaging exams such as magnetic resonance imaging (MRI), CT scanning, or ultrasound, which can be used to visualize disc herniation or other spine issues, as well as nerve compression or inflammation.
- Electrodiagnostic tests: nerve conduction studies and electromyography help to localize the problem; determine whether it may be related to the nerves, muscles, or something else; and assess your prognosis.
- Skin biopsy to look for nerve fiber loss, and sometimes muscle or nerve biopsy.
- Blood, urine, or spinal fluid testing to look for underlying causes of the condition.

#### **Patient Care Providers**

The ultimate goal is to figure out the cause of your neuromuscular symptoms and treat them, or at least make you more comfortable. Treatment options may include medication, physical therapy, or referral to another specialist. Healthcare providers who care for people with neuromuscular disorders include:

- Neurologists
- Physiatrists (rehabilitation medicine physicians)
- Orthopedic surgeons, including surgeons that specialize in spine surgery
- Rheumatologists (doctors specializing in joint and autoimmune problems)
- Pulmonologists (for people with disorders that affect breathing)
- Physical, occupational, speech/swallowing, and respiratory therapists
- Registered dietitians, who support patients' nutritional needs
- Nurses and nurse practitioners



Your providers support you and work closely with you and your caregivers to help you achieve the best quality of life possible.

# Common Neuromuscular Conditions

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There are literally hundreds of types of neuromuscular disorders, ranging from common conditions such as carpal tunnel syndrome in the wrist to rarer conditions like ALS. Here is a guide to some of the most common neuromuscular disorders and how they are treated.

#### **Carpal Tunnel Syndrome**

**What It Is:** Pressure on the median nerve, which runs from the forearm into the palm of the hand. The carpal tunnel is a narrow rigid pathway of ligaments and bones at the wrist that houses the median nerve and the tendons that bend the fingers.

May be caused by trauma or injury (such as repetitive stress), thyroid disease, rheumatoid arthritis, or fluid retention during pregnancy.

#### Symptoms:

- Pain, numbness, tingling, and weakness in the wrist.
- Difficulty driving, grasping small objects, or doing other tasks with the hands.

#### Treatments:

- Wrist splint worn at night.
- Nonsteroidal anti-inflammatory and other drugs to reduce pain.
- Injections of steroids such as prednisone.
- Occupational therapy to improve use of the hand and wrist and to reduce symptoms and muscle loss.
- Surgery when nonsurgical therapies don't work.

#### **Peripheral Neuropathy**

**What It Is:** Dysfunction of the peripheral nerves in the arms and legs.

May be caused by diabetes, toxins (such as heavy metals and high doses of vitamin B6), autoimmune diseases, nutritional deficiencies (such as vitamin B12), certain medications (including some cancer chemotherapies), alcoholism, a blood disorder called monoclonal gammopathy of undetermined significance (MGUS), and various metabolic disorders. More rarely, it may be genetic. It is not uncommon that a cause is not always identified.

#### Symptoms:

- Numbness, tingling, prickly sensations, sensitivity to touch, burning pain, balance problems, and muscle weakness.
- Difficulty driving, grasping small objects, or doing other tasks with the hands.

#### **Treatments:**

- Treatment is aimed at reducing symptoms, as well as treating the disorder causing peripheral neuropathy when one is identified.
- Treatment may include medications for neuropathic pain, vitamin supplementation in cases of nutritional deficiencies, avoiding exposure to toxins, or treating the underlying systemic disease causing peripheral neuropathy.
- Physical therapy is useful when weakness or poor balance are present.

#### **Myositis**

**What It Is:** Inflammation of the muscles involved in voluntary movements, which may be caused by injury, infection, certain medications, or an autoimmune disease. May occur on their own, or as part of a systemic disease. Examples include polymyositis and dermatomyositis.

#### Symptoms:

- Polymyositis causes muscle weakness, usually in the muscles closest to the trunk of the body.
- Dermatomyositis causes muscle weakness plus a skin rash.
- Other symptoms may include fatigue after walking or standing, tripping or falling, or trouble swallowing or breathing.
- Difficulty driving, grasping small objects, or doing other tasks with the hands.

#### **Treatments:**

- There are therapies that focus on reducing the underlying inflammation causing symptoms. High doses of a corticosteroid (such as prednisone) can help people with polymyositis and dermatomyositis.
- Various other medications aim to reduce the underlying immune response causing the disease. Examples include intravenous immunoglobulin, mycophenolate mofetil, and methotrexate, among others.





#### **Sports- or Trauma-Related Nerve Entrapment**

**What It Is:** Pressure on a nerve due to an injury or overuse. Examples include compression of the ulnar nerve (cubital tunnel syndrome) in the elbow, posterior tibial nerve in the foot (tarsal tunnel syndrome), nerves in the space between the collarbone and first rib (thoracic outlet syndrome), peroneal nerve at the knee (causing peroneal neuropathy), and suprascapular nerve in the shoulder.

