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CLINICAL CHECKLIST COL2A1

FOR COL2A1 SCREENING ONE LABORATORY REQUEST AND ONE CLINICAL CHECKLIST SHOULD BE COMPLETED PER PERSON.

These forms may be requested by phone or downloaded from the website.

Patient information

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

Referring Physician:
Address:

Material sent:
☐ EDTA blood ☐ DNA ☐ Skin biopsy ☐ Chorionic villi
☐ Heparin blood ☐ RNA ☐ Bone biopsy ☐ Amniocytes
☐ Buccal swab ☐ Fibroblasts ☐ Paraffin embedded material
☐ Other:

Referring diagnosis (maximum 1)

☐ Achondrogenesis type 2 / Hypochondrogenesis
☐ Platyspondylic 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:

Phenotypic features

Measurements ☐ **NE (not examined)** **Age:**
Height: cm Weight: kg Head circumference: cm
Other:

Name patient (please complete): ……

Ophthalmological

☐ NE

	Yes	No	NE		Yes	No	NE
Myopia:	☐	☐	☐	Type 1(*) vitreous anomaly	☐	☐	☐
If yes, degree:				Type 2 (**) vitreous anomaly	☐	☐	☐
Cataract:	☐	☐	☐	Optically empty vitreous	☐	☐	☐
Glaucoma:	☐	☐	☐	Vitreous veils and bands	☐	☐	☐
Retinal tear:	☐	☐	☐				
Retinal detachment:	☐	☐	☐				
Chorioretinal atrophy	☐	☐	☐				
Perivascular pigmentation	☐	☐	☐				

(*) 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.

Orofacial

☐ NE

	Yes	No	NE
Cleft palate	☐	☐	☐
Submucous cleft	☐	☐	☐
Bifid uvula	☐	☐	☐
Flat face/midfacial hypoplasia	☐	☐	☐
Micrognathia	☐	☐	☐
Low nasal bridge	☐	☐	☐

Ears (please provide copy of audiogram)

☐ NE

	Yes	No	NE
Conductive hearing loss	☐	☐	☐
Sensorineural hearing loss	☐	☐	☐

Musculoskeletal

☐ NE

	Yes	No	NE	Beighton score (***)	Age
Joint hypermobility	☐	☐	☐		
Degenerative arthropathy	☐	☐	☐		
Joint replacement surgery	☐	☐	☐		
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

☐ 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

Family

	Yes	No	NE
Family history positive	☐	☐	☐

If Yes, please provide copy of family pedigree.

Versie 001/11-06-2010

Please see overleaf