Patient name:_____

DOB: _____ DOV:_____

MRN: _____

Provider: _____

CONNECTIVE TISSUE DISORDER PATIENT QUESTIONNAIRE

Patient's Name: _____ Date of Birth: _____

If patient is under 18, Parents/Guardians' Names:_____

Contact Phone Number(s): _____

This form is confidential and will become part of the patient's medical record.

Referring physician: _____
Address: _____
_____
_____

Primary Care Physician: _____
Address: _____
_____
_____

Other Physician: _____
Address: _____
_____
_____

Other Physician: _____
Address: _____
_____
_____

By signing below, you give us permission to send a copy of the clinic note from the clinic visit to the healthcare providers you listed above.

_____ _____

Patient's Signature/Signature of Parent or Guardian Date

Your Reason for Referral (Please circle all that apply)

Marfan syndrome Ehlers-Danlos Syndrome Tall Stature Scoliosis Flexible joints
Joint Dislocations Hernias Dilated blood vessels Lens dislocation Stretch Marks

Your Questions for the Doctor

Please list any specific questions or concerns you would like to discuss during the clinic visit.

_____
_____
_____
_____
_____
_____
_____

Patient Name: _____

This form will need to be completed and returned to us prior to your visit or your visit may be cancelled.

Cardiac: Have you ever had an echocardiogram (heart ultrasound)? **Y / N**

Date performed _____ Hospital where the echocardiogram was done _____

You must have an ECHO prior to your appointment!!! If this has not been completed, please ask your primary physician to have one completed at least 4 weeks before your appointment AND to send us the report! If this is not done, your appointment may be cancelled!

Below are questions and also information about connective tissue disorders like Ehlers-Danlos Syndrome (EDS). Some people will complete this packet and attached information sheet and determine that they now have all the information they need about EDS and no longer need to see the Geneticist.

It is important, however, that you DO see the Geneticist if you or your family have any personal or family history of aneurysms:

Have you or your family members had an **aneurysm or dissection of the aorta in the chest or by the heart?**
Yourself? Y / N Family members? Y / N Which family members? _____

Have you or your family members had an **aneurysm or dissection of the abdominal aorta?**
Yourself? Y / N Family members? Y / N Which family members? _____

Have you or your family members had a **brain aneurysm?**
Yourself? Y / N Family members? Y / N Which family members? _____

If you answered "yes" to any of these questions, please call our office 545-8000 and ask to speak to the Genetics nurse so that we can see you in the next few weeks, as you may have a more serious connective tissue disorder.

If you answered "no" to all questions in the box above, please proceed with the remainder of the questionnaire:

Cardiac: Were you or any family members born with a heart defect? Y / N If yes, please describe:

_____

_____

Do you or any family members have a problem with a heart valve or needed surgery on a heart valve? Y / N
If yes, please describe:

_____

_____

Joints: Have you had any of the follow problems with your joints:
Dislocations? Y / N If "yes", which joints?

_____

_____

Patient Name: _____

Subluxations (the joint feels like it is going out of place, but is not dislocated)? Y / N If "yes", which joints?

_____

_____

Have you ever had surgery on any joints? Y / N If "yes", please list joint surgeries:

_____

_____

Are you able to do "joint tricks", such as putting your feet behind your head? Y / N If "yes", please list:

_____

_____

Do you have hand pain after writing? Y / N

Have you been diagnosed with fibromyalgia? Y / N

If you have joint pain, which joints bother you?

_____

_____

_____

_____

Does your joint pain keep you from daily activities? Y / N If yes, please describe:

_____

_____

_____

_____

_____

_____

_____

_____

_____

What, if any, medications do you routinely use for joint pain?

