A13/S(HSS)/b

2013/14 NHS STANDARD CONTRACT
FOR COMPLEX EHLERS DANLOS SERVICE (ALL AGES)

PARTICULARS, SCHEDULE 2- THE SERVICES, A- SERVICE SPECIFICATIONS

Service Specification No.	A13/S(HSS)/b
Service	Complex Ehlers Danlos service (All Ages)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

The estimated prevalence of Classic Ehlers Danlos syndrome (EDS) is 1 in 20,000 (Byers PH 2001). The prevalence of vascular EDS has been estimated at 1 in 50,000. (Steinmann B et al 2002). 50% of affected people have affected family members and 50% are new mutations.
The service aims to achieve equality of excellent holistic care for all children and adults with suspected complex EDS throughout the country. The first step is accurate diagnosis.

Currently patients suspected of having EDS are assessed and diagnosed in UK regional clinical genetics clinics and various other secondary and tertiary speciality clinics such as dermatology, rheumatology, paediatrics, vascular surgery, gastroenterology and neurology. The EDS specialist diagnostic service runs two specialist clinics for patients who meet the referral criteria.

Description of the disease/condition

EDS is a group of heritable disorders of connective tissue. The major clinical features are hyperextensible skin, hypermobile joints and tissue fragility. There is significant morbidity and mortality associated with some subtypes (Steinman B, Royce P, Superti-Furga A 2002a): EDS type IV patients are at particular risk of lethal arterial aneurysms anywhere between the aortic arch and distal medium sized arteries, including especially the renal, internal carotid and ileofemoral arteries, (Byers PH 1995, Pope et al 1996); whilst spontaneous colonic perforations and pneumothoraces are also common. Late aortic rupture or aortic dilatation is also a risk for some EDS families. Pregnancy is associated with significant morbidity in EDS type IV for reasons such as uterine rupture (Peaceman AM, Cruikshank DP 1987), whilst perineal tearing is common to all EDS subtypes (Lind J et al 2002). The

premature rupture of foetal membranes is particularly associated with EDS types I & II (Smith LT *et al* 1992). The latest classification (Beighton P *et al* 1998) delineates seven subtypes that are defined by major and minor criteria. Each type of EDS is a distinct disorder.

1.1 National/local context and evidence base

A key role of the specialist clinic is to develop, audit and refine the referral criteria and diagnostic pathways to ensure that testing is strictly selected and therefore cost-effective. In addition the clinic provides a hub for the development of model pathways and treatments for the regional and local services.

By providing a specialised expert service for atypical or complex cases of EDS the clinical service aims to reduce national costs by:

- reducing times to diagnosis
- reducing the number of hospital appointments required to achieve a specific diagnosis
- matching treatment to diagnosis
- reducing unnecessary tests
- maximising the management efficiency of uncomplicated cases in secondary and tertiary care.

Cost effectiveness of the clinics will progressively improve from factors such as:

- better local secondary and tertiary diagnosis and selective testing as a result of quaternary co-ordination and education
- improving tertiary expertise will reduce inappropriate quaternary referrals, except for complex or difficult diagnostic problems. Although these should plateau, there will be significant demand
- reduction in average laboratory costs by exploiting automation and technological development
- increasing understanding of genotype-phenotype correlations that will result in better targeting of molecular genetic testing
- earlier predictive testing of at-risk individuals with the aim of minimising long term complications and developing efficient preventative screening.

Development of other laboratory analyses such as further development of collagen analysis to deliver easier and more rapid profiling of collagen species or improvements in electron microscopy may contribute to more accurate and cheaper diagnostic pathways.

2. Scope

The role of the specialist clinic.

The service will provide a fully comprehensive service under the auspices of the clinical genetics service for the precise clinical diagnosis and management of a sub-set of patients with all types of EDS in whom either:

- the clinical diagnosis is not straight forward, or the clinical diagnosis is one of EDS but laboratory testing has not confirmed the diagnosis and further clinical evaluation is necessary.

2.1 Aims and objectives of service

The aims of the clinic will be to:

- correctly diagnose and investigate complex cases of EDS
- minimise inappropriate and costly investigations
- educate referrers about diagnosis, investigation and management of patients with EDS, encouraging appropriate referral to the clinic
- develop guidelines and pathways of care for the different subtypes of EDS that can be used in secondary and tertiary care.

