

## REFERRAL FORM CONNECTIVE TISSUE DISORDER

Thank you for referring your patient.

Isolated hypermobility is a common condition for which there is currently no role for genetic evaluation or genetic testing to confirm the diagnosis. We therefore are no longer accepting referrals for **isolated hypermobility**. You may wish to refer your patient to another medical specialist for management of symptoms, if required.

# Additional information about hypermobility/Ehlers Danlos Type III can be found at: https://www.ncbi.nlm.nih.gov/books/NBK1279/

However, for patients who display hypermobility and/or other connective tissue signs and symptoms, please complete this form. The information provided will help determine whether an evaluation in genetics is indicated. Please make special note of the items marked with an « \* », which are required accompanying documentation. Incomplete referral forms will not be triaged.

All adolescent and adult patients should have had a screening echocardiogram within the last 3 years\*.

Referring physician information Name Specialty Mailing address Telephone Fax Email Reason for referral (please check all applicable factors) ☐ Aortic root enlargement or aneurysm / dissection ☐ Tissue fragility; e.g. hernias, recurrent rectal or uterine prolapse ☐ Intestinal/uterine fragility or rupture ☐ Generalized arterial tortuosity or aneurysm ☐ Ectopia lentis (dislocated lens) ☐ Pneumothorax, spontaneous ☐ Clubfoot deformity ☐ Spontaneous organ rupture (specify) \_ ☐ Severe scoliosis ☐ Anxiety, depression or other psychiatric diagnosis (specify) ☐ Pectus deformity ☐ Bifid uvula or cleft palate ☐ A family member with a genetic mutation which causes a ☐ Joint dislocation (requiring visit to emergency room connective tissue disorder (include copy of result) for reduction); which joints, amount of occurrences, ☐ Family history of sudden death at age of 40 years old or less activity at time of dislocation ☐ Beighton ccore\* (see chart on the next page) □ Previous joint surgery (specify) ☐ Skin fragility; abnormal stretch marks; hyperextensibility/abnormally stretchy skin ☐ Previous amputation (specify) ☐ Thin, translucent skin ☐ Easy bruising ☐ Wheelchair-bound ☐ Atrophic scars; specify site(s) ☐ Other pertinent features in your patient

#### Please complete the **Beighton score** as indicated below (required\*).

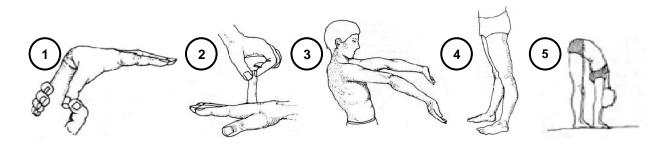
Please refer to the web link below for the technique of the Beighton Testing.

### https://www.youtube.com/watch?v=uYRYsCW0gl8

	CLINICAL MANEUVER	UNABLE TO PERFORM (0 POINT)	ABLE TO PERFORM (1 POINT)
	Appposition of thumb to forearm		
(1)	Right	0	1
	Left	0	1
	Extension of 5 <sup>th</sup> finger beyond 90 degree	ees	
(2)	Right	0	1
	Left	0	1
	Extension of elbow beyond 10 degrees		
(3)	Right	0	1
$\mathbf{C}$	Left	0	1
	Extension of knee beyond 10 degrees		
(4)	Right	0	1
$\bigcirc$	Left	0	1
(5)	Forward flexion of trunk,		
しり	legs straight, palms touching floor	0	1
•	<b>Total Beighton score</b> (sum of points for each maneuver - 0 to 9 points)		

#### « Beighton score positive;

for child: 6 points or more; from puberty to 50 years old: 5 points or more; for 50 years old or more: 4 points or more »



For all patients, please provide the following:

- Copy of screening echocardiogram\*
- Consultation reports from any pertinent specialists, recent laboratory tests and imaging (e.g. MRI)

We will provide assessment and management recommendations for your patient. In some cases, management and referrals may be initiated by our service.

However, after initial consultation, the patient will be returned to you for ongoing care.

We will proceed with triaging the referral upon receipt of a complete referral form. Please fax the referral back to the Genetics Clinic at 514-412-4296 or email to genetics @muhc.mcgill.ca

Signature	Date (YYYY/MM/DD)