_____

Do you receive any physical therapy services? Y / N If yes, please describe when and which joint was treated:

_____

_____

_____

Have you or your family members ever been diagnosed with scoliosis (curving of the spine)? Y / N
If yes, please list all those affected and describe any treatment needed:

_____

_____

Patient Name: _____

Do you or your family members have an unusual shape to your chest, either as a pectus (breast bone is sunken in or pushes out too far) or significant chest asymmetry (one side sticks out further than the other)? Y / N
If yes, please list all those affected and describe:

_____
_____

Skin: Do you have any problems with your skin? Y / N If yes, please circle any issues you have and describe below:

Easy Bruising / Skin tears easily / Poor or slow wound healing / Stitches that tear / Scars are wide and thin / Stretchmarks (prior to having children if female) / Hernias

_____
_____
_____

Vision: Do you have any problems with your vision? Y / N

Dislocated lens (ectopia lentis) Y / N Age at dx _____
Nearsighted (myopia) Y / N Age at dx _____
If nearsighted, please give correction in diopters if known: _____
Please list any other vision problems_____
Date of last eye exam _____ Where was it performed? _____

Dental : Do you have any problems with your teeth? Y / N Circle any issues you have.

Multiple cavities / Enamel problems / Needed orthodontics (braces) for dental "crowding" /
Bleeding of the gums/ TMJ (clicking of the jaw) / Teeth that break off without cavities in the tooth
Other _____

Gastrointestinal: Do you have any problems with your stomach and/or intestines? Y / N Circle any issues:

Irritable Bowel Syndrome / Reflux (heartburn) / Hemorrhoids / Rectal fissure / Rectal prolapse

Genitals/Urinary Tract: Circle any issues you have:

Uterine prolapse / Bladder prolapse / Recurrent urinary tract infections

If you are female and have gone through puberty, do you have any problems with your periods? Y / N
Circle any issues you have: Heavy bleeding / Periods that last more than 7 days

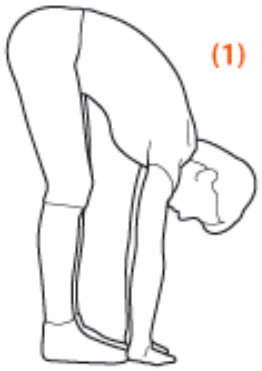
If you have had a pregnancy, were there any complications with delivery or after delivery? Y / N
Circle any issues you have: Pre-term delivery / Excessive bleeding / Uterine rupture / Other: _____

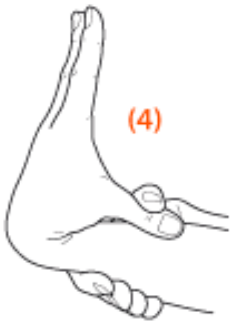
Has anyone in your family been diagnosed with a connective tissue disorder such as Marfan syndrome or Ehlers-Danlos syndrome? Y / N If yes, who, with which disorder, and where are they treated or followed?

_____
_____

<u>Ehlers-Danlos syndrome (EDS), hypermobility type</u> is diagnosed in people who have some of the joint problems and other symptoms listed above AND also have joint hypermobility.
<u>Please complete the Beighton hypermobility tests on the following page. A score of 5 points or higher out of the total of 9 points is positive for hypermobility.</u>

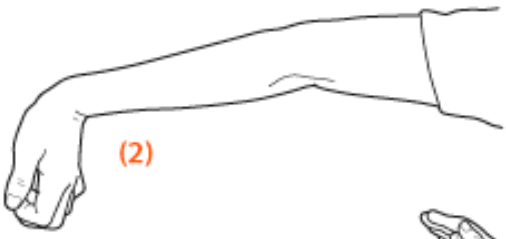
Patient Name: _____

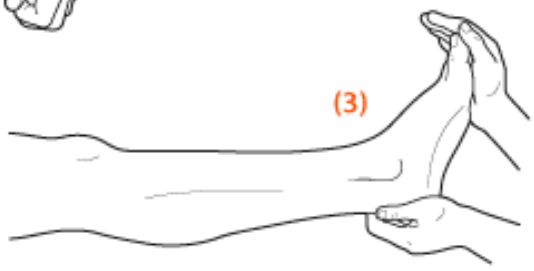


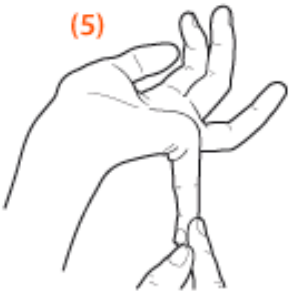


The Beighton score

Beighton's modification of the Carter and Wilkinson scoring system. Give youself 1 point for each of the manoeuvres you can do, up to a maximum of 9 points.







