

McKusick-Nathans Institute of Genetic Medicine

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Thank you for your inquiry regarding a clinical evaluation regarding your diagnosis of hypermobility and/or joint pain and diagnostic evaluation for the hypermobility type of Ehlers-Danlos syndrome (EDS). Currently, we are only accepting new patients between the ages of 7-21 years for this referral reason. For those beyond that age limit, generalized counseling information is below.

The genetic causes of hypermobility and hypermobile Ehlers-Danlos syndrome are unknown, thus treatment is symptomatic and typically not dependent on a diagnosis. There is currently no genetic test to confirm the diagnosis.

Joint hypermobility can be observed in hundreds of genetic syndromes, however these would typically go along with developmental delay, birth defects, growth issues (overgrowth, failure to thrive). Thus, if there are other indications for genetics evaluation, this should be discussed with your primary care physician and referral could be generated for those reasons. The presence of these additional features could prompt referral to genetics:

1. Short stature: If less than 18 years old, less than 1% for height; >18 years old, males under 5'0" and females under 4'10".
2. Developmental delay or intellectual disability
3. Low muscle tone (hypotonia)
4. Multiple low or no-trauma fractures, or fractures that do not heal
5. Ocular concerns such as retinal detachments, lens dislocation, corneal ruptures, myopia ≥ 7 diopters
6. Personal history of aneurysm or dissection of aorta or arteries
7. Family history of aneurysm or dissection of aorta or arteries
8. Uterine or bowel rupture
9. Collapsed lungs without injury
10. Birth defects such as congenital heart defects, clubfoot, cleft palate, contractures, hip dysplasia
11. Hearing loss
12. Episodic swelling of joints, without trauma; ANA-negative.

If any of these are occurring, please call the main clinic office at 410-955-3071 x1 and request additional intake based on other diagnoses.

Sincerely,

Ada Hamosh, MD
Clinical Director

INFORMATION & RESOURCES FOR JOINT HYPERMOBILITY AND RELATED SYMPTOMS

Hypermobility describes joints that easily move beyond the normal range expected for that particular joint; it tends to be inherited in families in an autosomal dominant pattern (meaning you can see it inherited from a parent, who may have more or less severe symptoms). It can be seen as a somewhat isolated feature causing musculoskeletal pain and joint dislocations/subluxations. Unlike most genetic disorders that are very rare, it is very common in the general population. Many patients with hypermobility have other symptoms/conditions, including anxiety, gastrointestinal problems (slow motility, irritable-bowel syndrome, gluten sensitivities) and orthostatic intolerance (disorder of autonomic function that leads to difficulty tolerating standing and/or sitting for periods of time due to the excessive pooling of blood in the extremities and episodes of syncope, increased heart rate, dilated pupils, and distractibility, difficulty in concentration and exercise intolerance.)

The question of the vascular type of Ehlers-Danlos syndrome often gets raised in patients with joint hypermobility. This subtype of Ehlers-Danlos syndrome is typically NOT associated with widespread joint hypermobility and/or dislocations or subluxations of the large joints. There may be some mild hypermobility of finger joints. People with the vascular type of Ehlers-Danlos syndrome frequently have very fragile skin (friable skin with easy scar formation), severe bruising, collapsed lung(s), and/or aortic/arterial tear or dissection. A genetic test is available for Vascular Ehlers-Danlos syndrome. If there is concern about the vascular type of Ehlers Danlos syndrome, a genetic counseling appointment can be made to discuss genetic testing for this subtype.

Online resources:

- The Ehlers-Danlos Society
 - (<https://www.ehlers-danlos.com>)
 - In 2017, management and care guidelines were produced for hypermobility and hypermobility EDS. The collection of articles can be found at: <https://www.ehlers-danlos.com/2017-eds-international-classification/>
- Ehlers-Danlos Syndrome, hypermobility type Gene Reviews
 - (<https://www.ncbi.nlm.nih.gov/books/NBK1279/>)

Book resources:

- Dr. Bradley Tinkle: "Joint Hypermobility Handbook- A Guide for the Issues & Management of Ehlers-Danlos Syndrome Hypermobility Type and the Hypermobility Syndrome"; amazon.com
- Rosemary J Keer and Rodney Grahame (resource for PTs): "Hypermobility Syndrome: Diagnosis and Management for Physiotherapists"; amazon.com
- Claire Davies (resource for myofascial trigger point release at home to address muscle spasms and myofascial pain): "The Trigger Point Therapy Workbook"; amazon.com
- Mindfulness-based stress reduction can aid in coping with chronic pain. Good resources are Dr. Jon Kabat-Zinn's books called "Full Catastrophe Living: Using the Wisdom of Your Body and Mind to Face Stress, Pain, and Illness" and "Wherever You Go, There You Are."
- JWG Jacobs, LJM Cornelissens, MC Veenhuizen, BCJ Hamel "Ehlers-Danlos Syndrome: A MultiDisciplinary Approach". 370 page e-book (or can order hard copy). <http://ebooks.iospress.nl/ISBN/978-1-61499-878-5>

Below are generalized resources and management tools; any individual should discuss medical concerns and/or initiation of any new therapies with their Primary Care Physician.