#### Symptoms:

- Numbness and tingling in the affected part of the body.
- Sometimes muscle weakness and pain.

#### Treatments:

- Many of these injuries get better on their own. Nonsteroidal anti-inflammatory drugs and splints can help.
- Physical therapy or occupational therapy to correct any imbalances causing the entrapment and increase flexibility and strength may be effective.
- People with pain due to nerve entrapment may achieve relief with steroid injections.
- When nonsurgical treatments are not sufficient to relieve symptoms, surgery may be necessary to release pressure on the nerve.

If you have any of the symptoms listed here and they don't seem to be getting better on their own, seeing your doctor to determine their cause will put you on the road to feeling better.

# **Building Strength, Maintaining Nutrition**

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For people with the most serious and incurable neuromuscular disorders—such as ALS, muscular dystrophy, or spinal muscular atrophy—building strength, maintaining flexibility, maximizing mobility, and getting enough nutrients are essential components of care. Therapists are therefore critical members of their multidisciplinary care teams, including physical and occupational therapists, speech/swallowing therapists, and registered dietitians.

#### Keeping Patients—and Caregivers—Strong and Mobile

Exercise can provide physiological and psychological benefits for people with neuromuscular disorders, improving quality of life. Research shows that moderate intensity, low-resistance exercise can be beneficial. Before beginning any exercise program, it is important for patients to discuss any restrictions with their doctors.

Physical and occupational therapies can maximize existing capabilities, decrease pain and functional loss, and increase range of motion. The key is to remain active and mobile to prevent further muscle atrophy while enjoying daily activities. Exercise programs may include:

- Cardiovascular conditioning, such as walking, aquatic therapy, and recumbent cycling.
- Stretching and range of motion activities to reduce pain and stiffness and improve posture.

 Strengthening exercises, which should focus on activities to improve function, while avoiding heavy resistance or trying to strengthen already atrophied muscles.

A physical therapist can also educate patients about the use of assistive devices (such as canes or walkers), an ankle foot orthosis (a brace worn on the ankle and lower leg) to prevent foot drag and reduce falls, and wheelchairs to promote independence and reduce fatigue.

Occupational therapy addresses fine motor skills and activities of daily living, such as self-feeding, dressing, and grooming. Therapists work one-on-one with patients to teach them how to conserve energy and use their bodies in the most efficient way possible. Patients may learn how to use widegrip utensils, buttoning aids, braces, walkers, canes, and other devices to make mobility and daily activities easier. Some patients are hesitant to accept these devices because they feel it means they are losing the battle against their illness. But therapists remind them that learning how to expend less energy to perform a routine task, such as brushing teeth, means they will have more energy for another activity that they really enjoy like gardening.

Caregivers can benefit, too, by learning from physical therapists how to move a patient without injuring themselves. Many caregivers don't have the help they need, so learning to lift or move a loved one in the safest, most efficient way possible is extremely important for their own health.

#### Maintaining Good Nutrition

Routine activities that you do each day may require significant effort for someone with a neuromuscular disorder. For example, while you may get dressed in 5 or 10 minutes, someone with ALS may require 30 to 45 minutes to achieve that goal. They may therefore expend far more calories performing their activities of daily living than the average person, requiring them to take in more nutrients each day.

It is the job of registered dietitians and speech/swallowing therapists to ensure that people with neuromuscular disorders receive adequate caloric intake to compensate for their increased energy needs. Consuming protein at each meal can help to preserve muscle mass. Preliminary research suggests that medium chain triglycerides such as coconut oil may help slow progression of illnesses such as ALS; studies are under way to see if this holds true in patients. Fat is also an excellent source of calories. since a gram of fat provides 9 calories (compared with 4 calories each for a gram of protein or carbohydrate).

For people whose illness affects their swallowing and digestive motility, swallowing therapists and nutritionists work together to help patients find ways to eat what they want safely. Oral nutrition supplements and protein powders can maximize nutrient intake. Occupational therapists can help with self-feeding by educating patients how to use devices such as a high-low table, which minimizes the distance someone has to raise a utensil to move food from a plate to the mouth.

Despite this support, there may come a time when a feeding tube should be considered. With percutaneous endoscopic gastrostomy (PEG), a flexible feeding tube is placed through the abdominal wall and into the stomach. PEG allows nutrition. fluids and/or medications to be put directly into the patient's stomach, bypassing the mouth and esophagus. The timing of this placement can be challenging to determine, since it deprives the patient of many of the pleasures of eating. The dietitian looks at the length of meal times, weight changes, and overall ability to meet a patient's estimated

nutrient needs to help determine when it may be time for a feeding tube. People using a PEG may still be able to have some of their favorite foods, such as ice cream, while receiving the majority of their nutrition through the tube.