The subdivision of EDS into its various subgroups reflects the current state of clinical practice in other single gene disorders and has the following general benefits:

- it enhances the accuracy of genetic risk assessment
- if the gene and mutation is identified, predictive and diagnostic testing is consequently available, for probands and their relatives
- it improves the co-ordination of health surveillance of specific rare disorders
- it provides novel information and focussed expertise to various primary, secondary and tertiary practitioners and other medical carers involved in the management of these particular disorders
- it refines and enhances all aspects of clinical management of these complex disorders, which would not otherwise be accessible to the primary, secondary and tertiary practitioners listed above;
- it enhances the accuracy of genetic registers, as judged by, complete ascertainment of affected individuals, risk estimation and family planning;
- it fulfils various recommendations of the clinical genetic community for the testing of rare genetic single gene disorders such as:
  - the testing for those individual defects, which are too rare to be economical for individual genetic regional units to assess and test
  - it fulfils the recommendation that for such rarities two genetic centres are sufficient to provide efficient back-up and quality testing assurance
  - it promotes UK self-sufficiency in this complex field, which would not otherwise be addressed in the UK
  - it fosters comprehensive coverage of this specialised field and provides

otherwise unavailable expertise to non-expert primary, secondary and tertiary UK practitioners, who do not have specialised expertise in these matters.

For example the pathway described in section 5.4 of that publication is very similar to that of Col3a1 deficient patients with vascular EDS IV. Similar considerations apply to all those EDS subtypes with testable parameters, such as electron microscopical, structural protein, enzyme or DNA mutations, including EDS types I/II, VIA, VIIA, B & C and certain EDS III variants. Similarly such analysis is also desirable in future for all other subtypes in which the molecular defects is so-far unknown.

2.2 Service description/care pathway

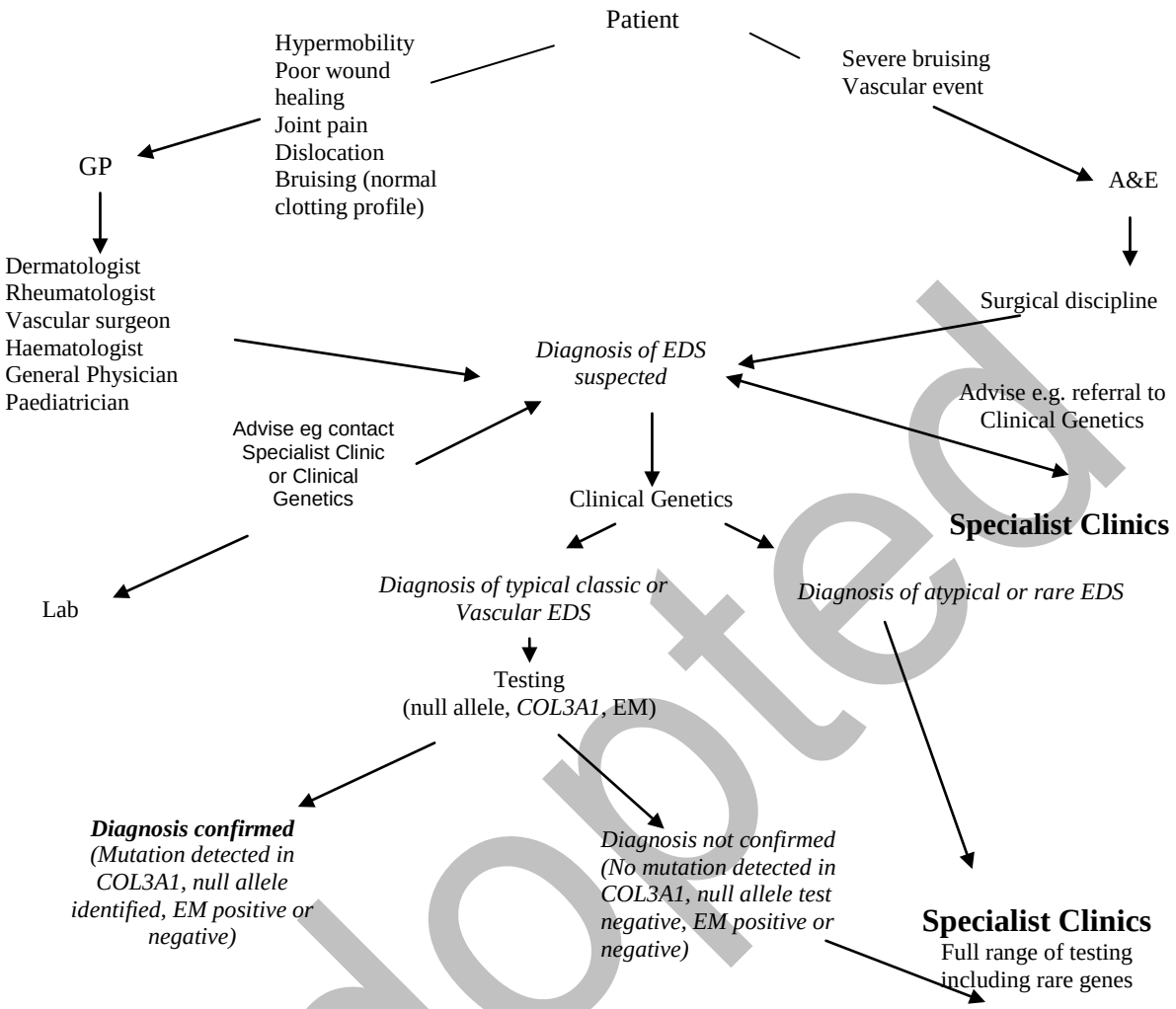
Service description

The service provides a fully comprehensive service under the auspices of the clinical genetics service for the precise clinical diagnosis and management of a sub-set of patients with all types of EDS in whom either:

- the clinical diagnosis is not straight forward, or
- the clinical diagnosis is one of EDS but laboratory testing has not confirmed the diagnosis and further clinical evaluation is necessary.

The detail of the service includes:

- correct diagnosis and investigation of complex cases of EDS
- minimising inappropriate and costly investigations
- educating referrers about diagnosis, investigation and management of patients with EDS, encouraging appropriate referral to the clinic
- developing guidelines and pathways of care for the different subtypes of EDS that can be used in secondary and tertiary care.



Days/hours of operation

As a specialist diagnostic service, the hours of operation are office hours Monday to Friday 0900 – 1700 hours

2.3 Population covered

The service is commissioned by NHS England for all eligible patients from England and Scotland.

2.4 Any acceptance and exclusion criteria

Referral criteria, sources and routes

Referrals are accepted from consultants in secondary care/tertiary care for patients in whom the diagnosis of EDS is suspected but not confirmed for one of the

following reasons:

- diagnostic criteria according to Villefranche Classification not met

- diagnostic testing does not confirm diagnosis suspected
- diagnostic criteria of more than one type of EDS identified
- significant additional findings aside from diagnostic criteria.

Referrals are also accepted where the patient or clinician requests a second opinion after initial consultation.

The provider has a duty to co-operate with the commissioner in undertaking Equality Impact Assessments as a requirement of race, gender, sexual orientation, religion and disability equality legislation.

Exclusion criteria

The exclusion criteria refer to patients who do not fulfil the referral criteria. If patients do not fulfil the referral criteria, the referring physician is contacted and advised on the appropriate route for the patient.

Response time & detail and prioritisation

All patients referred to the specialist service are required to meet the targets of the 18-week patient pathway.

Discharge criteria

Patients are discharged following their final appointment at which results are given and explained. Detailed letters are written to the referring doctor and to the patient these will include information on their advised ongoing management (to be provided locally).

2.5 Interdependencies with other services

Whole system relationships and interdependencies
After diagnosis, GPs and referring clinicians have a responsibility to care for the patient in line with their individual management plan.

Relevant networks and screening programmes
Not applicable.

3. Applicable Service Standards

3.1 Applicable national standards e.g. NICE, Royal College

Providers will carry out a mandatory patient note and consent audits in accordance with the Trust audit requirements.

All audits and the patient satisfaction survey must be registered with the trusts audit department.

Providers must comply with the trusts requirements for safe haven status when transferring patient information between NHS organisations.

4. Key Service Outcomes

Quality Performance Indicator	***Threshold***	***Method of measurement***	***Consequence of breach***	***Report Due***
Clinic Letters	to be sent out to patient and referrer within 14 days			
Waiting time	patient to be seen in clinic within 18 weeks			
Molecular Laboratory	genetic test results to be obtained within 20 weeks			

5. Location of Provider Premises

Sheffield

The clinical service is provided by a consultant dermatologist with training in clinical genetics under the governance of the Sheffield clinical genetics service, Sheffield Children's NHS Foundation Trust

The diagnostic service is housed within the directorate of diagnostics in the molecular and biochemical genetics laboratories under the heads of each department at Sheffield Children's NHS Foundation Trust

Sheffield Children's NHS Foundation Trust
Western Bank
Sheffield
S10 2TH

Electron microscopy is provided the Northern General Hospital, who presently processes the Sheffield Electron Microscope requests.

Electron Microscopy
Department of Histopathology
Northern General Hospital
Sheffield Teaching Hospitals NHS Foundation Trust,
Herries Road
Sheffield
S5 7AU

London

Clinical service provided by consultant dermatologist and honorary Professor of Clinical Genetics and a consultant clinical geneticist, under the governance of:

North West Thames Regional Genetics Service (Kennedy Galton Centre)
North West London Hospitals NHS Trust,
Level 8
Watford Road
Harrow
Middlesex
HA1 3UJ

Laboratory service provided by a senior scientist with appropriate technical and administrative support under the governance of the molecular genetics department at Northwick Park Hospital (part of North West London Hospitals NHS Trust).

Electron Microscopy is provided by:

Nuffield Department of Clinical Laboratory Science
University of Oxford
John Radcliffe Hospital
Oxford
OX3 9DU.

Sub-contractors

Not applicable