	SCORE Left	Right
1. Can you put your hands flat on the floor with your knees straight?	1	
2. Can you bend your elbow backwards?	1	1
3. Can you bend your knee backwards?	1	1
4. Can you bend your thumb back on to the front of your forearm?	1	1
5. Can you bend your little finger up at 90° (right angles) to the back of your hand?	1	1
		9

Your score: ___ / 9

Score of others in your family:

_____

_____

_____

_____

_____

_____

Patient Name: _____

Thank you for completing the questionnaire so far!

Attached are two pages of information about how to manage **Ehlers-Danlos Syndrome (EDS), hypermobility type**. There are several types of EDS, but hypermobility type is by far the most common. If you have joint issues as described above (dislocations, subluxations, joint pain), some additional symptoms as described above, and also a positive Beighton score (5 or higher), you most likely have EDS, hypermobility type.

If you feel your questions about **Ehlers-Danlos Syndrome (EDS), hypermobility type** have been answered by this information sheet, you do not have to see a Geneticist. If you still have questions and would like to have a Genetics consultation, we would be happy to see you and we ask that you finish the rest of this form. As stated on the first sheet, if you or family members have a history of aneurysms, we need to see you sooner, so please call us directly at 545-8000 and ask for the Genetics nurse.

Medications ☐ No current medications

Medication Name	**Reason** (example: allergies or seizures)	**Dose** (examples: 25 mg tablet or 100 mg/5 ml suspension)	**Frequency Taken** (examples: 1 tablet twice daily or 2 teaspoons 3 times a day)

Do you have any **allergies to medications**: Y / N
If yes, please list medication name(s) and describe allergic reaction(s): _____
_____

If you are over age 18, please answer the following questions IF APPLICABLE:

What is your occupation? _____

What is your highest level of education? _____

Do you have any learning disabilities, require any special education, or have resource room assistance?
Y / N If yes, please explain:_____

For patients under age 18, please complete next two sections: Pregnancy and Birth History AND Developmental History.

Pregnancy and Birth History

Mother's age at delivery: _____ What number pregnancy was this child for the mother? _____

Did the mother have any complications during ***this child's*** pregnancy? For example, diabetes, high blood pressure or infections? If yes, please list the complication and how it was treated below.

_____
_____
_____

Medications used during pregnancy: _____

Patient Name: _____

How much alcohol did the mother consume during pregnancy? _____
How many packs per day of cigarettes did the mother smoke during pregnancy? _____
Please list any street drugs (marijuana, cocaine, etc) used during pregnancy: _____

The child was born: ☐ at full-term ☐ prematurely (weeks premature: _____)
If premature, please list the reason: _____

The child was born: ☐ Vaginally ☐ by C-section; if so, why: _____

Birth weight: _____ Birth Length: _____ Birth head circumference:_____

Did your child have any problems after delivery or require admission to the Neonatal Intensive Care Unit (NICU)?
Yes / No If yes, please explain: _____

Developmental History

Are you concerned about your child's development? **Y / N** If yes, since what age:_____

If yes, circle the developmental concerns that you or your child's doctor(s) have about your child:

Hyperactivity	Delay in motor development	Autism/Asperger/PDD-NOS
Short attention span	Delay in language development	Learning difficulties

List any other developmental or behavioral problems that your child has or may have: _____
_____
_____

What do you think is the developmental age of your child? _____

Please check what skills your child currently has and *list the age* when achieved if known:

Developmental Skills	✓	**Age**		✓	**Age**
First word (not dada/ mama)			Print name		
Talk in Sentences			Write in cursive		
Dress and Undress self			Pull to stand		
Builds a tower w/ blocks			Cruise		
Button or Zip clothing			Go up steps w/o help		
Walk independently			Riding a bicycle		

If your child is school-aged: Current Grade _____

Has your child ever had an **evaluation** by therapists, an IEP, or formal IQ testing? **Y / N**
If yes, at what age(s)? _____ What were the results?_____
_____

