BRIGHAM AND WOMEN'S HOSPITAL

HARVARD MEDICAL SCHOOL

BWH Genetics and Genomic Medicine Service: Hypermobility Evaluation

You are receiving this information because our records indicate that you will be undergoing evaluation related to joint hypermobility and/or Ehlers-Danlos syndrome (EDS) at Brigham and Women's Hospital. Please reference the following overview and summary of the clinical services provided by the BWH Genetics and Genomic Medicine Service. We hope that this information is helpful in preparing for your appointment.

- Your clinic session will include an educational video to review the diagnosis of hypermobility EDS, general management recommendations, and genetic testing options.

- Common tests and recommendations for hypermobile/EDS patients typically include the following:
    - An echocardiogram (ultrasound of the heart).
    - Genetic testing for a panel of genes associated with connective tissue disorders.
    - Referral to physical therapy.
    - We may recommend other referrals for the management of medical problems associated with joint hypermobility, however, *we will not provide follow-up or ongoing care coordination*.

- What to expect at your appointment. The purpose of the hypermobility clinic is to provide specific clinical services through an initial genetics evaluation. We will try to make this experience as efficient as possible.
    - You will be seen by a genetic counselor and geneticist.
    - Our clinical scope of practice is medical genetics- we are not musculoskeletal specialists and are not able to directly manage joint pain or instability.
    - Hypermobility clinic does not offer PT services and will not manage or prescribe pain medication.

- What to expect after your appointment. We will send you a copy of the clinic note from your appointment, as well as the results of any testing ordered (i.e. echocardiogram and/or genetic testing).
    - The purpose of our clinic is to provide an initial genetics evaluation for patients who are referred for indication of joint hypermobility. *Please note that we are not a care coordination clinic.*
    - We understand that many patients would like to establish continued care in clinic, but due to limited clinical resources, we are unfortunately unable to offer follow-up appointments.

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Name: _____

Date of Birth: _____

Joint Hypermobility and Connective Tissue Disorders

Many patients who are referred to the Adult Genetics Clinic for a history of joint hypermobility are evaluated in clinic for a connective tissue disorder. Connective tissue disorders are genetic conditions that are characterized by a defect in the connective tissue. Connective tissue provides support to many parts of the body, such as the skin, muscles, GI tract, heart, and ligaments. Because connective tissue is found throughout the body, individuals affected by connective tissue disorders can experience a wide variety of symptoms and medical problems.

Ehlers-Danlos syndrome (EDS) is a condition that falls into the broad category of connective tissue disorders. There are multiple subtypes of EDS that are associated with variable features and severity. The most common type of EDS is called 'hypermobility-EDS', or 'type III' EDS. Hypermobility-type EDS is often characterized by joint hypermobility, joint instability, and joint pain. While some connective tissue disorders are associated with specific and distinctive features, there may be symptoms that overlap between EDS types or other connective tissue disorders.

Medical Intake Screening

Please indicate the status of each symptom with an "X" in one of the boxes to the right.

Please also complete the more detailed medical history questionnaire intake on the following pages of this document.

Symptom	Yes, I have this	I don't have this, but a first degree relative does
Arterial aneurysm or dissection ***NOT including abdominal aortic aneurysm*		
Organ rupture (uterine, intestinal, etc.)		
Lens dislocation (ectopia lentis) or **detached retina**		
Spontaneous lung collapse (pneumothorax)		
Intellectual disability (i.e. mental retardation)		
Multiple (5+) **broken bones** with no clear cause		
Have you ever had **genetic testing**? For what indication?		Indication:
Have you ever been evaluated before by **medical genetics** and/or in a **genetics clinic**? If so, where?		

Medical Record Review

Below is a list of medical records that are helpful for us to have prior to your appointment in clinic, if possible.

Please refrain from sending any types of records other than those listed below:

- Echocardiogram report
- Other cardiac or chest imaging report
- Records related to aneurysm(s), if applicable
- Ophthalmology records
- Genetic testing report
- Clinical genetics note

If you have not had these tests, clinic visits, or imaging studies done before, there is no need to send us any records. The tests, clinic visits, and imaging studies listed here are not things that need to be done for everyone, and do not need to be done for you prior to your appointment unless medically indicated and recommended by your doctor.

Please return this completed questionnaire to us by mail, fax, or email (contact details below). Thank you!

Fax: (617) 264-3018 **Email**: GGMS@partners.org

Mail: BWH Hypermobility Clinic c/o Victoria Perroni
41 Ave Louis Pasteur, #303
Boston, MA 02115

Some of the symptoms described in the following table are associated with EDS, or other types of connective tissue disorders. Please indicate the status of each symptom with an "X" in one of the boxes to the right.

