McKusick-Nathans Institute of Genetic Medicine
600 N. Wolfe Street/Blalock 1008
Baltimore, MD 21287-4922
410-955-3071 T
410-614-9246 F

September 19, 2018

Dear Patient,

Thank you for contacting our office to see a geneticist for the diagnosis or possible diagnosis of Ehlers-Danlos syndrome due to hypermobility or pain issues. We are a diagnosis-only center. Currently, we are able to schedule appointments for individuals between the ages of 7-21 with a geneticist. Any geneticist can see you/your child to establish or rule out the diagnosis of hypermobility type of Ehlers-Danlos syndrome. However, we are not a comprehensive multidisciplinary clinic for this patient population.

For additional resources, we recommend connecting with the Ehlers- Danlos Society (https://www.ehlers-danlos.com), asking your primary care physician for other referral options, or contacting your local geneticist (https://www.acmg.net/; click on "find genetic services."). Specifically, the Ehlers-Danlos Society holds an internal list of physicians that diagnose/manage patients with EDS and they also hold a yearly learning conference for patients. Other support resources include EDS International (http://eds-international.com/) and the EDS Network CARES, Inc. (http://www.ehlersdanlosnetwork.org/).

Many individuals with joint flexibility are looking for management of joint pain. Good information can be found at https://www.genetests.org/ (click on Gene Reviews, type in "Ehlers Danlos", under Ehlers Danlos, hypermobility type click on "reviews.") This may answer many of your questions about the physical findings and management of symptoms. As noted, the Ehlers- Danlos Society has good resources. Finally, some book resources that may be applicable for management tools include:

1. Dr. Bradley Tinkle: "Joint Hypermobility Handbook- A Guide for the Issues & Management of Ehlers-Danlos Syndrome Hypermobility Type and the Hypermobility Syndrome"; www.amazon.com

2. Rosemary J Keer and Rodney Grahame (resource for PTs): "Hypermobility Syndrome: Diagnosis and Management for Physiotherapists"; www.amazon.com

3. Claire Davies (resource for myofascial trigger point release at home to address muscle spasms and myofascial pain): "The Trigger Point Therapy Workbook;" www.amazon. Com (https://www.painscience.com/tutorials/trigger-points.php)

Thank you.

Sincerely,

Ada Hamosh, MD, MPH
Clinical Director, McKusick-Nathans Institute of Genetic Medicine

Hal Dietz, MD
Director, Cardiovascular CTD Clinic

Joann Bodurtha, MD, MPH
Professor of Pediatrics and Oncology

Revised: May 2015

New Patient Appointment Questionnaire
Institute of Genetic Medicine, Johns Hopkins University
Ehlers Danlos syndrome/hypermobility

Dear Patient/Parent,

Thank you for your interest in having a clinic appointment in the Institute of Genetic Medicine at Johns Hopkins.

IMPORTANT: If you are pregnant, OR, have an urgent vascular problem
please DO NOT complete this packet and call 410-955-0316 (Gretchen MacCarrick)

In order for us to prepare for your appointment, we ask that you fill out the attached questionnaire. We are working to have this become a confidential electronic process through MyChart. Once completed, please return the questionnaire back with any additional records by:

Fax: 410-614-9246 **Phone:** 410-955-3071 **Email:** *CTDinfo@jhmi.edu*

Mail: Genetic Counselors--CTD
Institute of Genetic Medicine, Johns Hopkins University
600 N. Wolfe St, Blalock 1008
Baltimore, MD 21287

Once we receive your questionnaire and any additional records, a genetic counselor will review your information and determine the appropriate physician to see the patient and determine if there are any needed ancillary appointments to be scheduled before or on the day of the appointment. Appointments will be scheduled in the order questionnaires are received. We will do our best to have the patient scheduled as soon as possible; however please be patient as we have a high volume of appointment requests. We schedule about 4-6 months in advance, depending on urgency and diagnosis.

Sincerely,
Clinical Staff, Institute of Genetic Medicine

Revised: May 2015

Institute of Genetic Medicine
Johns Hopkins Medicine
Ehlers Danlos syndrome/hypermobility

This questionnaire is to be completed if you or your child was referred due to hypermobility or the possibility of Ehlers Danlos syndrome, hypermobile type. If you/he/she was supposed to be referred a general clinic or another specialty clinic, please contact our office at 410-955-3071 option 1 to be sent another questionnaire.

Patient's Name: _____

Patient's Date of birth: _____/_____/_____ **Phone:** _____

Name of referring doctor (indicate self-referral if no referring doctor)**:**
Doctor Name: _____**Phone:** _____Fax: _____
Address: _____

Reason(s) for appointment: _____

1. Has the patient ever seen a genetics specialist before? Yes / No (circle one)
If **YES**, please list the name and the location of this specialist(s) and attach evaluation notes:

_____

_____

2. Does the patient have a <u>confirmed</u> genetic diagnosis of Ehlers Danlos syndrome? <u>If there is a confirmed genetic diagnosis, please attach genetic evaluation and/or testing reports.</u> Please circle the appropriate choice:

<u>Confirmed</u> diagnosis of (please attach genetic records):
Classic or hypermobile Ehlers Danlos syndrome
Vascular Ehlers Danlos syndrome
Other: _____

3. If the appointment request is to evaluate for a potential diagnosis, please circle the appropriate reason(s):
Joint hypermobility/subluxation/dislocation
Aortic dilation/dissection—rule out syndrome (Marfan, Loeys Dietz or Vascular Ehlers Danlos)
Family history of _____
Other: _____

4. It is helpful for us to better understand the patient and his/her family's <u>MEDICAL PROBLEMS</u>. Please check which symptoms the patient and anyone in their family have.

Findings in Family	Present in patient	Present in relative (age)	Specify relation
Aortic dilation/dissection			
Arterial aneurysm			
Arterial tortuosity syndrome			
Bicuspid aortic valve			
Carotid/coronary dissection			
Arrhythmias, pacemakers			
POTS			

Neurally mediated hypotension			
Congenital heart defects			
Dislocated lenses			
Retinal detachment			
Myopia			
Ehlers Danlos (type)			
Familial thoracic aneurysm			
Loeys Dietz			
Marfan syndrome			
Shprintzen Goldberg syndrome			
Stickler syndrome			
Joint hypermobility			
Joint dislocations			
Joint pains			
Pectus			
Pneumothorax			
Intestinal rupture			
Poor wound healing			
Stretchy skin			
Unusual bleeding			
Flat feet			
Craniosynostosis			
Scoliosis			
Hip dysplasia			
Hernias			
Short stature			
Very tall stature			
Sudden deaths			

5. Please list any additional medical diagnoses that you have (use back of form if necessary)

- _____
- _____
- _____
- _____
- _____

6. Please list any <u>SURGERIES</u> that you have had and age and/or year (use back of form if necessary)

- _____
- _____
- _____
- _____
- _____

7. Has the patient ever had any <u>NON VASCULAR IMAGING</u> (e.g. MRI, CT, X-rays, dexa scans) or <u>LAB TESTS</u> that were abnormal? Yes / No (circle one) *If YES, *Please include a copy of any abnormal imaging reports and/or lab test results with this questionnaire and provide details:*

- _____
- _____

- _____

8. Please list ALL doctors/specialists who this patient sees and the reason(s):

- _____
- _____
- _____
- _____

9. Is there any other important information about the patient we should know prior to scheduling his/her appointment? Please describe:

10. What are the patient's goals for the visit?