EMORY CLINIC

Ehlers-Danlos Syndrome (EDS) Referrals for Pediatric Patients

Date:_____

Dear Referring Provider:

Thank you for entrusting us with the care of your patient:_____
DOB:_____ for possible Ehlers-Danlos Syndrome (EDS).

Please note: we cannot complete an accurate assessment in patients under the age of 12.

For patients aged 12-18, **one of the following must be confirmed:**

- ☐ Aortic dilatation confirmed by echocardiogram - please attach echo result
- ☐ Aneurysm or arterial dissection - please attach imaging results or operative note
- ☐ Prolapse in nulliparous female. - please attach gynecology note
- ☐ Family member with a likely pathogenic or pathogenic variant on a genetic test. - please attach relative's results
- ☐ Personal or family history of spontaneous organ rupture. -please provide patient or relatives' pertinent records relating to rupture.

Please check the reason for referral above and fax this checklist, along with supporting documentation and patient demographics to 404-778-8562 Attn: EDS Referrals. **If one of the above reasons is not selected and/or if the supporting documents are not sent, we cannot schedule your patient**.

Once we receive your reply with the supporting documentation we will contact your patient to schedule an appointment.

If you would like to speak with the geneticist on call about your patient, please call 404-686-5500 PIC#50263

Sincerely,

The Physicians of Emory Clinic, Department of Human Genetics

1365 Clifton RD., Building B, Suite 2200, Atlanta, Georgia 30322

Tel 404.778.8570 Fax 404.778.8562

*Please see page 2 of this fax for information regarding hypermobility syndrome that we hope is helpful to both the patient and the provider. Please feel free to share this information with your patient.

HYPERMOBILITY SYNDROME: HELPFUL INFORMATION

EDS is not a single condition but a group of disorders that primarily affect the connective tissue. The majority of people with suspected EDS have one of many Hypermobility Spectrum Disorders (HSD), including hypermobile EDS (hEDS). The criteria for the diagnosis were revised in 2017. The best estimate is that about 2% of the population have HSD/hEDS.

There is no known causative gene for HSD/hEDS. Inheritance in some families follows an autosomal dominant pattern while in others it resembles polygenic inheritance. There is a spectrum of disease that starts at benign joint hypermobility and ends at hEDS. The syndrome includes POTS, fibromyalgia-like pain, diffuse joint pain that does not necessarily correspond to the loose joints, chronic fatigue, irritable bowel syndrome, migraines, TMJ dysfunction, bladder irritability, early degenerative joint disease in some. Life expectancy is normal.

Management often requires the input of many specialists such as cardiology, physiatry and pain medicine. It is very important to establish a single PCP that coordinates care. Beta blockers, midodrine and fludrocortisone may be effective in the management of POTS syndrome. Often patients have either major depression or dysthymia and treatment with antidepressants may be indicated. Amitriptyline and duloxetine may help with the fibromyalgia-like pain as well. When pain seems confined to a few joints, physical therapy and the use of devices to stabilize the joint may be of benefit. Chronic fatigue responds best to exercise regimens, such as swimming or leg-intensive workout. There is anecdotal evidence that gluten free diet, acupuncture and high doses of vitamin C may be of benefit for some patients.

This diagnosis is not a contraindication for orthopedic or other surgical procedures.