Symptom	Yes, I have this	No, I don't / am not known to have this	Not me, but a family member has this	Unsure
Vision and ENT (ear, nose, and throat)				
Vision problems				
Myopia (near sighted): *Able to see close objects, difficult seeing objects far away.*				
Hyperopia (far sighted): *Able to see objects far away, difficult seeing close objects.*				
Lens dislocation (if yes, please provide age when occurred)				
Retinal detachment (if yes, please provide age when occurred)				
Hearing loss (if yes, please provide age of onset)				
Teeth crowding				
Teeth extraction *(how many have been pulled? Wisdom, baby teeth, etc?)*				
Gum recession				
High or narrow palate				
Other (please describe):				

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Please indicate the status of each symptom with an "X" in one of the boxes to the right.

Symptom	Yes, I have this	No, I don't have this	Not me, but a family member has this	Unsure
Cardiology				
Current or past care with a cardiologist *(if so, what for)*				
Prior heart imaging *(please note if results were normal or not):*				
Echocardiogram (ultrasound of the heart)				
Diagnosed heart problems *(if yes, please describe):*				
Aneurysms *(if yes, please provide details and location):*				
Strong, irregular heartbeat, feeling like your heart skips a beat				
Frequently feeling lightheaded, dizzy				
Palpitations when resting				
Episode(s) of fainting, fully losing consciousness				
Mitral valve prolapse				
Unusual pectus (chest bone) shape				
Pectus excavatum- concave shape (sinks in, "hollowed chest")				
Pectus carinatum- convex shape (protrudes out, "pigeon chest")				
Other (please describe):				

Please indicate the status of each symptom with an "X" in one of the boxes to the right.

Symptom	Yes, I have this	No, I don't have this	Not me, but a family member has this	Unsure
Pulmonary				
Lung collapse *(if yes, please describe and provide age of onset)*				
Other (please describe):				
Symptom	**Yes, I have this**	**No, I don't have this**	**Not me, but a family member has this**	**Unsure**
Gastrointestinal and Genitourinary				
Very frequent problems with: nausea, reflux, diarrhea, constipation, abdominal pain *(if yes, please circle all that apply)*				
Hernias *(now or in the past, please specify where)*				
Trouble urinating				
Diagnosed kidney problems				
Rupture or prolapse or organs – uterus, rectum, etc.				
Other (please describe):				

Please indicate the status of each symptom with an "X" in one of the boxes to the right.

Symptom	Yes, I have this	No, I don't have this	Not me, but a family member has this	Unsure
Musculoskeletal				
Joint hypermobility, looseness *Please list the joints with this problem:*				
Joint pain *Please list the joints with this problem:*				
Joint instability and laxity *Please list the joints with this problem:*				
Joint dislocations *Please list the joints with this problem:*				

Subluxations or partial dislocations *Please list the joints with this problem:*				
Arthritis *(please list the joints with this problem):*				
Muscle or joint weakness				
Recurrent or easy fractures *(please list fractures and indicate if caused by trauma or not):*				
Scoliosis (curve in the spine)				
Kyphosis				
Flat feet				
Club feet				
Joint surgeries *(please list the joints that have had surgery):*				
Other (please describe):				

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Please indicate the status of each symptom with an "X" in one of the boxes to the right.

Symptom	Yes, I have this	No, I don't have this	Not me, but a family member has this	Unsure
Skin				
Easy bleeding from minor cuts and scratches				
Easy bruising				
Fragile, thin, velvety skin				
Abnormal scarring				
Thick, raised scars				
Thin, stretched scars				
Keloid scars				
"Cigarette paper" scars				
Poor wound healing				
Stitches that have popped out or needed to be redone				
Stretch marks (if yes, please indicate where):				
Varicose veins (bulging veins)				
Other (please describe):				

Please indicate the status of each symptom with an "X" in one of the boxes to the right.

Symptom	Yes, I have this	No, I don't have this	Not me, but a family member has this	Unsure
Endocrinology				
Growth problems				
Thyroid problems				
Hypothyroidism (low thyroid)				
Hyperthyroidism (high thyroid)				
Bone density				
DEXA study (bone density scan) – please indicate results:				
Osteopenia (please indicate where, if known):				
Osteoporosis (please indicate where, if known):				
Other (please describe):				
Symptom	**Yes, I have this**	**No, I don't have this**	**Not me, but a family member has this**	**Unsure**
Neurology				
Frequent headaches or migraines				
Brain imaging studies (if done, please note results):				
Other (please describe):				