

Stickler syndrome types 1 & 2 OMIM: 108300; 604831

Gene: COL2A1; COL11A1 Locus: 12q13.11-q13.2; 1p21 OMIM: 120140; 120280

SERVICE: mutation and dosage analysis of the COL2A1 and COL11A1 genes
microsatellite analysis to determine linkage to either COL2A1 and COL11A1 loci

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

REFERRALS: **Clinical Geneticists, Cleft Services and Ophthalmologists 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 (single gene sequence and dosage)*
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
- PCR and fluorescent sequence analysis of exons 1-68 and splice site boundaries of the COL11A1 gene
- Multiplex ligation dependent probe amplification for COL2A1 or COL11A1 gene

Following clinical assessment to assign vitreous phenotype, sequence analysis detects >90% of mutations in patients with the type 1 vitreous phenotype (COL2A1), and >60% of mutations in patients with the type 2 vitreous phenotype (COL11A1)

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
Level 6, Addenbrooke's Treatment Centre
Addenbrooke's Hospital
Cambridge CB2 0QQ
Tel: +44 (0) 1223 348866
Fax: +44 (0) 1223 348870
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):	Stickler syndrome type 1 or 2; STL1 & STL2
Name of gene(s):	COLLAGEN, TYPE II, ALPHA 1; COL2A1 COLLAGEN, TYPE XI, ALPHA 1; COL11A1

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"/>
Consultant Ophthalmologist with vitreoretinal specialty expertise	<input type="checkbox"/>

Minimum criteria required for testing to be appropriate:

Criteria	Tick if this patient meets criteria
Clinically affected individuals, assessed by Consultant ophthalmologist with vitreoretinal specialty expertise AND	<input type="checkbox"/>
For type 1:	<input type="checkbox"/>
a) Type 1 congenital vitreous anomaly (Type 1 membranous vitreous anomaly: is visualised on slit-lamp biomicroscopy as a highly folded membrane bordering a vestigial secondary gel in the immediate retrolental space) AND	<input type="checkbox"/>
b) Sensorineural deafness AND/OR	<input type="checkbox"/>
c) Midline cleft AND/OR Pierre Robin sequence AND/OR	<input type="checkbox"/>
d) Arthropathy OR epiphyseal dysplasia	<input type="checkbox"/>
For type 2:	<input type="checkbox"/>
a) Type 2 congenital vitreous anomaly (type 2 vitreous anomaly: visualised on slit-lamp biomicroscopy as sparsely and irregularly thickened bundles of collagen fibres distributed throughout the secondary vitreous cavity) AND	<input type="checkbox"/>
b) Sensorineural deafness AND/OR	<input type="checkbox"/>
c) Midline cleft AND/OR Pierre Robin sequence AND/OR	<input type="checkbox"/>
d) Arthropathy OR epiphyseal dysplasia	<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