National Specialist Services Committee (NSSC)

STAGE 2 Specialist Service: Detailed Proposal

Applicants must refer to NSSC Guidance which can be found on the NSD Website before completing a proposal: NSSC-Guidance for completing Proposals - Specialist Service.pdf

Section 1: Summary information
1.1 Full name of proposed service
Scottish Centre for Complex Ehlers Danlos Syndrome
1.2 Short title for proposed service
SCCEDS
1.3 Name, title, and contact details of lead applicant
Professor Stuart H Ralston Professor of Rheumatology and Honorary Consultant Rheumatologist Centre for Genomic and Experimental Medicine University of Edinburgh Western General Hospital Edinburgh EH5 3LH Email: [redacted]@ed.ac.uk Phone: [redacted] Peter McLoughlin Strategic Planning Programme Manager NHS Lothian Western General Hospital Email: [redacted]@nhslothian.scot.nhs.uk
1.4 **Any additional information that was requested at stage 1**
NPPPRG requested that the patient group be defined more precisely, that consideration should be given to including other non-inflammatory connective tissue disorders, and that questions around diagnostic routes, referrals, staffing levels and relationships with existing services should be addressed. NSSC added that input should be sought from rheumatology and orthopaedic colleagues and that that the longer term plans of the service with regard to continued national designation are outlined.

Section 2: Description of proposed service
2.1 Summary of patient need
5. Summary of patient need and scale of the problem There is a substantial unmet clinical need to establish a centre that offers assessment and treatment for patients with complex Ehler's Danlos Syndrome hypermobility type (hEDS). Patients with complex hEDS are getting a raw deal at present as is evident from representations made by patient testimonials and patient support groups to the Scottish Parliament and other agencies. At the present time patients with complex hEDS report significant delays in the correct diagnosis being reached and even once a diagnosis has been made they are not being offered that advice and support they need to take control and self-manage their condition effectively. In the absence of a specialist service in Scotland, many patients are being referred to tertiary centres in England at significant cost, whereas other individuals who can afford it, are being seen by consultants in the private sector based in London. Frequently, the patients then return to their local NHS service with demands for a range of treatment and investigations

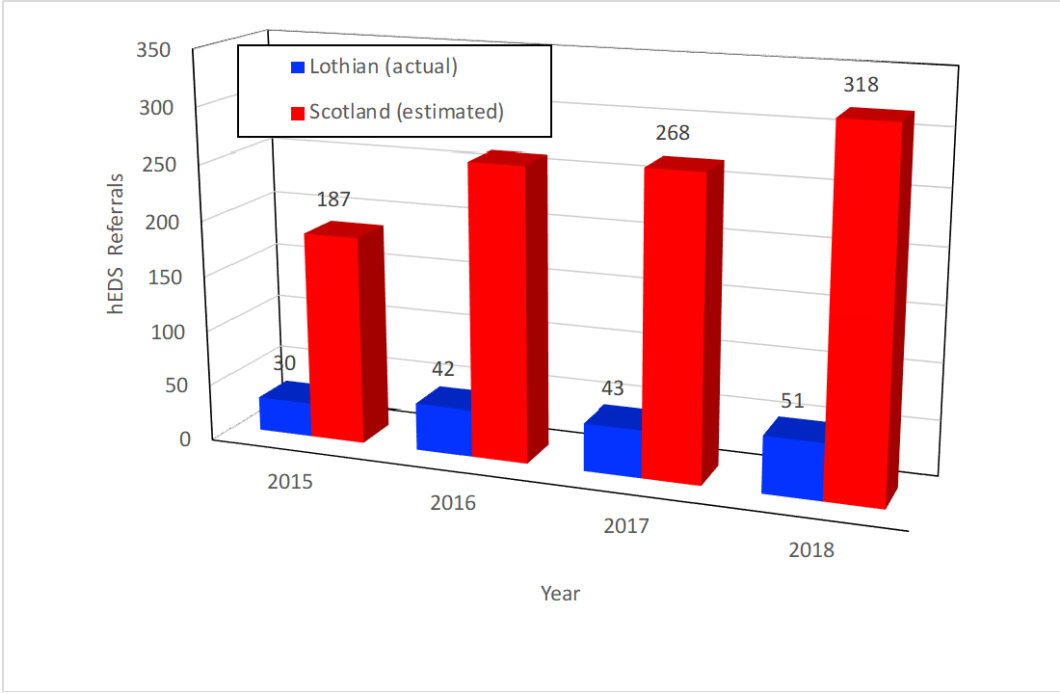
including genetic testing that lack an evidence base. Often these treatments are not available and investigations are not indicated, leading to dissatisfaction from the patient perspective and resentment from the perspective of the health care professional.

The defining feature of Ehler's syndrome is ligament laxity, which is often accompanied by disabling musculoskeletal pain and functional impairment. In a proportion of cases, other features such as recurrent dislocation, skin laxity, kyphoscoliosis and symptoms referable to the cardiovascular system, gastrointestinal system, or genitourinary system may be observed.

There are few data on the prevalence of hEDS, but current estimates suggest it affects between 1 in 5000 to 1 in 10,000 people, which means that there may be between 500 and 1000 people in Scotland with the condition. Many people with hEDS do not require specialist input since the diagnosis can be made clinically based on history and examination and often no specific treatment is required. However, a proportion of patients with hEDS present with chronic and disabling and pain, recurrent dislocations, and kyphoscoliosis, along with various cardiovascular, gastrointestinal or genitourinary problems. We define this as complex hEDS.

For example, a survey of 384 patients conducted by the Ehlers Danlos Support in Scotland revealed an average time of 19 years between the presenting symptoms and a diagnosis being reached; that only 16% had been able to see a healthcare professional that was aware of EDS and that 87% of patients did not feel their needs were being adequately addressed by the NHS in Scotland due to a lack of interest by the Medical Profession in helping these individuals to understand and to manage their condition effectively. A common theme of patient testimonials is that even after a diagnosis had been made, health care professionals were either not interested or felt they did not have the skills to managing their condition effectively.

In order to gain an insight into the scale of the problem we have audited the number of referrals with a diagnosis of EDS that have been treated by physiotherapy and/or occupational therapy at rheumatic diseases unit in Edinburgh. Some of these referrals have derived from primary care, others from rheumatology clinics and others from genetics clinics. Based upon this we expect that up to 350 hEDS patients in Scotland may need referred to secondary care per year but it's likely that a smaller number will have complex hEDS as defined above.



2.2 Aims and Objectives of service

To establish a centre for the assessment, and management of patients with complex Ehler's Danlos Syndrome (hEDS) whose needs are not being adequately met at a local level. The centre would accept referrals from secondary care in NHS Boards throughout Scotland, where it was felt that the patients' needs could not be adequately addressed locally. The centre would offer a holistic package of care. This would start with a detailed assessment by a consultant experienced in the condition to reaffirm the diagnosis, provide education on the nature of the condition, and develop a treatment plan for the musculoskeletal and extra-skeletal manifestations of the disease. This would be followed by an assessment by an experienced physiotherapist to deliver advice on movement and muscle strengthening exercises as well as manual therapy if appropriate. This would be followed by an assessment by an occupational therapist who will provide advice on helping patients to improve their ability to function independently so that they can participate in activities that are important to them. If necessary, the patients will be assessed by a clinical psychologist to address issues such as depression or behavioural, emotional and psychological distress with the aim of helping patients come to terms with their condition and promoting well-being.

It is anticipated that patients accepted into the programme would have an initial assessment and treatment package delivered over two days, with a further review and treatment after 2-3 months and a final assessment and review after 6 months at which point, the patient would be discharged to the referring NHS board with a long-term treatment plan that can be delivered locally.

Since feedback from patient organisations is that there is a lack of awareness about hEDS and the management strategy for the condition, an important role of the centre will be to develop educational materials resources for doctors and AHP across Scotland and run courses on hEDS management so that the shared care treatment model will work effectively in the longer term. The SCCEDS would also develop a research programme into EDS to better understand the pathophysiology of the condition with an aim of identifying new treatment strategies. In summary, SCCEDS would act as a centre for excellence for the management of EDS as well as a hub for education, training and research.

2.3 Description of proposed delivery of service including the patient pathway of care

Referrals into the service will be accepted from Consultant Rheumatologists across Scotland. The criteria for referral will be:

1. Clinical diagnosis of hEDS confirmed by a consultant rheumatologist with debilitating symptoms that persist despite an adequate trial of supportive therapy delivered locally.

Referrals will be reviewed by one of the SCCEDS consultants and if the patient meets the criteria an appointment will be made by the SCCEDS manager. The service will be based at the Western General Hospital (WGH) in Edinburgh. Patients entering the service will have an initial assessment spread over two days; will have a follow up appointment at 3 months and then will have a final appointment and will be discharged from the service after 6 months. In exceptional circumstances patients may be seen for further review visits if this is thought to be clinically indicated and agreed by the referring rheumatologist and one of the SCCEDS consultants.

<u>Initial assessment</u>

The initial assessment will take place at the outpatient clinic by a consultant rheumatologist where there will be a 45-minute consultation to reaffirm the diagnosis to educate the patient about the nature of their condition and to develop a treatment plan. The patient will then be seen at the therapies department at the WGH where they will have a 60-minute consultation with a physiotherapist (PT) following by a 60-minute consultation with an occupational therapist (OT). In some circumstances this may be followed by a 60-minute consultation with a clinical psychologist (CP). The following day, the patient will have further 30-minute therapy sessions with the PT and OT to reinforce the treatment plan.

Initial review
The patient will be reviewed at 3 months and seen by the consultant (30 minutes) PT (30 minutes), OT (30 minutes) and (if necessary) the CP.

Final review and discharge
There will be a final review marking discharge from the service at 6 months with consultant, PT and OT review. The patient will then be discharged to the referring health board with a long-term treatment plan.

Summary of 3 step new patient clinic model:
1/ 1st visit – Initial Assessment. Consultant and AHP. Initial review and care planning. Patient experiences circa .05 day contact with clinician on day 1, returning day 2 to receive care plan and advice from AHP's. Overnight accommodation required for 1 night per person (both patient and relative)
2/ 2nd visit – Review Assessment. Undertaken after 2 – 3 months. Circa 2 hours. Nurse and AHP lead.
3/ 3rd visit – Appointment with full MDT – Undertaken after circa 6 months - circa 30 mins with consultant and circa 2 hours AHP. Review, plan, and discharge to referring NHS board.

A specialist national service – building regional capacity
This new national service will provide specialist assessment, and management of patients with complex Ehler's Danlos Syndrome (hEDS) whose needs are not being adequately met at a local level. Following the initial period of specialist assessment and formulation of a long-term care plan, patients will then be discharged to the referring health board. Given that this model lends itself to sharing learning and building capacity in Rheumatology and related therapy services across Scotland, it would be our intention to work towards developing a 'Regional Hub and spoke' model of service. In this model, increasingly specialist levels of assessment could be undertaken in local units in the West and North regions of Scotland, thereby reducing over time the continuing caseload demand on the Edinburgh national specialist service. It is anticipated that developing capacity, knowledge and skills would take some time to deliver. We would envisage that this would be a stated aim of the specialist national service in Edinburgh, which would work towards sharing the learning and approach to assessment and management of the patient group, and provide opportunities for training, visits, research and other collaborative development. The engagement of regions and local units would be required explicitly for this, and a form of network management resource may be required to facilitate the development.

Section 3: Evidence of the Clinical Benefits

3.1 Evidence for clinical effectiveness and potential for health gain of the proposed service

The diagnosis of hEDS does not require any special facilities in terms of imaging or other diagnostic tests. Very rarely, genetic testing may be required in individuals where a monogenic form of hypermobility is suspected.

The management of hEDS is mainly based on well-established techniques such as physiotherapy, occupational therapy and clinical psychology to promote self-management. Therapeutic intervention may also be required for chronic pain and this will be delivered along the principles outlines in SIGN 136 (Management of Chronic pain).

The potential for health gain is considerable. By using these interventions appropriately, we expect to be able to help patients to maximise their quality of life and return to normal functioning

3.2 Benefit over current NHS alternatives

The NHS in Scotland is currently failing these patients who are already extensive users of NHS resources. They are typically being referred to multiple specialist clinics including rheumatology, pain medicine, gastroenterology, cardiology and gynaecology but are not getting the treatment that is required. Currently, following referral, typical waits for AHP assessment is circa 8 months (NHS Lothian).

The centre offers the prospect of saving NHS resources by confirming the diagnosis, by educating the patient about their condition and by developing a treatment plan that will empower the patient to optimise function and effectively self-manage their condition.

3.3 Clinical safety and risk record of the proposed service

The proposed service will be based in the rheumatic disease unit, NHS Lothian. The unit has an exemplary record of clinical safety and managing risk.

3.4 Performance and Quality Monitoring Measures

We will used standardised tools including to document quality of life (SF36 and ED5D questionnaires), pain (BPI questionnaire) and physical functioning (HAQ). These will be assessed at the time of referral to the centre, at follow up visits and on discharge. In addition, we will administer patient satisfaction questionnaires to gain feedback on the patient view of their journey and to identify any unmet needs that exist. We will also ask physicians in referring health boards to complete a survey on their satisfaction with the service and to suggest any gaps that may exist in what is being delivered.

3.5 Does this proposal fit with principles of realistic medicine

Yes. The proposed service is entirely consistent with the principles of realistic medicine since it will empowerment the patient to work with physicians and therapists to optimise function and gain control of their condition. Such an approach offers potential to reduce demand on various specialties, whilst supporting delivery of efficient, integrated and personalised care.

Section 4: Societal benefits:

4.1 Details on the equity impact of the proposed service on patients and the wider society

The proposed service is expected to provide hEDS patients with the assessments and treatment they need and are at present sadly lacking. Therefore, patients with this disorder will for the first time have equity with patients who have other rheumatological disorders.

4.2 Details of the impact of providing the proposed service on further clinical research and innovation

We expect that SCCEDS will lead the way in promoting clinical research into this disorder. Current evidence suggests that hEDS has a genetic component but that it is inherited in a polygenic manner. The predisposing genetic variants are unknown. Patients attending the centre will be invited to take part in a research programme which will seek to identify the predisposing genetic variants for hEDS, genotype phenotype relationships and environmental triggers. Funding for this will be sought by application to external agencies.

Section 5: Financial impact

5.1 Estimated costs

Staff Type	WTE	Band	Cost/ WTE	Total Cost
Consultant	1	Cons	136453	136453
Physio	1.5	7	52206	78309
OT	1.5	7	52206	78309
Psychologist	0.8	8A	60252	48201.6
Specialist Nurse	1	7	54034	54034
Business Manager	1	5	34925	34925

Medical Secretary	1	3	25774	25774
Health Records	0	2	23663	0
Outpatient Resource	0	5	34925	0
Staff Costs				**456005.6**
Non Pay				15000
Overheads @24%				113041.344
Total				**584047**

5.2 Current costs

Approximately 133k PA based on 60 Lothian patients. These costs are derived from case cost estimates from the service model described and costed in this submission. Actual costs are currently being absorbed by NHS Lothian and would require to be funded as part of any new service designation.

5.3 Potential savings to NHS Scotland

Potential to reduce demand on various specialties**.** This would require more detailed assumptions to be developed in order to estimate avoidable costs and will be considered in the next stage.

Section 6: Support for the proposal

6.1 Evidence of support for the proposal

A recent survey of consultant rheumatologists that deal with EDS has confirmed that while the majority feel comfortable in making the diagnosis, 64% feel that they are poorly placed or very poorly placed in managing the condition. In addition, 78% of rheumatologists felt that it would be helpful or very helpful to have a national centre for referral and 85% felt that it would be helpful or very helpful for the centre to provide education and training on the needs of these patients. A similar survey of orthopaedic surgeons in Scotland revealed that most felt poorly or very poorly placed in making a diagnosis and in deciding upon the need for clinical genetics referral, but that they were reasonably well placed in managing the condition from an orthopaedic standpoint. All of the orthopaedic surgeons surveyed (100%) felt that it would be very helpful to have a referral centre for EDS based in Scotland. Full details of the survey and provided as an Appendix B.

Letters of support has been obtained from [redacted] at the Royal College of Physicians of Edinburgh and Ehlers Danlos Support UK (Appendix C). We have written to the Scottish Society of Rheumatology (SSR), the Scottish Committee for Orthopaedics and Trauma (SCOT), the Scottish Cardiac Society (SCS) and the Scottish Society of Gastroenterology (SSG) outlining the issues experienced by hEDS patients in anticipation of obtaining for further letters of support but these are not available at the present time. Should letters of support be forthcoming from these organisations, they will be forwarded in due course.

6.2 Potential opposition to the proposal

We are unaware of any opposition to this proposal

6.3 Any other relevant information

Scottish Centre for Complex Ehlers Danlos Syndrome
Results of clinician survey

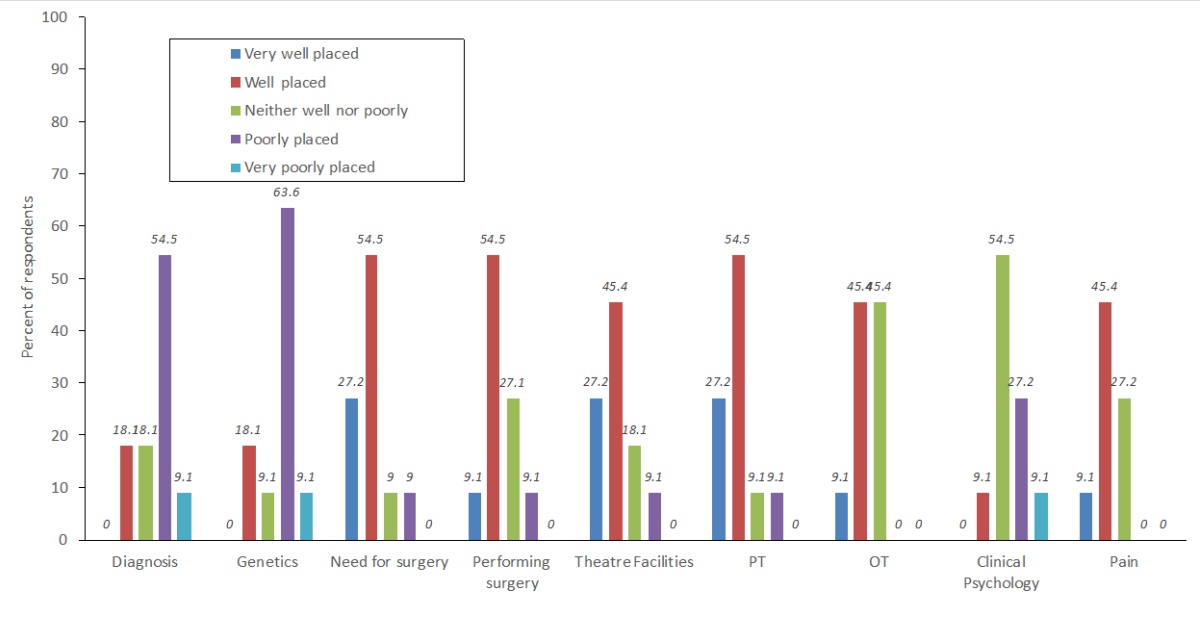
Executive summary
A web-based survey of orthopaedic surgeons and rheumatologists 'views on how well placed they were to deliver various aspects of care for patients with complex Ehlers Danlos Syndrome (EDS) was conducted between June and July 2019. Views were also sought in how useful it would be to have a national referral centre for EDS. The majority of respondents saw less that 5 patients annually but 43% of rheumatologist saw between 5-10 patients per year. Orthopaedic surgeons reported that they were poorly placed to make a diagnosis and to decide upon the need for referral to clinical genetics but were general well placed to decide upon the need for surgery and to conduct that surgery. They felt reasonably well placed with regard to physiotherapy, occupational therapy and management of pain. Rheumatologists felt well placed to make a diagnosis of EDS and to access other specialists, but views were mixed on how well placed they were to decide upon the need for a referral to clinical genetics. Most rheumatologists (78%) felt poorly placed with regard to their ability to manage EDS overall. Most felt they were poorly placed with regard to occupational therapy support and reported that they were neither well nor poorly placed with regard to support from clinical psychology and management of pain. All orthopaedic surgeons and 78% of rheumatologists felt that establishing a referral centre for EDS would be helpful or very helpful. In addition, 85% of rheumatologist and 90% of orthopaedic surgeons felt that if the centre was to provide training opportunities that would be helpful or very helpful. In conclusion, the survey does provide support within the orthopaedic and rheumatology community to establish a centre for complex EDS.

Methodology
A web-based survey of consultant orthopaedic surgeons and consultant rheumatologists in Scotland was performed between 25th June and 12th July 2019 by NSD in collaboration with the Scottish Orthopaedic Society and Scottish Society for Rheumatology. The survey was sent to all consultant members of both societies and responses were invited.

Results
Responses were obtained from 22 orthopaedic surgeons of whom 11 (50%) cared for patients with complex EDS as part of their normal practice. Of these 11 consultants, 9 (81%) saw fewer than 5 patients per year and 2 (19%) saw between 5-10 patients per year. Responses were obtained from 29 rheumatologists of whom 14 (48.2%) cared for complex EDS. Of these 8 (57%) of respondents saw fewer than 5 patients per year and 6 (43%) saw between 5 and 10 patients per year.

Orthopaedic surgeons
The orthopaedic surgeons generally felt poorly or very poorly placed with regard to making a diagnosis and deciding on the need for referral to clinical genetics but felt well placed or very well placed in deciding on the need for surgery, performing surgery and in terms of physiotherapy and occupational therapy support after surgery. Most respondents felt neither well placed nor poorly placed in regard to clinical psychology support. They felt reasonably well placed with regard to pain management. The responses are summarised in Figure 1.

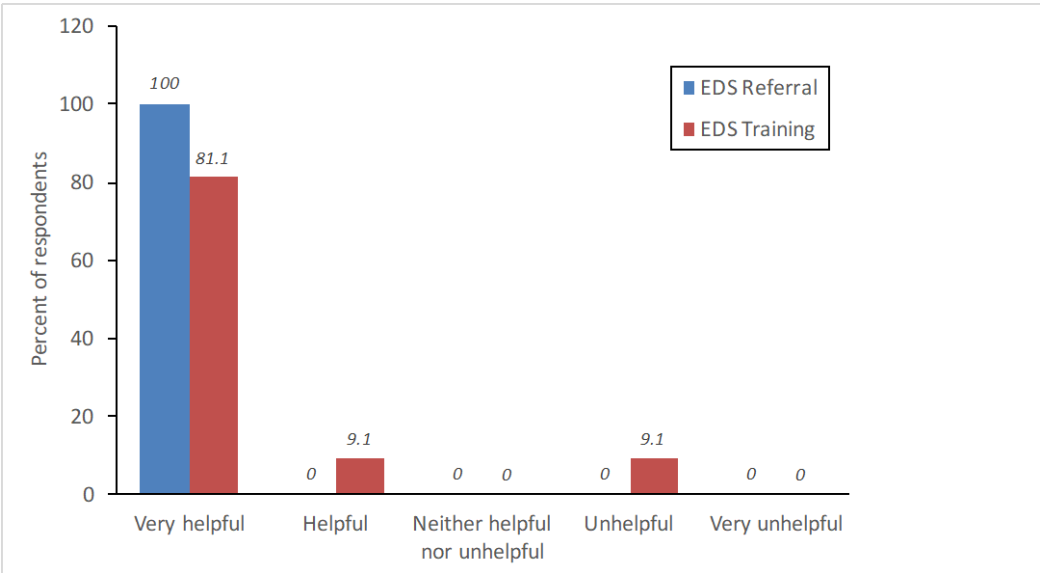
Figure 1. Orthopaedic views on their ability to deliver various aspects of EDS care



The graph shows how well orthopaedic surgeons felt they were able to care for EDS patients across various domains of care (PT- physiotherapy; OT – occupational therapy)

Orthopaedic views on the need for a specialist referral centre are shown in Figure 2. All respondents (100%) felt that it would be very helpful to have a specialist centre for patient referrals and 9 (81.1%) felt that training and educational opportunities provided by the centre would be very helpful.

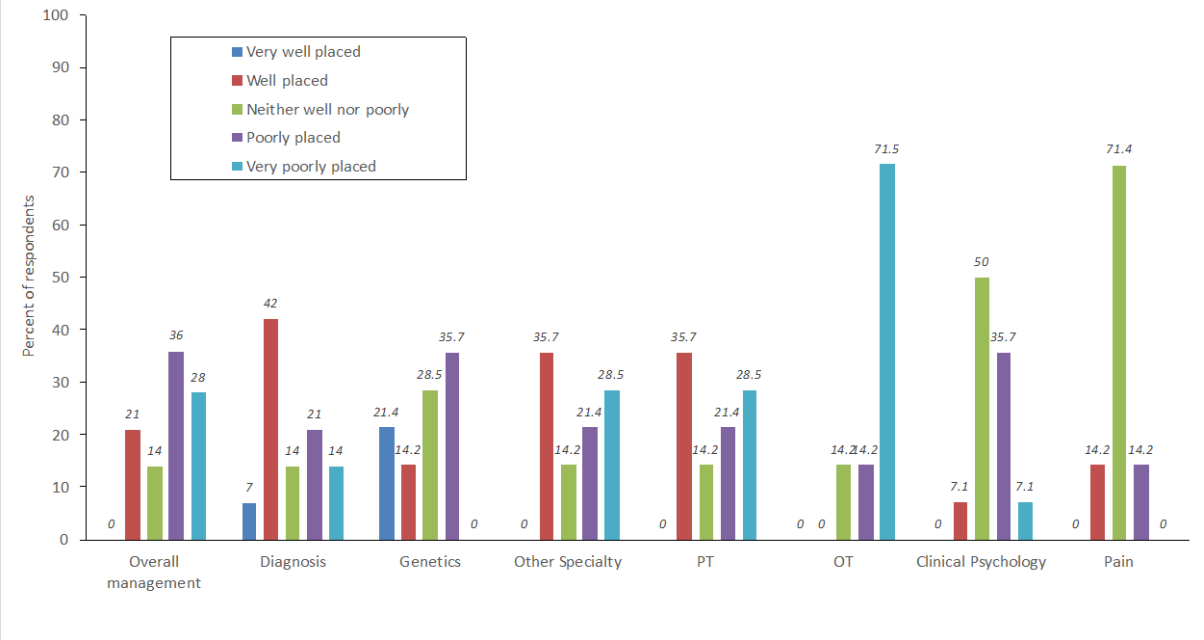
Figure 2. Orthopaedic views on need for an EDS centre



Rheumatologists

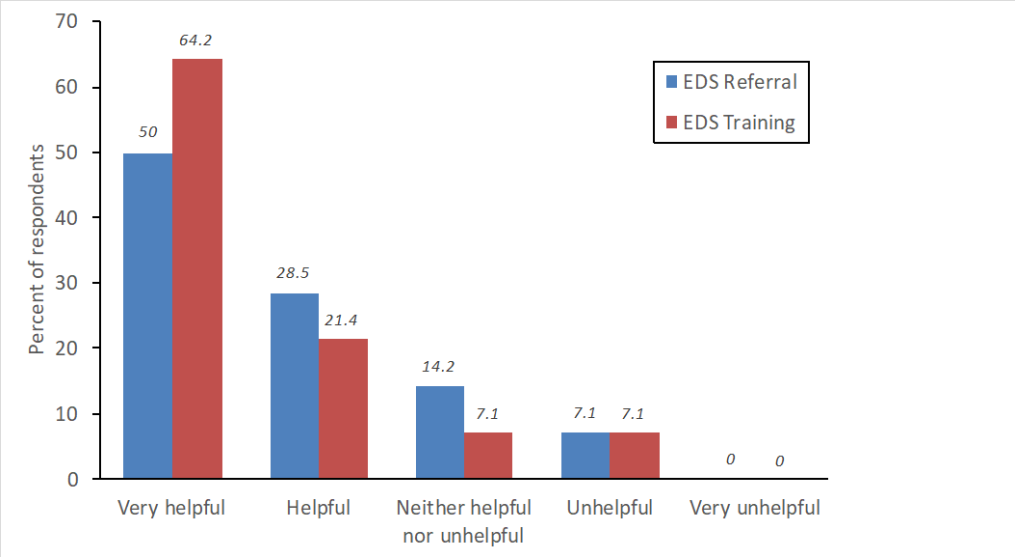
The majority of rheumatologists felt well placed with regard to making a diagnosis of EDS, but views were mixed on how well placed they felt in deciding on the need for referral to clinical genetics. Most rheumatologists felt well placed on accessing other specialist input, but the majority felt poorly placed to manage the condition overall. Most felt reasonable well placed with regard to physiotherapy support but very poorly placed with regard to occupational therapy. The majority of respondents reported that they felt neither well placed nor poorly placed in regard to clinical psychology support and pain management. The responses are summarised in Figure 3.

Figure 3. Rheumatologists views on their ability to deliver various aspects of EDS care



The graph shows how well rheumatologists felt they were able to care for EDS patients across various domains (PT- physiotherapy; OT – occupational therapy)

Rheumatologists views on the need for a specialist referral centre are shown in Figure 4. The majority of respondents (78%) felt that it would be very helpful or helpful to have a specialist centre for patient referrals and an even higher proportion (85%) felt that training and educational opportunities offered by the centre would be very helpful or helpful.



ROYAL COLLEGE of PHYSICIANS of EDINBURGH

26 July 2019

President's Office
[redacted]

Professor Stuart H Ralston, MD, FRCP, FMedSci, FRSE
Edinburgh Western General Hospital
Edinburgh
EH4 2XU

[redacted]@ed.ac.uk

Dear Professor Ralston

I am very pleased to write in support of your application to establish a referral centre for the assessment and management of patients with complex Ehler's Danlos Syndrome (EDS) within Scotland.

It is clear from the information summarised in your application that the needs of these patients are not currently being adequately addressed by the NHS in Scotland. I was also interested in the results of your survey of both rheumatologists and orthopaedic surgeons in Scotland who take care of these patients. I noted that while the majority of rheumatologists felt they were well placed to make the diagnosis, 64% felt that they were poorly placed or very poorly placed to manage the needs of these patients and that 78% of respondents felt that it would be helpful or very helpful to have a national referral centre to deal with the needs of these patients. I also noted that all of the orthopaedic surgeons surveyed supported such a centre.

On behalf of the college, I wish you success in establishing the proposed centre since it offers the prospect of initiating a personalised treatment pathway that is likely to have enduring effects in helping to support patients in self-managing their own condition in the longer term.

With kind regards
Yours sincerely

[redacted]

President

9 Queen Street, Edinburgh, EH2 1JQ
[redacted]

EHLERS-DANLOS SUPPORT UK

[redacted]
National Services Division
Area 062, Gyle Square
1 South Gyle Crescent,
Edinburgh
EH12 9EB

25th July 2019

Dear [redacted]

Ehlers-Danlos Support UK (EDS UK) is very pleased to provide this letter of support for the development of a new national specialist centre in Edinburgh for the diagnosis and treatment of complex hypermobile Ehlers-Danlos syndrome (hEDS) and hypermobility spectrum disorders (HSD).

As you are aware patients with complex forms of these conditions are currently badly served in Scotland. Results of surveys of our members in Scotland (575) support this and are provided.

Our organisation is over thirty years old and throughout this time, we have seen that without help, support, and access to treatment, EDS can have a significant impact on people's lives. Those affected face challenges to their physical and mental health, both of which can deteriorate rapidly without appropriate diagnosis, support, and treatment. EDS can force those with the conditions into poverty by its impact on their lives, especially if their ability to pay for the help and support they need is limited.

EDS UK works closely with the two specialist hypermobility services in England, UCLH and the Royal National Orthopedic Hospital in Stanmore, reinforcing and complementing the advice provided to patients by the medical teams there and providing long-term support to help people self-manage their conditions where possible. We would be very pleased to build the same relationship with a new centre in Scotland. Our volunteer-led support network is well-established in Scotland and is free and easy for patients to access.

Part of our strategic plan over the next five years is to work with the NHS nationwide to establish EDS UK centres of excellence. This will provide recognition to medical teams providing excellent care, with potential access to funding for both services and research.

We continue to build on the work we have already done to improve knowledge about EDS and related conditions in the medical community, for example our partnership