- **Joint Surgery:** Joint stabilizing surgeries should be avoided unless absolutely necessary. There is an increased risk of immediate or intermediate-term failure of joint stabilizing surgery in patients with inherited disorders of connective tissue.
- **Physical therapy:** Physical therapy may help with overall fitness, to decrease pain and increase joint stability. This can include physical therapy for muscle relaxation (i.e. massage, deep heat, ultrasound, TENS, etc) as well as myofascial trigger point release to address muscle spasms, myofascial trigger points, and musculoskeletal pain, which should be followed by muscle toning exercises to increase muscle tone around the joint.

Hydrotherapy (swim therapy) and recreational swimming, particularly in a heated pool for added muscle relaxation, can also be helpful. Epsom salts baths or footbaths may help replete magnesium and assist with muscle relaxation. Other resources and tools include wearing compression gloves, Imak arthritis gloves, or ring splints which can limit hyperextension in the small joints of the hands, or using an ergonomic pen.

- **Activity Recommendations:**
 - Regular physical activity is important and can prevent deconditioning. It is usually recommended to avoid, or at least minimize, high-impact activity, and resistance exercises such as running or heavy weight lifting. Those would be expected to worsen the joint instability and pain and increase the risk for the development of osteoarthritis over time.
 - The cornerstone of managing joint laxity and pain is to try to improve joint stability by first relaxing any tense muscles and ultimately increasing muscle tone. Tone is the resting state of muscle contraction and should not be confused with strength. Strength is a voluntary force exerted at will. Exercises should be very low resistance, gradually increasing duration, frequency, or repetitions. Good examples could include walking, bicycling, swimming, water exercise, elliptical trainer, yoga, Pilates, and core toning/stability exercises.
 - It is important to recognize and not exceed current physical limitations, not only in minimizing resistance, but also in avoiding excess repetitions. Some patients can initially tolerate only a few minutes of repetitive motion exercise, and need to be careful not to advance too quickly.
- **Muscle Relaxation:** The current thinking is that a lot of the pain and fatigue associated with joint hypermobility is due to muscle spasm, triggered by the underlying joint instability. Passive treatments such as myofascial release, stretching, acupuncture, and muscle relaxants can bring relief and are worth considering
- **Lightheadedness/Dizziness/Postural orthostatic hypotension (POTS):**
 - (POTS) is defined by excessive heart rate increments upon upright posture.
 - Maximizing blood volume by increasing salt intake and fluid intake (water or drink with electrolytes) is the foundation for management. In addition to adding salt to food, salt tablets are sometimes necessary.
 - Aggressive hydration is recommended. (Practically, someone should drink enough water to have clear urine every 1-2 hours, this may require up to 16 ounces of water every two hours. If this is problematic with work or school setting, please talk with your primary care physician about documentation to have access to water and bathroom breaks.)
 - Increase salt intake. Strategies include adding table salt to food or eating foods with high salt concentrations. One of the easiest solutions is to eat 1-2 pickles per day. Some people find salt tablets (Nuun) to their water more helpful.
 - Some medications may be considered if increased hydration and salt is not effective.
 - Consider cardiology evaluation for any fainting WITHOUT change of positioning.

- **Psychological Approaches:**
 - Some individuals with joint hypermobility and/or chronic pain have significant fatigue, depression and psychological burden. Many find psychological counseling helpful for coping with chronic medical problems, which frequently have significant impact on psychological health and well-being. Achieving better psychological health can reduce the subjective experience of chronic pain and other medical problems. In particular, cognitive behavioral therapy has been shown to improve overall coping and function in patients with hypermobility and pain.
 - Mindfulness-based stress reduction (MBSR) has also been shown to improve overall psychological well-being, specifically in those with joint hypermobility/hypermobility Ehlers-Danlos syndrome.
 - Dr. Neda Gould offers MBSR at the Johns Hopkins Bayview Campus. More information can be found online: https://www.hopkinsmedicine.org/psychiatry/for_faculty_staff/mindfulness/index.html
 - Link to other resources in the Baltimore area: http://www.trishmagyari.com/links_to_resources
 - Find an MBSR program: <https://umassmed.edu/cfm/mindfulness-based-programs/mbsr-courses/find-an-mbsr-program/>
 - Good sleep hygiene, coping skills, and relaxation techniques are helpful in managing chronic pain associated with joint hypermobility.
- **Pain Medication:**
 - Medication for pain may be under prescribed, and should be tailored to the individual's subjective symptoms and objective measures of pain, not to physical examination or radiologic findings.
 - Combinations of multiple medications may be required (rather than a single medication).
 - Prevention or control of pain with regularly scheduled dosing is often more successful than acute treatment with as-needed dosing.
 - Skeletal muscle relaxers and neuropathic pain medications are often useful adjuncts, in addition to analgesics and anti-inflammatories.
- **Irritable Bowel syndrome:**
 - A gastrointestinal evaluation for diagnosis and management of lower GI symptoms may be considered.
 - Management of irritable bowel syndrome is no different in patients with or without underlying hereditary connective tissue disorders. Management typically includes stress reduction, avoidance of dietary triggers, and pharmacologic therapy as needed.