Program of Genetics & Metabolism,
HSC Winnipeg

FE229 820 Sherbrook Street
Winnipeg, MB R3A 1R9
P: (204) 787-2494
F: (204) 787-1419

Dear Doctor

Joint hypermobility is a common finding in the general population. While we would like to be able to offer a genetic appointment to all patients with a suspected diagnosis of hypermobile Ehlers-Danlos Syndrome (EDS), our wait list has unfortunately become unmanageable. **We are therefore returning this referral to you** with general advice that has been helpful to other patients with joint hypermobility.

The differential diagnosis for hypermobile EDS includes a number of heritable connective tissue disorders. Some of these can be associated with thoracic aneurysms/vasculopathy, which is why it is important for you to ensure that your patient does not have a personal or family history of aneurysm, vascular dissection, hollow organ rupture or sudden death.

- All patients suspected of having EDS should have an echocardiogram. If your patient's echocardiogram shows an aortic dilatation, a mitral valve prolapse or a bicuspid aortic valve, please refer them back and include a copy of the echocardiogram with your referral.

- If your patient has a personal or family history of thoracic or brain aneurysm or vascular dissection or hollow organ rupture or sudden death, please also refer them back indicating this history on the referral.

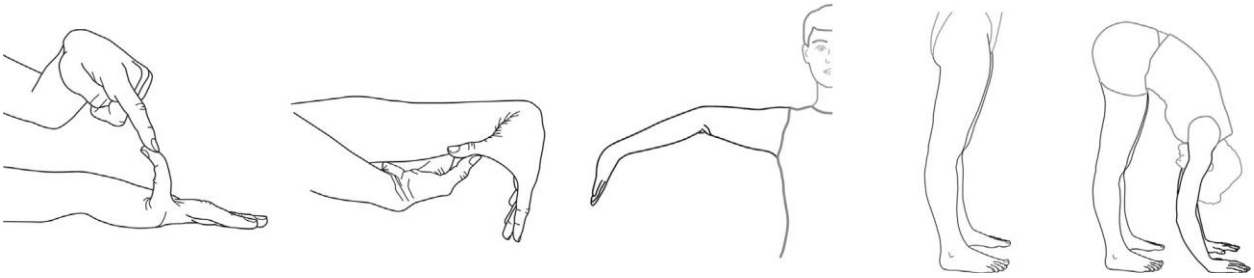
- If your patient's relatives were diagnosed with another subtype of EDS other than hypermobile EDS, please refer your patient back and indicate this history on the referral.

- If your patient displays any of the following, please refer them back and indicate this history on the referral:
    - Very translucent skin
    - Skin hyper extensibility (2 cm or more on the volar surface of the forearm)
    - Atrophic scars
    - Very wide-spaced eyes (hypertelorism)
    - A bifid uvula
    - Intramuscular or joint hematomas
    - Tendon / muscle rupture
    - Extremely large bruises with minimal trauma (not just easy bruising)
    - Pneumothorax

The diagnosis of hypermobile EDS remains clinical as there is yet no reliable genetic test in the vast majority of patients. The diagnostic criteria for hypermobile EDS and other forms of EDS can be found in the following article: Malfait et al, The 2017 *International Classification of the Ehlers–Danlos Syndromes.* American Journal of Medical Genetics Part C (Seminars in Medical Genetics) 175C:8–26 (2017).

According to this guideline, hypermobility is required but is not sufficient for the diagnosis of hypermobile EDS. Other criteria must be met. Hypermobility is defined by the Beighton scoring system i.e. ≥ 6 for pre-pubertal children and adolescents, ≥5 for pubertal men and women up to the age of 50, and ≥4 for those >50 years of age for hypermobile EDS.

Each joint is scored independently as outlined(A) With the palm of the hand and forearm resting on a flat surface with the elbow flexed at 90°, if the metacarpal-phalangeal joint of the fifth finger can be hyperextended more than 90° with respect to the dorsum of the hand, it is considered positive, scoring 1 point. (B) With arms outstretched forward but hand pronated, if the thumb can be passively moved to touch the ipsilateral forearm it is considered positive scoring 1 point. (C) With the arms outstretched to the side and hand supine, if the elbow extends more than 10°, it is considered positive scoring 1 point. (D) While standing, with knees locked in genu recurvatum, if the knee extends more than 10°, it is considered positive scoring 1 point. (E) With knees locked straight and feet together, if the patient can bend forward to place the total palm of both hands flat on the floor just in front of the feet, it is considered positive scoring 1 point. The total possible score is 9



Medical Genetics Clinic
www.wrha.mb.ca/prog/genetics
Enclosure: General Advice Regarding Patients with Joint Hypermobility

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General Advice Regarding Patients with Joint Hypermobility

This handout provides general recommendations regarding management of joint hypermobility and some symptoms that may also be present in patients with joint hypermobility. Joint hypermobility is a primary feature of Ehlers Danlos syndrome hypermobility type (hypermobile EDS) but by itself is not sufficient to make this diagnosis.