Nutritional guidance and supportive therapies are integral components of care because they keep patients as active and engaged as possible in their daily lives.

If you or a loved one has been diagnosed with a neuromuscular disorder, be sure to begin a therapeutic program as soon as possible to ensure you have the best quality of life.





## Hope on the Horizon

**Advances in Neuromuscular Research** 

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For years, the diagnosis of a serious neuromuscular disorder carried with it no hope for a cure. Yet advances in molecular biology are changing the futures of people with these diseases, providing hope for some and extending the lives of others. Scientists have identified altered proteins involved in the progression of many diseases, and in some cases pinpointed inherited genetic mutations involved in their development. These advances have led to the design of novel targeted therapies that—while not outright curing the disease—slow its progression and reduce the severity of symptoms so patients have a better quality of life, and often a longer life. Here are a few examples of those successes:

#### Eculizumab (Soliris<sup>®</sup>) for generalized myasthenia gravis (MG). Patients with this disorder may have trouble speaking and swallowing, double or blurred vision, shortness of breath and episodes of respiratory failure, and need assistance walking. In a form of MG that is positive for an antibody called AchR, the immune system turns on itself and produces antibodies against AchR, which plays an important role in nerve-to-muscle communication. Eculizumab blocks a substance in the immune system which injures the muscle surface in people with AchR-positive MG; the drug improves activities of daily living and reduces muscle weakness. It was approved for MG by the U.S. Food and Drug Administration (FDA) in October 2017.

- Nusinersen (Spinraza®) for spinal muscular atrophy (SMA). People with SMA lack sufficient levels of a protein called SMN, which motor neurons need to function. Nusinersen increases SMN levels when given directly into the spinal canal and has been shown to improve motor function in patients with SMA, especially when given early in the course of the illness. The FDA approved the drug in December 2016.
- Eteplirsen (Exondys 51<sup>™</sup>) for Duchenne muscular dystrophy (DMD). Patients with DMD are missing the protein dystrophin, which maintains muscle integrity. Eteplirsen was approved by the FDA in September 2016 for patients with a particular genetic mutation in the DMD gene, which governs dystrophin production. Eteplirsen has been shown to raise dystrophin levels in skeletal muscle.

Alglucosidase alfa (Lumizyme®) for Pompe disease. Lack or insufficiency of a vital enzyme called GAA causes Pompe disease, disabling the heart and skeletal muscles. Alglucosidase alfa replaces this missing enzyme and has been shown to improve function and extend lives in patients with Pompe disease. It was first approved by the FDA in 2006.

Edaravone (Radicava®) for ALS. This medication has been shown to slow the decline in the loss of physical function in people with ALS by scavenging free radicals substances that are part of cellular processes and are normally cleared by the body, but may be one of the causes of nerve cell death in ALS. Edaravone was approved by the FDA in May 2017.

Note: None of the medications described are effective in all patients with these diseases, and all medications carry a risk of side effects. No medication should be used without first discussing its potential benefits and risks with your healthcare provider.

#### Restoring Mobility and Support

Other advances against neuromuscular disorders occur when neuroscientists and engineers team up to combine their expertise. A battery-powered robotic "exoskeleton" that someone wears and operates may reduce the amount of energy and muscle exertion needed to initiate and control the process of walking, for example. Even a robotic arm device may help a patient accomplish daily feeding and grooming more efficiently. The ultimate goal is to devise an exoskeleton that would be wired to the patient's brain and controlled by his or her own nerve impulses; research is under way in several laboratories to develop such a device.







Treatments using stem cells and gene therapy are showing promise in clinical trials.

**Stem cells** are retrieved from a patient's body, modified to produce factors that promote the growth of motor neurons, and placed into the spinal cord of patients with ALS. One approach under clinical study uses stem cells that mature into glial cells that produce a growth factor called GDNF. In another approach, modified stem cells that are placed into the spinal cord produce neuron-promoting growth factors that are taken up by neurons and glial cells. The latter method, now being assessed in a Phase III clinical trial, is expected to slow disease progression, alleviate symptoms, and improve patient survival. **Gene therapy** uses inactivated viruses that serve as vectors to ferry genetic material or functional proteins to cells to correct genetic defects causing neuromuscular disorders. A Phase I clinical trial of VM202-a piece of circular DNA that contains a protein called HGF which improves neuron survival and growth—improved function in people with ALS. Another early-stage study found that AVXS-101, a gene therapy designed to deliver a functional copy of the SMN1 gene to motor neurons, improved motor function in all 15 infants with SMA who received it, helping many to control their head movement or sit unaided. A Phase III study of this promising treatment is now under way.

