

Ehlers-Danlos Syndrome

Condition: Ehlers-Danlos Syndrome (EDS) is a group of disorders that affect the skin, joints, internal organs and/or blood vessels.

Background: The actual prevalence of EDS is unknown because many patients are never diagnosed, particularly if they only have one symptom. Presentation may be in childhood or later in life when more vascular complaints are identified.

Risk Factors: EDS is a genetic disorder, which means it is passed from parent to child. If a parent has one of the most common types of EDS, there's a 50% chance their child or children will be born with the same EDS gene.

History and Symptoms: Children can present with developmental delay, easy bruising, and floppiness. EDS patients often have unusually flexible joints, loose skin and skin that scars easily and does not heal well. Some people with EDS only have one symptom, others have more, and they can range from mild to severe. It is important to know that symptoms can also vary widely among the different types of EDS.

For example, a baby with one of the more common types may be born with dislocated hips; an individual with EDS affecting the blood vessels may have nearly translucent skin with visible veins; patients with Kyphoscoliosis EDS may have eye issues and scoliosis. The only form of EDS with shortened life expectancy is the Vascular which can present with a significant medical problem by age 40, involving rupture of blood vessels and bowel involvement. Patients can have problems with hypotension (low blood pressure) as well as difficulties with gastrointestinal motility, causing constipation.

Physical Exam: In addition to examining skin and joints, the physical medicine and rehabilitation (PM&R) physician (rehab physician) may ask patients to bend and place their hands flat on the floor without bending the knees, or other tests that make up the Beighton Score, to check for hypermobility. PMR physicians also can evaluate gait patterns to make appropriate recommendations. Other exams may be performed to help identify specific type of EDS.

Diagnostic Process: Diagnosis of EDS is confirmed by skin biopsy and blood sample test to perform genetic test and rule out other issues. Cardiac, abdominal and thoracic imaging are recommended prior to clearance for competitive sports.

Rehab Management: Rehabilitation should focus on improving posture and spatial orientation, building strength and motor control, stabilizing joints, and increasing physical activity. Education is also important so patients can remain active but do so in a safe manner that includes sports and activities that minimize risk to joints. Medications can also be used to manage pain. Swimming or cycling are non-weight bearing and hence are useful to develop strength and help with balance. Avoid high intensity sports and heavy weightlifting. Help is also provided for activities of daily living like utensils with wide grip to decrease pain, and strain on finger joints. Braces to support feet can be helpful.

School accommodations can also be instituted. PMR physicians are specialists who can help coordinating care, especially interdisciplinary management. Family members should be provided with education about

protection of skin and joints, use of protective gear, avoid contact sports if they have a history of heavy bruising. Patients with EDS involving blood vessels should avoid competitive sports.

Patients should be monitored for early arthritis. Surgery should be cautiously approached due to poor wound healing, and risk of increased bleeding.

Other Resources for Patients and Families: The Ehlers-Danlos National Foundation offers many resources for patients and families, such as [support group information](#) and a [helpline](#).

Frequently Asked Questions

What is PM&R?

Physical medicine and rehabilitation (PM&R), also known as physiatry, is a primary medical specialty that aims to enhance and restore functional ability and improve quality of life to those with injuries, physical impairments or disabilities affecting the brain, spinal cord, nerves, bones, joints, ligaments, muscles and tendons. PM&R physicians, known as physiatrists, evaluate and treat the whole body, maximize patients' independence in their daily life and are experts in designing comprehensive, patient-centered treatment plans to empower patients to achieve their goals. By taking the whole body into account, they can accurately pin-point problems, decrease pain, assist in recovery from devastating injuries and maximize overall outcomes and performance with non-surgical and peri-surgical options. To learn more, visit www.aapmr.org/aboutpmr.

What makes PM&R physicians unique?

PM&R physicians' training focuses not just on treating medical conditions, but on enhancing the patient's performance and quality of life in the context of those medical conditions. They focus not only on one part of the body, but instead on the development of a comprehensive program for putting the pieces of a person's life back together – medically, socially, emotionally and vocationally – after injury or disease. PM&R physicians make and manage medical diagnoses, design a treatment plan and prescribe the therapies that physical therapists or other allied therapists perform or that are carried out by the patients themselves. By providing an appropriate treatment plan, PM&R physicians help patients stay as active as possible at any age. Their broad medical expertise allows them to treat disabling conditions throughout a person's lifetime.

Why see a PM&R physician?

A PM&R physician will thoroughly assess your condition, needs, and expectations and rule out any serious medical illnesses to develop a treatment plan. By understanding your condition and goals, you and your PM&R physician can develop a treatment plan suited to your unique needs.

How do I find a PM&R physician near me?

Visit www.aapmr.org/findapmrphysician or contact your primary care physician for a referral.