

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

Osteogenesis Imperfecta

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

- Osteogenesis Imperfecta type I (mild)
- Osteogenesis Imperfecta type II (lethal)
- Osteogenesis Imperfecta type III (severe)
- Osteogenesis Imperfecta type IV (moderate)
- Recessive Osteogenesis Imperfecta
- Bruck syndrome
- Other (e.g. battered child):

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

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

SYSTEMIC CLINICAL CHECK-LIST

NA = not available
 NE = not examined

Facial							
<input type="checkbox"/> NA	Yes	No	NA	Yes	No	NA	
Blue sclerae	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Triangular face	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Other:							

Osteo-articular						
<input type="checkbox"/> NE						
Height:	cm	Weight:	kg	Head circumference:		cm
Arm span:	cm	Lower segment:		cm		
				Yes	No	NA
Short stature/dwarfism				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
- Short and deformed extremities				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Pectus deformity				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Osteopenia/osteoporosis				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Joint hypermobility				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Contractures				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Kyphoscoliosis				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
- Degree :						
- Age of onset :						
- Progressive :				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Fractures						
-						
-						
-						
-						
-						
Multiple, recurring				<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>

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

	Yes	No	NA
Dentinogenesis Imperfecta : (discolored/translucent teeth)	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Hearing loss	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
- unilateral	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
- bilateral	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
- type of hearing loss			
.....			
.....			
Other :			
- Skin manifestations	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Describe			
.....			
.....			
- Hernia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
- Cardiac/Vascular manifestations	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Describe			
.....			
.....			
X-rays			
-			
-			
-			
Audiometry			
-			
-			
-			



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