# **Programs and Resources**

Hospital for Special Surgery offers a variety of wellness exercise classes designed to help you gain endurance, strength and flexibility. Meditation, relaxation and general wellness programs are also offered.

#### Pain and Stress Management Series

This series includes lectures and workshops that introduce techniques such as yoga, meditation, and exercise to manage pain and stress.

#### Therapeutic Yoga

The slow, controlled physical movement of yoga can provide pain relief, relax stiff muscles, ease sore joints and help build strength.

#### **Pilates**

A series of specific movements designed to strengthen the powerhouse muscles of the abdomen, back and waist.

#### Yogalates

A popular form of exercise that blends the best of yoga and Pilates.

#### T'ai Chi Chih®

Simple, rhythmic movements that provide benefits such as improved balance, strength, flexibility and maintenance of bone mass.

#### **Dance for Fitness and Fun**

Studies have shown that dance maintains cardiovascular fitness, enhances emotional well-being, strengthens weight-bearing bones and slows loss of bone mass.

For more information on the schedule, location and cost of these classes, visit **hss.edu/pped** or call **212.606.1613**. Additional programs and offerings can be found by visiting **hss.edu/pped**.

#### Integrative Care Center (ICC)

The ICC, located in mid-Manhattan and affiliated with Hospital for Special Surgery, offers alternative care services including Pilates, acupuncture, massage therapy, chiropractic medicine and pain management. Please visit hss.edu/icc for more information or call **212.224.7900**.

### 2017 Report to the Community



The HSS Community Benefit Report provides information about the Hospital's contributions to the community in the areas of community programs and services, research and health professional education. Visit **hss.edu/community** for more information and to download a copy of the 2017 Community Benefit Report and the 2016-17 HSS Community Service plan or the Community Health Needs Assessment.

### **Neuromuscular Medicine Resources**

- ALS Association: alsa.org
- Brain & Life: brainandlife.org
- The Foundation for Peripheral Neuropathy: foundationforpn.org
- Muscular Dystrophy Association: mda.org
- National Institute of Neurological Disorders and Stroke: ninds.nih.gov/Disorders/Patient-Caregiver-Education

### **Health Video Library**

Check out our complimentary HSS health video library at hss.edu/health-videos. Featured topics include:

- Active and Aging
- Osteoarthritis
- Bones Health
- Pain and Stress Management
- Inflammatory Arthritis
- Health and Wellness

A short video excerpt on **"Meditation for Pain Management"** is also available for patients via **our YouTube playlist**, *Education for Public and Patients.* 

### HealthConnection FastFacts



This recurring publication is a convenient one-page online health education newsletter designed to provide the public with fast, current, accurate musculoskeletal, and general health information.

View the latest edition at **hss.edu/hcfastfacts**.

For more information, visit **hss.edu**.

To make an appointment, call our Physician Referral Service at **800.796.0486**.

Most major insurance plans are accepted.

#### **Financial Assistance**

Hospital for Special Surgery (HSS) offers a Financial Assistance Program to patients who have limited or no insurance coverage and experience difficulty in meeting their financial responsibility for our services. For further information, please contact the HSS Financial Advisory Department at 212.606.1505 or visit hss.edu/ financial-assistance.

**2014 National Health Information Bronze Award HealthConnection Newsletter** Winter 2014 Aging Well issue

#### 2015 Graphic Design USA Health and Wellness Design Award HealthConnection Newsletter Summer 2015, Healthy Feet issue

**2016 Gold Aster Award HealthConnection Newsletter** Winter 2016, Keeping Your Children Healthy Issue

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HSS Ortho Injury Care 1233 2nd Avenue at 65th Street New York, NY 10065

**HSS Long Island Outpatient Center** 333 Earle Ovington Boulevard, Suite 106 Uniondale, NY 11553

**HSS Paramus Outpatient Center** 140 East Ridgewood Avenue, Suite 175 S Paramus, NJ 07652

**HSS Queens Outpatient Center** 176-60 Union Turnpike, Suite 190 Fresh Meadows, NY 11360

HSS Stamford Outpatient Center 1 Blachley Road Stamford, CT 06902

HSS Orthopedics at Stamford Health Stamford Hospital One Hospital Plaza Stamford, CT 06904

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**Integrative Care Center** 635 Madison Avenue, 5th Floor New York, NY 10022 The Public and Patient Education Department of Education Institute provides information to the general public and patients through a variety of health education programs. Professionals provide practical information to help prevent or manage orthopedic and rheumatological conditions. Programs are held at the hospital as well as in the community. The department is dedicated to providing education today, so that everyone can have a healthier tomorrow.

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