STAGE 1 Specialist Service: Outline 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 Provider & Location
NHS Lothian Western General Hospital Rheumatic Diseases Unit Edinburgh
1.4 Contacts and support from NHS Board Management
Professor Stuart H Ralston Centre for Genomic and Experimental Medicine University of Edinburgh Western General Hospital Edinburgh EH5 3LH
1.5 Brief description of proposed service ***(no more than 100 words)***
The aim of the service would be to provide a centre for the assessment, and management of patients with complex Ehler's Danlos Syndrome (EDS) whose needs are not being adequately met at a local level. The centre would offer a detailed evaluation of symptoms and needs of referred patients and develop an individualized care pathway for patients that would address the musculoskeletal and extra-skeletal manifestations of the disease. A shared care model would then be developed with specialists from the referring NHS board so that care could be delivered locally on a longer-term basis. As the musculoskeletal system is almost invariably affected it is anticipated that most referrals would come from consultant rheumatologists.
Section 2: Outline Proposal
2.1 Description of current provision and why there is a need for national commissioning?
The current provision for management of patients with complex EDS in Scotland is inadequate. Patient testimonials and surveys indicate that the diagnosis is often missed resulting in multiple hospital attendances to different specialists including rheumatologists, gastroenterologists, cardiologists and gynaecologists. Even once the correct diagnosis has been made holistic pathways of care for patients with complex EDS in Scotland are lacking and as such many patients are referred to specialist centres in England for residential care programmes. Almost all patients with this syndrome experience significant musculoskeletal pain and morbidity associated with hypermobility. A proportion of patients

also suffer from symptoms referable to the gastrointestinal tract, genitourinary tract and cardiovascular system. In addition to diagnostic delays, patients often experience a lack of a support structure to enable them to manage their condition once a diagnosis has been made.

Less than 10 patients per year are referred to the specialist diagnostic service commissioned by NHS England as well as various residential treatment programmes (Royal Hospital for Rheumatic Diseases Bath, or Royal Orthopaedic Hospital Stanmore) for advice on pain management. The actual number could be higher as some health boards may not seek funding from the Out of Area Risk Share and anecdotal evidence from patient organisations suggests that a significant number of patients fund access to residential programmes privately.

A survey of 384 patients conducted by the EDS society in Scotland revealed an average time of 19 years between presenting with symptoms and a diagnosis being reached and that only 16% had been able to see a healthcare professional that was aware of EDS. In addition, 87% of patients did not feel their needs were being adequately addressed by the NHS in Scotland. 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 take responsibility for managing their condition in a holistic way.

It would be impractical and costly to try and establish specialised services at a local health board level for this condition. Complex EDS is a rare disorder and patients' needs would be optimally served by a centralised model of assessment and developing a care plan, followed by shared care and local delivery in the longer term.

The care pathway would be led by a consultant rheumatologist with expertise in EDS. The reason for this is that a key defect at present is the need for a senior, medically qualified member of the care team to take overall responsibility for the management of the condition. The lead consultant will be responsible for initial evaluation of the patient; for deciding on the initial care plan and for deciding upon whether referral to other specialities such as clinical genetics, cardiology, gastroenterology, gynaecology or obstetrics is required. Other members of the care team would include a full time (100% FTE) specialist physiotherapist (PT), a full time (100% FTE) occupational therapist (OT), a part time (60% FTE) clinical psychologist (CP) and a part time (60% FTE) clinical nurse specialist (CNS). The OT and PT will play central roles in developing the package of care by providing advice on lifting, and handling, pacing, sleep hygiene, work/life balance (OT) as well as posture, physical activity, strengthening exercises, and stretching (PT). The CNS will work with the OT and PT in role in goal setting, managing fatigue, flare management, coping strategies and in setting up and dealing with enquiries from a patient help line / email advice service. Many EDS patients suffer anxiety and depression along with other psychological problems in coming to terms with their condition. The involvement of a CP is essential to successful management of this disorder and will be involved in helping patients to adjust to their condition; in the management of depression and anxiety; and in the management of pain.

2.2 Number of patients expected to benefit?

There is little information on the overall prevalence of EDS, with estimates ranging widely between 1 in 5000 to 1 in 100,000 of the population, but by far the most common is hypermobility type EDS (hEDS) which is estimated to account for 90% or more of patients with the disorder. During the past 4 years we have audited the number of referrals for diagnosis and management of EDS in the Rheumatic Diseases Unit in Edinburgh (Figure 1). Please note 2018 figures to August have been extrapolated to give projected numbers for the whole year. Based upon this we expect that up to 350 patients in Scotland may need specialist input that the centre could offer.

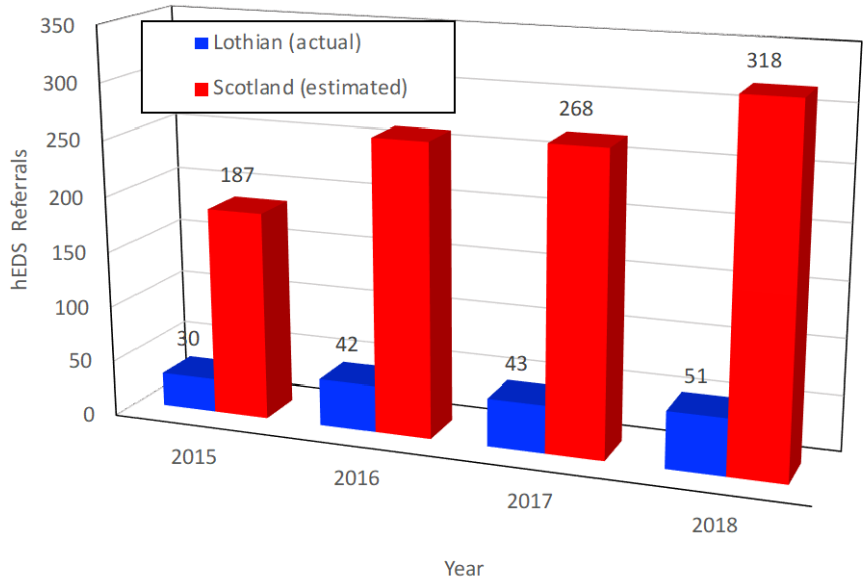

Figure 1: Number of referrals for diagnosis and management of EDS in the Rheumatic Diseases Unit - Edinburgh

2.3 Expected benefits/impact in relation to patient experience, quality, outcomes and wider NHS in Scotland?

It is expected that the proposed service would improve the experience and outcomes for people with severe forms of EDS by providing a comprehensive assessment of their clinical needs and a care plan for implementation at local level. Therefore, it is also envisaged that the service would support local services in managing patients' ongoing care including any complications and flare-up of symptoms.

Supporting individualised care pathways and shared care
Referrals into the service would initially undergo a detailed assessment by the lead consultant rheumatologist to evaluate the extent of their symptoms and to address the need for other specialist input. All patients would be evaluated by the physiotherapist, occupational therapist and specialist nurse. Input from the clinical psychologist will be sought in patients where this input is considered necessary by other members of the care team. The desired outcome of the assessment will be to develop and initiate a personalised care plan for patients referred into the service. It is anticipated that following the initial assessment patients would be reviewed on one or at most two occasions at the SCCEDS to monitor progress. Subsequently, patients would be discharged to the clinical team in the referring health board with a clearly defined care plan for long-term management which will be delivered locally.

Education
A key aim of the SCCEDS will be to develop an education programme on the diagnosis and management of EDS, primarily aimed at rheumatologists, general practitioners, physiotherapists, occupational therapists and specialist nurses across Scotland. All of these practitioners are capable of managing EDS but many feel that they do not have adequate training to treat the condition effectively. To achieve this aim, the SCCEDS would run regular courses on EDS and in combination with EDS support organisations, help to develop educational materials for patient use.

Research
The SCCEDS will establish a programme of clinically based research on EDS to try and gain greater understanding of the disease mechanisms and through this to develop more effective management strategies. An important aim will be to better understand the genetic contribution to hEDS.

2.4 What level of evidence is available to support the proposal?
• In line with most rare diseases the evidence base underpinning various treatment options is patchy. Many recommendations are based on expert opinion. Therapies are usually tailored to individual needs. Multidisciplinary input may be necessary. There are some diagnostic and treatment guidelines, including physiotherapy and psychological evaluation. • Results from patient questionnaires and patient testimonials.
2.5 Is there external support for the development of the service?
The proposal is strongly supported by the EDS Society and by the Scottish Society of Rheumatology