AKRON CHILDREN'S HOSPITAL (ACH)
PROVIDER REFERRAL FOR JOINT HYPERMOBILITY/CONCERN FOR EHLERS-DANLOS SYNDROME

Name:
MRN:
DOB:
Patient Identification

Joint hypermobility (JH), in isolation, is often a common trait that does not indicate the need for genetic testing. These patients may still experience symptoms from JH. Patients may benefit from a referral to a physician who is knowledgeable regarding the symptoms and treatment for JH.

An individual with JH may have additional features that would qualify them for a diagnosis of Hypermobile Ehlers-Danlos Syndrome (most commonly chronic pain, joint dislocations, and family history of similar symptoms). A diagnosis can be made by history and physical examination and there is no genetic test available to help with this diagnosis.

Clinical Genetics is a service that provides consultations for patients with rare conditions and makes recommendations for testing. Geneticists do not have special training in the management or treatment of chronic pain or biomechanical dysfunction. Rheumatology, Pain Management and/or Physical Therapy may be better equipped to help with management of pain and joint instability. In situations where there is a personal or family history of any of the features noted in the table below, genetic evaluation may be considered.

Please check which box(es) most closely matches your clinical suspicion:

A referral for a genetic evaluation is indicated in patients with hypermobility (Beighton score >4) PLUS a personal or family history of the following:	**Personal History**	**Family History**
Aneurysm or dissection of aorta or arteries	☐	☐
Bowel or uterine rupture	☐	☐
Rectal prolapse; pelvic organ prolapse WITHOUT a history of vaginal childbirth	☐	☐
Atraumatic, spontaneous pneumothorax	☐	☐
Retinal detachment, lens dislocation, corneal rupture, myopia > 7 diopters	☐	☐
Marfanoid body habitus	☐	☐
Bilateral hip dysplasia	☐	☐
Abnormal genetic testing	☐	☐
Cleft palate or bifid uvula	☐	☐

*If Box is Checked Please Provide Copy of Results/Evaluation

High Suspicion of life-threatening vascular complications: ☐ Yes *or* ☐ No

If yes, please include a reason for your high clinical suspicion

_____

_____

Please explain: What is the specific question or questions that you want addressed in this consultation:

_____

_____

Referring Provider: _____

Name of Form Preparer: _____

Best Contact Number for Follow-up: _____

Please fax this form back to (330) 543-3677. We will evaluate the information provided and schedule if appropriate. If a genetics appointment is not indicated or no longer of interest, we will provide general recommendations for management of hypermobility

Hypermobility Preventative Guidelines

Thank you for your referral to the Akron Children's Hospital Genetics Clinic for the concern of Joint Hypermobility (JH). Based on the evaluation of the required questionnaire for this type of referral, there is no indication for further evaluation nor genetic testing. In lieu of a Genetics Clinic appointment, some resources and recommendations related to management with a healthcare provider are given.

There is a collection of resources provided below that can help to summarize the causes and prevention techniques of Joint Hypermobility that raise concerns for Ehlers Danlos Syndrome. Patients who do not require genetic testing for their diagnosis of Joint Hypermobility may refer to the references provided to broaden their knowledge on preventative measures and general recommendations (current as of August 2023) based on expert consensus. All general recommendations should be discussed with your Primary Care Provider (PCP). These resources should be reviewed if a healthcare provider has noted joint hypermobility and no other concerning features. These recommendations and guidelines do not substitute for clinical management with a healthcare provider.

1. Joint stability may be improved with low-resistance exercise to increase muscle tone. Examples of low-resistance exercise include walking, bicycling, water exercise, and simple range-of-motion exercises without added resistance. Progress should be made by increasing repetitions, frequency, or duration, not resistance. However, if an individual has osteopenia or osteoporosis there needs to be a balance between exercises that provide some low-level resistance training to enhance bone strength versus exercises to maximize joint stability.
2. Some patients with FJH will benefit from physical therapy. Information about physical therapy for patients with hypermobile joints are available through the following resources:
    a. The book "Hypermobile Syndrome recognition and Management for Physiotherapists" by R. Keer, R. Graham (eds.), Butterworth Heinemann, London, 2003
    b. The journal article "Examination and Treatment of a Patient with Hypermobility" by Leslie Russek in Physical Therapy 2000; 80:386-396.
    c. Through the Hypermobility Syndrome Association in the section on physical therapy at www.hypermobility.org.
3. The literature for fibromyalgia and EDS/joint hypermobility have recently shown the effectiveness of mindfulness techniques in helping chronic pain symptoms. These provide exercises to stop the cycle of thinking about pain and having pain dictate activity. These are not cure-alls but part of a comprehensive plan to help alleviate symptoms over time. Resources are available online (such as https://www.swedish.org/services/painservices/pain-management-guide/mindfulness) and through free apps on phones such as Headspace.
4. Consider following up with a rheumatologist or provider who cares for patients with fibromyalgia as they would be up to date on the best management of symptoms.
5. Gastrointestinal conditions such as Irritable Bowel Syndrome (IBS), gastroesophageal reflux disease (GERD), and gastroparesis can arise; treat according to symptoms with proton pump inhibitors, antispasmodics, antidiarrheals, laxatives, or promotility agents. Awareness of foods that trigger symptoms can also help reduce symptoms.
6. Annual ophthalmologic evaluation to screen for myopia and astigmatism
7. Serotonin/norepinephrine receptor inhibitors (SNRIs), such as venlafaxine and duloxetine, or gabapentin-like treatments which may offer combined benefit for depression, anxiety and neuropathic pain. A trial of the SNRI medications through your primary care provider may be beneficial.
8. Patients with joint hypermobility may be at risk for orthostatic hypotension (low blood pressure and high heart rate on changing positions). If symptomatic, one should consider increasing salt and fluid intake and be more conscious of positional changes to reduce the risk for fainting. You could also consider compression stockings. You can discuss with your PCP if referral to a Cardiologist with special interest in postural tachycardia/orthostatic intolerance is indicated.
9. Easy bruising and bleeding can be seen but does not usually cause major medical problems. Further testing or evaluation with a hematologist is only indicated if there are concerning symptoms or plans for elective surgery with considerable bleeding risks.

The websites listed below can help to provide the most up-to-date information and research found by physicians and researchers that can be used as a reference by patients.

- The Ehlers-Danlos Society
    - https://www.ehlers-danlos.com

- EDS International Consortium
  - https://www.ehlers-danlos.com/international-consortium/
- hEDS Diagnostic Criteria
  - https://www.ehlers-danlos.com/heds-diagnostic-checklist/
- EDS Network CARES, Inc.
  - http://www.ehlersdanlosnetwork.org
- GeneReviews
  - https://www.ncbi.nlm.nih.gov/books/NBK1279/