

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

Skeletal dysplasias

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

- Achondrogenesis type 2 / Hypochondrogenesis
 - Platypondylic lethal skeletal dysplasia, type Torrance (PLSD-T)
 - Spondyloperipheral dysplasia (SPD)
 - Spondyloepiphyseal dysplasia congenita (SEDC)
 - Spondylometaphyseal dysplasia, type Strüdwick (SEMD)
 - Kniest dysplasia
 - Stickler syndrome type 1
 - Czech dysplasia metatarsal type
 - Avascular necrosis of the femoral head
- Other:

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 SUMMARY

PEDIGREE

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

Phenotypic features

Measurements	
<input type="checkbox"/> NE (not examined)	Age:
Height:	cm
Weight:	kg
Head circumference:	cm
Other:	

Ophthalmological								
<input type="checkbox"/> NE	Yes	No	NE		Yes	No	NE	
Myopia:	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Type 1(*) va	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
If yes, degree:				Type 2 (***) va	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Cataract:	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Optically empty vitreous	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Glaucoma:	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Vitreous veils and bands	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Retinal tear:	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Retinal detachment:	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Chorioretinal atrophy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Perivascular pigmentation	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Sheathing of peripheral	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Posterior perivascular	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					

(*) type 1 = an apparently vestigial vitreous gel occupies the immediate retrolental space and is bordered by a distinct folded membrane.
 Snead MP, Yates JRW. *Clinical and molecular genetics of Stickler syndrome. J Med Genet* 1999;36:353-359.

(**) type 2 = with sparse and irregularly thickened bundles of fibres throughout the vitreous cavity. Snead MP, Yates JRW. *Clinical and molecular genetics of Stickler syndrome. J Med Genet* 1999;36:353-359.

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

Orofacial			
<input type="checkbox"/> NE	Yes	No	NE
Cleft palate	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Submucous cleft	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Bifid uvula	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Flat face/midfacial hypoplasia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Micrognathia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Low nasal bridge	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Ears (please provide copy of audiogram)			
<input type="checkbox"/> NE	Yes	No	NE
Conductive hearing loss	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Sensorineural hearing loss	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

Musculoskeletal					
<input type="checkbox"/> NE	Yes	No	NE	Beighton score (***)	Age
Joint hypermobility	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		
Degenerative arthropathy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		
Joint replacement surgery	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>		
Other musculoskeletal problems					

(***) Articular hypermobility is assessed by using the 9-point Beighton score, which assigns one point for each side of the body on which the patient can (1) passively dorsiflex the 5th finger >90 degrees with the forearm flat on the table, (2) passively appose the thumb to the flexor aspect of the forearm, (3) hyperextend the elbow beyond 10 degrees, and (4) hyperextend the knee beyond 10 degrees and one point for forward flexion of the trunk with the legs straight so the palms rest flat on the floor. Beighton P. *McKusick's heritable disorders of connective tissue*. 5th ed. St Louis: Mosby, 1993.

Skeletal X-rays
<input type="checkbox"/> NE
Please provide radiographs in case of short stature or radiological abnormalities. Minimal requirements: skull AP and lateral left hand AP pelvis AP knees AP thoracolumbar spine AP and lateral

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

Family	Yes	No	NE
Family history positive	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
If Yes, please provide copy of family pedigree.			