Is your child receiving special education or resource room assistance? **Y / N**
If yes, please explain the type and amount of assistance your child receives: _____
_____
_____

Patient Name: _____

EHLERS-DANLOS COUNSELING

Hypermobility and joint pain:

Let's review the signs and symptoms of Ehlers-Danlos syndrome (EDS), including surveillance, management, treatment and prognosis. The hypermobility form of EDS is characterized by generalized joint laxity, recurrent joint dislocations and sprains, spontaneous joint subluxation and joint pain. There is a natural history of the joint problems associated with EDS. Children often have significant growing pains and may have delays in their gross and fine motor skills, including handwriting. As children move into elementary school, they may start to have pain during or after PE or sports activities. Joint problems may become more evident with increased frequency and rigor of the physical activity. Some children with EDS require modified PE class. Joint pain may not occur with physical activity, but may be present hours after the activity. Flat feet are also common and can result in lower extremity pain of the foot, ankle, knee or hip. Adults may not appear as hypermobile as they were in childhood, but may instead experience chronic joint pain. Males generally have fewer problems with joint instability than females, possibly due to greater muscle mass to stabilize the joint.

Treatment of the joint problems related to EDS focuses on avoiding activities that cause dislocations and sprains and controlling chronic joint pain. Nonsteroidal anti-inflammatory agents are the first medication to try for joint pain. Over the counter medications such as Ibuprofen or other longer-acting NSAIDs such as Naproxen should be taken on a regular basis for several weeks at a large enough dose to decrease inflammation. Then the dose or frequency can be titrated as needed. These medications should be taken with food to avoid stomach irritation. Some patients find that prescription medications are necessary to control their joint pain. Some form of exercise (such as swimming, walking, or biking) is important to improve muscle tone to stabilize joint hypermobility. Sometimes a physical therapist can aid in planning or adapting strengthening exercise. It is important not to overdo the exercise or therapy to avoid exacerbating joint pain. For flat feet, we suggest trying shoes with a good arch support, such as New Balance shoes. Shoes with ankle support can also aid in prevention of ankle sprains and joint pain.

Aortic root dilatation:

A very small number of individuals with hypermobility type EDS will have aortic root dilation. Our data suggest that aortic root dilatation is more common in the 6-12 year age range, with resolution of the dilatation by high school. Cardiac valvular problems may also be seen, such as mitral valve prolapse. Because of the potential for cardiac involvement, a screening echocardiogram should be performed at diagnosis. If this study is normal, a follow up study is recommended in 3-5 years.

POTS (postural orthostatic hypotension and tachycardia syndrome):

EDS patients with POTS have symptoms of increased heart rate, sweating, dizziness, lightheadedness and near fainting when moving to a standing position. POTS symptoms can be helped by increasing fluid and salt intake and by rising more slowly and sitting at the side of the bed when moving from a lying to standing position.

Other symptoms:

Most people with EDS have skin which is very soft and sometimes even "stretchy". Easy bruising and poor wound healing are also common in both children and adults with EDS. To decrease bruising and improve healing, some patients have responded well to 1-4 grams of vitamin C supplementation per day.

Dental problems such as multiple caries, enamel problems, and gingival bleeding may also occur. Fibromyalgia, anxiety, depression, migraines, irritable bowel syndrome, heavy periods, hernias, varicose veins, and uterine, bladder, or rectal prolapse also seem to be fairly common in patients with EDS.

The above conditions seen in EDS patients are treated in the same way they are treated in non-EDS patients. It is also important to remember that the features of EDS may vary from one individual to another within a family and each person may not exhibit all or the features of the condition.

Inheritance pattern:

The hypermobility form of EDS is inherited in an autosomal dominant pattern. This means each child born to an individual with EDS has a 50% chance of inheriting the condition. **At the present time, confirmatory genetic testing of the hypermobility form of EDS is not available.** This may change in the future as genes associated with this form of EDS are identified.

Additional information about hypermobility EDS is available from the Ehlers-Danlos National Foundation, which can be found at **ednf.org.**