Medical Genetics Connective Tissue Questionnaire for Referring Providers

Thank you for referring your patient for evaluation in Medical Genetics. After reviewing the provided documentation, it appears the primary reason for this referral is to evaluate for Ehlers-Danlos Syndrome (EDS). There is such a high volume of patient referrals for this indication nationally and regionally and URMC Medical Genetics is one of the only clinics in upstate NY region accepting referrals for EDS. We are unfortunately experiencing a high volume of referrals for this indication, and the anticipated wait time for an appointment for EDS is around 3 years.

While we understand this wait time is not ideal, and regret not being able to promptly evaluate your patient, we would like to provide some general management guidelines and resources to help manage symptoms in the interim.

- As you know, there is a spectrum of severity across the different connective tissue disorders like EDS. In our experience, the most common type of Ehlers-Danlos is hypermobile Ehlers-Danlos (hEDS). HEDS is considered the least severe type of EDS, but there can be complications, mostly musculoskeletal. There is currently no clinical genetic test to diagnose this most common type of EDS. Attached are the diagnostic criteria to make a clinical diagnosis of Hypermobile (type III) Ehlers Danlos Syndrome. https://www.ehlers-danlos.com/wp-content/uploads/2019/09/hEDS-Dx-Criteria-checklist-1-Fillable-form.pdf
- Symptom Management: There are no targeted treatments for patients with a diagnosis of hEDS. Treatment is focused on supportive measures for the patient's symptoms. General recommendations to manage a diagnosis of hypermobile EDS include:
    - Echocardiogram screening every 3-5 years
    - Maintenance physical therapy to strengthen and stabilize joints (especially aqua therapy)
    - Low impact physical activity
    - Increased salt intake if the patient experiences POTS/dysautonomia
    - GI evaluation if patient has severe gastrointestinal symptoms
    - Pain Management Clinic for Chronic Pain symptoms
- However, there are other connective tissues disorders where genetic testing can be coordinated, and results could alter management for patients. Please alert our office if your patient has any of the following symptoms, as these may increase the urgency of their referral and shorten their time to schedule:
    - History of aneurysm and/or dissection
    - History of pneumothorax
    - History of retinal detachment or ectopia lentis (lens dislocation)
    - History of organ prolapse or rupture (uterine or rectal)
    - If you believe this evaluation is medically urgent for any other reason, please feel free to contact our office.
- **We are requiring the following prior to scheduling an appointment:**
    - **An echocardiogram, if not yet performed**
    - **Clinical information questionnaire**
    - **Family history questionnaire**

We hope you find this information helpful. Please contact our office at 585-275-3461 to discuss any further questions or concerns.

Thank you,

Medical Genetics Team
URMC Division of Clinical Genetics
601 Elmwood Ave Box 641
Rochester, NY 14642
P: 585-275-3461
F: 585-756-8054

Additional Resources:

A Practical Guide- What to do When You Suspect EDS:
https://www.mountainstatesgenetics.org/wp-content/uploads/sites/257/2022/02/2022-01-31-EDS-Algorithm-Booklet-Revised-PDF-for-Upload.pdf

Ehlers Danlos Society- Page for Healthcare professionals:
https://www.ehlers-danlos.com/healthcare-professionals/

URMC Medical Genetics Connective Tissue Questionnaire

Patient Name: _____

DOB: _____ / _____ / _____

Referring Provider's Name: _____

Referring Provider's phone number: _____ - _____ - _____

Referring Provider's fax number: _____ - _____ - _____

Please indicate "yes" or "no" depending on whether (the patient) has or has had any of the following features:

Presence or history of aortic/arterial dilation, aneurysm, or dissection?	○ Yes ○ No ○ Unknown
Has the patient had an echocardiogram (echo) in the past?	○ Yes ○ No
If the patient has not had an echocardiogram, please order one and have the results faxed to URMC Genetics at (585) 756-8054 as this is a requirement for the referral to be triaged appropriately and for your patient to be scheduled.	
Date of most recent echocardiogram:	_____ / _____ / _____
Were the echo results abnormal?	○ Yes ○ No ○ Unknown
If the Results were abnormal, please describe:	
Please fax the patient's echo results to URMC Genetics at (585) 756-8054	
History of organ prolapse or rupture?	○ Yes ○ No ○ Unknown
History of pneumothorax?	○ Yes ○ No ○ Unknown

History of retinal detachment or ectopia lentis (lens dislocation)?	○ Yes ○ No ○ Unknown
Signification/abnormal atrophic scarring?	○ Yes ○ No ○ Unknown
Reasons to suspect a genetic syndrome that may have been omitted in the original referral documentation. Explain:	
Notable family history in a close relative of symptoms listed above (1st or 2nd degree relative, under the age of 50 years) OR a confirmed diagnosis of a genetic condition (other than hEDS):	
If there is a known family history of a genetic condition, please fax a copy of the affected relative's genetic test report to URMC Genetics at (585) 756-8054	
If you believe this evaluation is medically urgent for any other reason, please specify here:	