CONNECTIVE TISSUE LABORATORY
Center for Medical Genetics – Ghent University Hospital - MRB – De Pintelaan 185 – B-9000 Ghent, Belgium
Department Chair: Prof. A. De Paepe – Supervisor Connective Tissue Lab: Prof. P. Coucke
Receipt of samples: Tel: 0032-(0)9-332 24 77 – Fax: 0032-(0)9-332 65 49
Website: http://medgen.ugent.be – e-mail: connective_tissue@medgen.ugent.be

CLINICAL INFORMATION SHEET

Ehlers-Danlos syndromes & Cutis laxa

Patient information

Name:
First Name(s):
Sex: ☐ M ☐ F
Date of Birth (dd-mm-yyyy): / /
Address:

Referring Physician:
Referring Center:

SAMPLE:
☐ EDTA blood ☐ DNA ☐ Skin biopsy ☐ Chorionic villi
☐ Heparin blood ☐ RNA ☐ Aortic biopsy ☐ Amniocytes
☐ Buccal swab ☐ Fibroblasts ☐ Paraffin embedded material
☐ Other:

Date (dd/mm/yyyy): / /
Sample arrived:

Suspected diagnosis

☐ Classic type of EDS (type I/II)
☐ Hypermobile type of EDS (type III)

☐ Vascular type of EDS (type IV)
☐ Kyphoscoliosis type of EDS (type VIA)
☐ Musculocontractural type of EDS (type VIB)
☐ Arthrochalasis type of EDS (type VIIA/B)
☐ Dermatosparaxis type of EDS (type VIIC)

☐ Autosomal dominant Cutis Laxa
☐ Autosomal recessive Cutis Laxa type I
☐ Urban Rifkin Davies syndrome
☐ Autosomal recessive Cutis Laxa type II
☐ Wrinkly Skin syndrome
☐ Occipital horn syndrome
☐ De Barsy syndrome
☐ RIN2 syndrome

☐ Other:

The differential diagnosis in patients referred for additional genetic testing with a clinical presentation of Ehlers-Danlos syndrome or cutis laxa syndrome is extensive. Please indicate which diagnosis you suspect in your patient and make sure to fill out the check-list as complete as possible so that we can set up the appropriate genetic testing.

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CLINICAL SUMMARY

PEDIGREE

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SYSTEMIC CLINICAL CHECK-LIST

	Suspected clinical diagnosis:		
	Height: ; Weight: ; Head circumference: ; Armspan: ; Lower segment:		
	SKIN (PLEASE PROVIDE CLINICAL PICTURES if possible)	Yes	No
Hyperextensible skin		☐	☐
Smooth velvety skin		☐	☐
Thin, transparent skin		☐	☐
Widened, atrophic scars		☐	☐
Acrogeria		☐	☐
Easy bruising		☐	☐
Cutis laxa: Age of onset Localisation (please specify): Progressive		☐	☐
Hernia: Recurrent: Localisation (please specify):		☐	☐
Other (please specify)		☐	☐
	MUSCULOSKELETAL		
Beighton score for joint hypermobility *			/9
Congenital hip dislocation Unilateral Bilateral		 ☐ ☐	 ☐ ☐
Recurrent dislocation/subluxations: Frequence: Localisation (please specify):		☐	☐
Club feet		☐	☐
Delayed closure of the fontanels		☐	☐
Muscle hypotonia		☐	☐
Tendon rupture: Muscle rupture:		☐ ☐	☐ ☐
Fractures: Number:		☐	☐

Localisation Osteoporosis on BMD:	☐	☐
Scoliosis Age at onset: Degree: Progressive:	Yes ☐	No ☐
CARDIOVASCULAR		
Arterial rupture/dissection Localisation (please specify):	☐	☐
Cardiac-valvular abnormality: Localisation (please specify): Severity:	☐	☐
Arterial Tortuosity Localisation (please specify):	☐	☐
Other vascular problems (please specify):		
OTHER SYSTEMIC FEATURES (please specify):		
• Ocular: • Central Nervous: • Gastro-intestinal: • Genito-Urinary: • Pulmonary:		

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*Beighton's Criteria for Joint Hypermobility			
Joint/Finding	**Negative**	**Unilateral**	**Bilateral**
Passive dorsiflexion of the 5th finger >90°	0	1	2
Passive flexion of thumbs to the forearm	0	1	2
Hyperextension of the elbows beyond 10°	0	1	2
Hyperextension of the knees beyond 10°	0	1	2
Forward flexion of the trunk with knees fully extended and palms resting on the floor	0	Present=1	

A total score of at least 5 defines hypermobility.