

Spondyloepiphyseal Dysplasia Congenita (SEDC) OMIM: 183900
Kniest Dysplasia OMIM: 156550
Gene: COL2A1 Locus: 12q13.11-q13.2 OMIM: 120140

SERVICE: mutation and dosage analysis of the COL2A1 gene

TESTING: **Diagnostic*:** clinically affected patients
Presymptomatic: patients at risk of SEDC/Kniest dysplasia (known mutation)
*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: **Clinical Geneticists, Fetal Medicine specialists and Perinatal pathologists only**
The laboratory does NOT accept referrals directly from patients

TARGET REPORTING TIME AND COSTS (NCG funded for patients from England)

(Non UK National Health Service patients are subject to a surcharge. Payment must be agreed prior to testing – please include invoice form A)

Diagnostic:	8 weeks	£850
Presymptomatic:	2 weeks	£145
Microsatellite analysis	2 weeks	£415 (family group of three individuals)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1-54 and splice site boundaries of the COL2A1 gene
- Multiplex ligation dependent probe amplification for COL2A1 gene

SAMPLE REQUIREMENTS

- 1-5ml blood in EDTA or 50ul DNA (concentration ~500ng/ul)
- All patient samples must be labelled with **name, date of birth and Hospital/NHS number**
- Samples should be accompanied by a FULLY completed request card (available from the laboratory)
- Please include details of test, family history, patient address & postcode, GP, referring clinician and unit/hospital
- **Samples and paperwork must include three unique and matching patient identifiers**

SHIPPING DETAILS

- DNA can be sent by first class post
- Blood must be appropriately packaged and preferably sent by courier to arrive as soon as possible.
- Do not freeze prior or during postage.

CONSENT

It is the responsibility of the referring clinician to ensure consent has been obtained for:

- testing and storage
- the use of the sample and the information generated from it to be shared with members of the patients family and their health professionals

After testing, part of this sample might be used anonymously for the development of new tests and to monitor the quality of laboratory results.

CONTACT DETAILS

Genetics Laboratories, Box 143
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Addenbrooke's Hospital
Cambridge CB2 0QQ
Tel: +44 (0) 1223 348866
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Email: becky.treacy@addenbrookes.nhs.uk

Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

UKGTN testing criteria

UK Genetic Testing Network

Name of disease(s): **Spondyloepiphyseal dysplasia congenita (SEDC)
Kniest dysplasia**

Name of gene(s): **COLLAGEN, TYPE II, ALPHA 1; COL2A1**

Patient name: _____ **Date of birth:** _____

Patient postcode: _____ **NHS number:** _____

Name of referrer: _____

Title/Position: _____

Department/Hospital: _____

Contact email/telephone number: _____

Referrals will only be accepted from one of the following:
(Please indicate with a tick which category refers to the referrer).

Referrer	Tick if this refers to you.
Consultant Clinical Geneticist	<input type="checkbox"/>
Fetal Medicine Specialist	<input type="checkbox"/>
Perinatal Pathologist	<input type="checkbox"/>

Minimum criteria required for testing to be appropriate:

Criteria	Tick if this patient meets criteria
<u>Prenatally:</u>	
Shortened long bones (<3rd centile) detected in the second/third trimester +/- reduced thoracic circumference, micrognathia	<input type="checkbox"/>
<u>Postnatally:</u>	
Short stature with other clinical signs of SEDC/KD including: Cleft palate, SNHL, vitreoretinal abnormalities, joint hypermobility, micrognathia	<input type="checkbox"/>
Characteristic radiographic features: Absent ossification of pubic rami, dumb-bell shaped long bones, platyspondyly and anisospondyly, epiphyseal delay and irregularity/fragmentation, odontoid hypoplasia, small carpal bones	<input type="checkbox"/>

If the sample does not fulfil these criteria and you still feel that testing should be performed please contact the molecular genetics laboratory