Many people with joint hypermobility do not have any symptoms; some people experience joint pains that seem to increase with age.

Exercise is an important component of overall health and wellness and individuals with hypermobility should be encouraged to be physically active. A physiotherapist can help by providing a thorough assessment and developing a tailored treatment plan for an individual with hypermobility. The aim of physiotherapy treatment is to strengthen muscles, improve proprioception, balance and coordination while promoting lifelong fitness through a program that combines targeted exercises and physical activities that the individual enjoys. A physiotherapist can also provide education to help with the management of pain and fatigue to maximize participation in daily activities. Please see information on "How to Find a Physiotherapist" below.

Of note, if joint pains become a chronic pain disorder with additional symptoms such as anxiety, depression, fatigue, and withdrawal from school, work, exercising, physical activities and social life, an approach based on the biopsychosocial model of illness (pain) is strongly indicated. Physical and psychological therapies are warranted in those situations. You can find further information here: https://everychildeverytime.ca/. This website is primarily directed to children but the principles are applicable to adults as well.

Please take the following into account when a patient with joint hypermobility presents to you:

- If your patient suffers from fatigue, counselling on sleep hygiene may be considered as the first approach to improve sleep and fatigue as in other patients; information on sleep hygiene can be found here: https://everychildeverytime.ca/healthy-living/restful-sleep/restful-sleep/
  Of note, while pain and fatigue are often reported and troublesome symptoms in hypermobile EDS, the degree of both pain and fatigue are poorly correlated with the degree of joint hypermobility.
- If your patient does have a tendency to grind or clench their teeth at night, they may consult their dentist to see whether they could benefit from a mouth guard.
- If your patient has migraines, please be aware that tryptans would become contraindicated if your patient was to develop enlargement of the aortic root.
- Cervical spine instability can be a feature in a number of heritable connective tissue disorders. Flexion-extension x-rays can be used as a screening test. If your patient experiences symptoms compatible with nerve compression secondary to cervical spine instability, a referral to a neurosurgeon or orthopedic surgeon for assessment for cervical spine instability may be indicated. Please note that chiropractic manipulations of the neck, particularly in individuals at higher risk of potential vertebral artery dissection because of joint hypermobility, are contraindicated.

- Further information on specific medical management of hypermobile EDS can be found in the references provided below.

How to Find a Physiotherapist

A list of **private physiotherapy clinics** can be found at www.mbphysio.org or on the College of Physiotherapists of Manitoba website at www.manitobaphysio.com. **Central Intake for Pediatric Physiotherapy**: https://mbphysio.org/find-a-physiotherapist/pediatric-central-intake

Review Articles on Joint Hypermobility syndrome/Ehlers-Danlos syndrome:

1. Castori M et al. Re-writing the natural history of pain and related symptoms in the joint hypermobility syndrome/Ehlers–Danlos syndrome, hypermobility type. Am J Med Genet 2013; 161A(12):2989-3004.
2. Castori M, et al. Management of pain and fatigue in the joint hypermobility syndrome (a.k.a. Ehlers–Danlos syndrome, hypermobility type): Principles and proposal for a multidisciplinary approach. Am J Med Genet 2012; 158A(8):2055-70.
3. Colombi M, Dordoni C, Chiarelli N, Ritelli M. Differential diagnosis and diagnostic flow chart of joint hypermobility syndrome/ehlers-danlos syndrome hypermobility type compared to other heritable connective tissue disorders. Am J Med Genet C Semin Med Genet. 2015;169C(1):6-22.
4. Donkervoort S, Bonnemann CG, Loeys B, Jungbluth H, Voermans NC. The neuromuscular differential diagnosis of joint hypermobility. Am J Med Genet C Semin Med Genet. 2015; 169C(1):23-42 .
5. Tinkle et al. Hypermobile Ehlers–Danlos Syndrome (a.k.a. Ehlers–Danlos Syndrome Type III and Ehlers–Danlos Syndrome Hypermobility Type): Clinical Description and Natural History. American Journal of Medical Genetics. 2017;175C:48–69.
6. Engelbert R., et al. The Evidence-Based Rationale for Physical Therapy Treatment of Children, Adolescents, and Adults Diagnosed With Joint Hypermobility Syndrome/Hypermobile Ehlers Danlos Syndrome. American Journal of Medical Genetics 2017; 175C: 158-167.
7. Smith et al. Physiotherapy and occupational therapy interventions for people with benign joint hypermobility syndrome: a systematic review of clinical trials. Disability and Rehabilitation 2014. 36(10):797-80.
8. Hakim et al. Cardiovascular Autonomic Dysfunction in Ehlers–Danlos Syndrome—Hypermobile Type American Journal of Medical Genetics 2017; 175C:168–174.

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