



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

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:

Phenotypic features

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

Name patient (please complete):

| Ophthalmological | | | | | | | |
|-----------------------------|--------------------------|--------------------------|--------------------------|------------------------------|--------------------------|--------------------------|--------------------------|
| <input type="checkbox"/> NE | Yes | No | NE | | Yes | No | NE |
| Myopia: | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> | Type 1 (*) vitreous anomaly | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| If yes, degree: | | | | Type 2 (**) vitreous anomaly | <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"/> | | | | |

(*) 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 | | | |
|--------------------------------|--------------------------|--------------------------|--------------------------|
| <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 | |

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