

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

Pseudoxanthoma Elasticum (PXE) syndrome

PXE- like syndrome

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

PXE
 PXE - like

Other:

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CLINICAL SUMMARY

PEDIGREE

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CLINICAL CHECK-LIST

NE = not examined

Antropometric data					
Height:	cm	Weight:	kg	Head circumference:	cm

Skin								
<input type="checkbox"/> NE		Yes	No	NE		Yes	No	NE
Cutis laxa (loose, sagging)	<input type="checkbox"/>		<input type="checkbox"/>	<input type="checkbox"/>	Peau d'orange	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Generalized					Location:			
Localized					Plaques of papules	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Yellowish papules	<input type="checkbox"/>		<input type="checkbox"/>	<input type="checkbox"/>	Location:			
Location:								
Skin biopsy:								
Other:								

Ophthalmological								
<input type="checkbox"/> NE		Yes	No	NE		Yes	No	NE
		Left				Right		
Angioid streaks	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Peau d'orange	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Comets/ comet tails	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Retinal hemorrhage	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Macular degeneration	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Retinal dystrophy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Current visual acuity								
Other:								

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Cardiovascular / Haematological / Gastro-intestinal								
<input type="checkbox"/> NE	Yes	No	NE		Yes	No	NE	
Hypertension	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Angor pectoris	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Intermittent claudication	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Myocardial infarction	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Early onset varicose veins	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Mitral valve pathology	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>					
Prolapse								
Regurgitation								
Coagulation disorder	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Hemoglobinopathy	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Type:				Type:				
GI - hemorrhage	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	Abdominal/ testicular calcifications	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Location:				Location:				
Other:								

Neurological			
<input type="checkbox"/> NE	Yes	No	NE
Stroke	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Ischemic			
Hemorrhagic			
Transient ischemic attack	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
Cerebral aneurysm	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>



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Other: