



Scottish Centre for Complex Ehlers Danlos Syndrome Stage 1 Proposal

1. Description of service

The aims 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.

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 inputs. All patients would be evaluated by the physiotherapist, and 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.

2. Provider & Location

NHS Lothian, Rheumatic Diseases Unit, Western General Hospital

3. Assessed Needs

Based on audits of referrals with a diagnosis and management plan for EDS to the Rheumatic Diseases Unit in Edinburgh over the past four years it is expected that up to 350 patients in Scotland may need specialist input that the centre could offer.

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 many such 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 the 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.

4. Level of evidence 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. There are some diagnostic and treatment guidelines, including physiotherapy and psychological evaluation. Results from patient questionnaires and patient testimonials

5. Level of external support for proposal

There is strong support from patient organisations and the Scottish Society for Rheumatology. Given the volume of ministerial correspondence and parliamentary questions on the topic it is expected that the Rare Disease Oversight Implementation Group would be supportive.

6. NSD Evaluation of proposal

Strengths of proposal

The proposal would address a number of service gaps for people with complex EDS. It would also provide clear guidance for referring clinicians about how patients should be managed and what continued care they should be offered locally. Given that the envisaged service would also provide education, it is hoped that the service would also equip local teams with the knowledge to manage people with non-complex EDS.

Weaknesses identified with proposal as stands

Referral criteria will have to be defined to ensure that only the most complicated cases are seen by the service. Implementation of care plans relies on availability of services locally (i.e. physiotherapy, psychology, occupational therapy).

7. NPPPRG Comments

NPPPRG supported a Stage 2 Application with the caveats 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.

8. Issues for Discussion

NSSC is asked to review the Stage 1 proposal and endorse the NPPPRG recommendation that the applicant be asked to prepare a Stage 2 application for the proposal.

**NSSC Secretariat
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